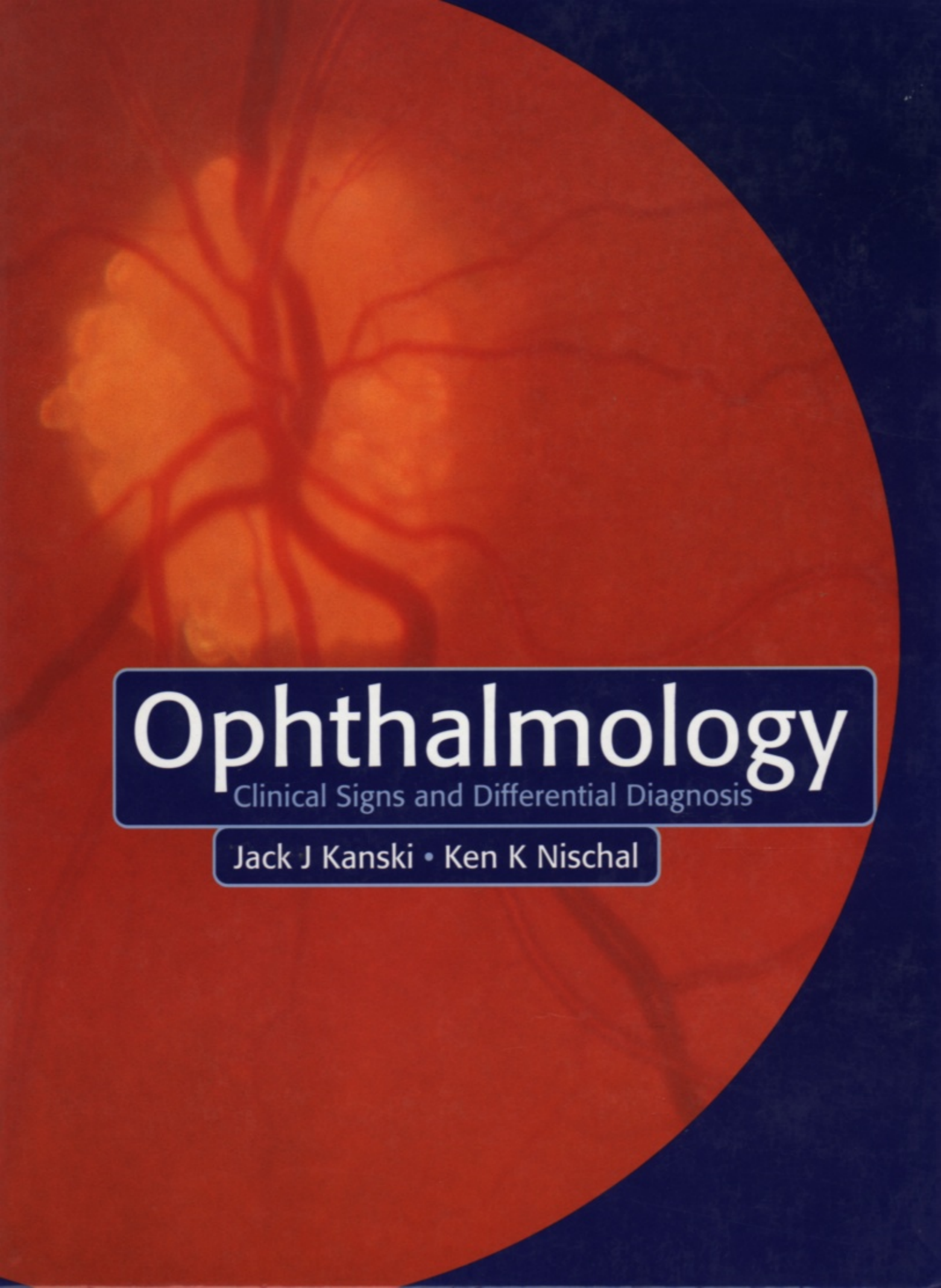

Ophthalmology

Clinical Signs and Differential Diagnosis

Jack J Kanski • Ken K Nischal

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Ophthalmology

Clinical Signs and Differential Diagnosis

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In memory of

Tony Shannan

and

Harbans Lal Nischal

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GUIDE TO ICONS


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Ocular associations


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PREFACE

There is an ever-increasing pressure in our specialist field of ophthalmology to know more and more detail about each particular sub-speciality such as the vitreo–retinal, pediatric ophthalmology, strabismus, and so on. However, all these different sub-specialities still have one very important common denominator: the eye and its component parts. All these component parts, such as the cornea, anterior chamber, iris, lens and so on, may exhibit different signs; it is the differential diagnosis of these signs that lies at the heart of our everyday clinical practice.

We have attempted to delineate these signs graphically and their differential diagnoses logically and practically, and structured the book so that each part of the eye is dealt with in a separate chapter. The book starts with the eyelids and progresses through to the fundus, with each sign clearly and succinctly explained and accompanied by color photos making this book invaluable as a desk reference for quick and easy diagnoses. We have also covered strabismus and nystagmus as separate disorders in great detail.

Furthermore, in recognition of the fact that ophthalmologists are expected to be more aware of the role of eye disease in systemic conditions and also of the genetic basis of disease as a whole, we have added an appendix which discusses the ophthalmic relevance of uncommon systemic disorders which have been mentioned in the text.

The text is concise yet comprehensive and pictorial icons are used to help the reader quickly and easily navigate through each section.

This book will be useful to the resident in training and primary care physicians as well as to practising ophthalmologists, orthoptists and optometrists.

Jack J Kanski
Ken K Nischal

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DISORDERS OF LASHES

Misdirection

Trichiasis

Trichiasis is a very common, acquired condition which may be unilateral or bilateral.

Signs

- Inturning of previously normal lashes on the lower (**Fig. 1.1**) or upper lid (**Fig. 1.2**). It should not be confused with inturning of lashes associated with entropion.

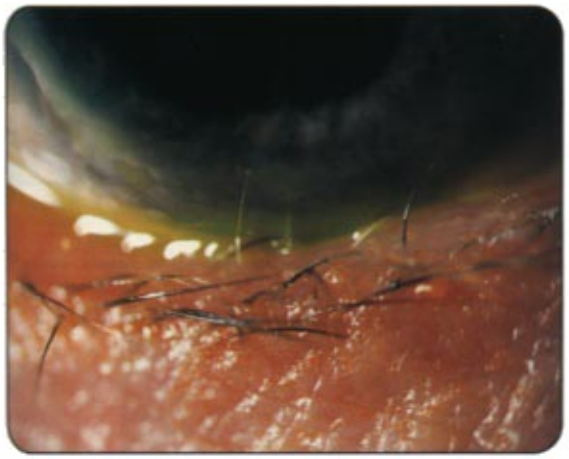


Fig. 1.1

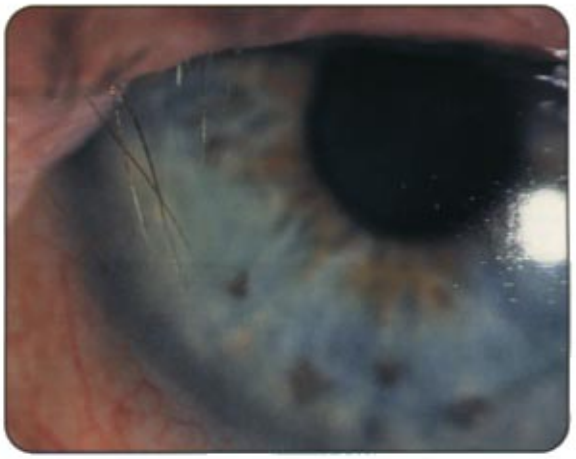


Fig. 1.2

Eyelash ptosis

Signs

- Downward drooping of upper-lid lashes (**Fig. 1.3**).

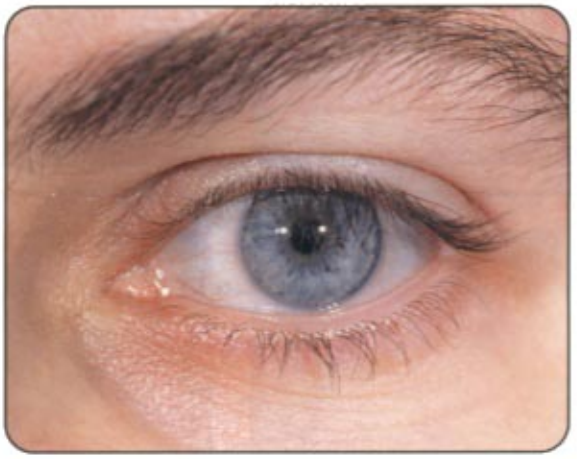


Fig. 1.3

Subcutaneous lash

Signs

- The outline of the lash is visible under the skin (**Fig. 1.4**).

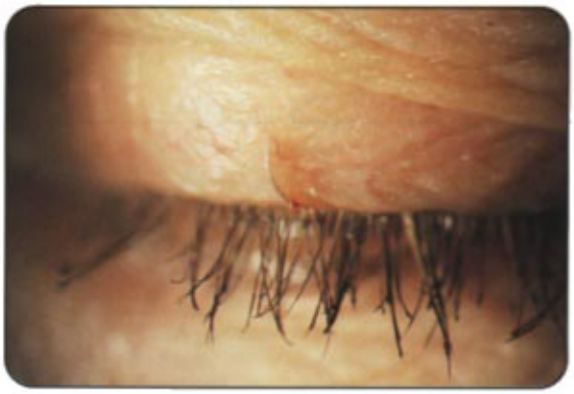


Fig. 1.4

Abnormal location

Congenital distichiasis

Congenital distichiasis is a rare, hereditary, bilateral condition which may be associated with lymphedema.

Signs

- Partial or complete second row of lashes originating from or slightly behind the meibomian gland orifices (**Fig. 1.5**). It should not be confused with metaplastic lashes, which is an acquired condition usually associated with cicatrizing conjunctivitis.

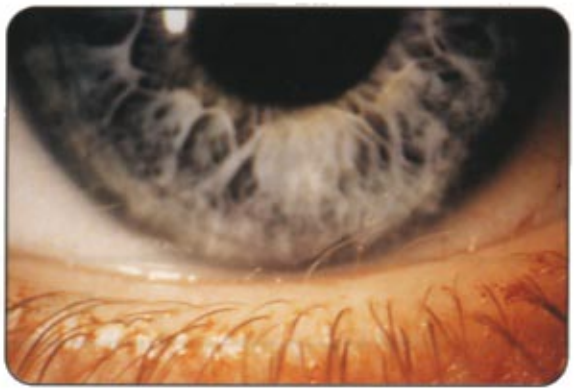


Fig. 1.5

Subconjunctival lash

Subconjunctival lash (**Fig. 1.6**) is a rare ocurrence which may be the source of redness and irritation.

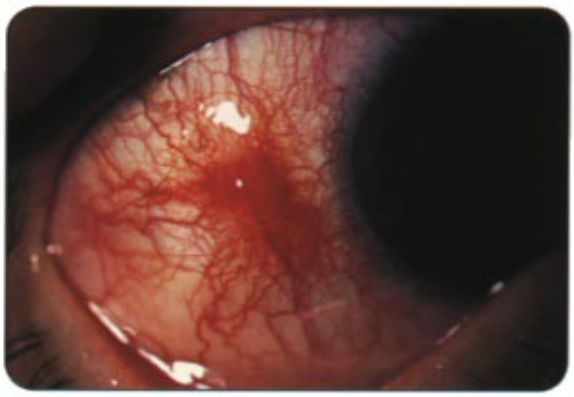


Fig. 1.6

Lash in punctum

Lash in punctum (**Fig. 1.7**) is an uncommon occurrence which may cause a pricking sensation on blinking.

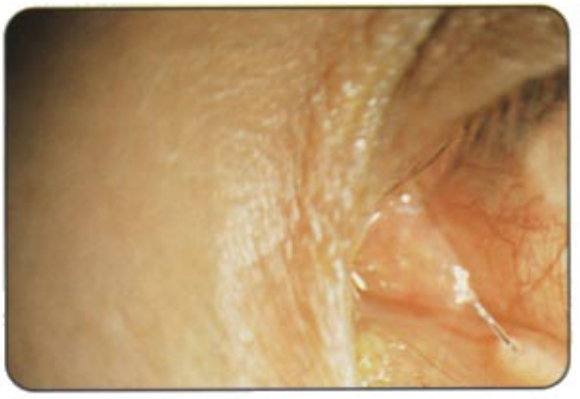


Fig. 1.7

Miscellaneous conditions

Madarosis

Signs

- Decrease in number or complete loss of lashes.

Table 1.1 Causes of madarosis

Local
• chronic anterior lid margin disease **(Fig. 1.8)**
• infiltrative lid tumors **(Fig. 1.9)**
• burns **(Fig. 1.10)**
Following removal
• iatrogenic for trichiasis **(Fig. 1.11)**
• trichotillomania – psychiatric disorder of habitual hair removal
Systemic
• myxedema
• generalized alopecia
• systemic lupus erythematosus
• psoriasis
• syphilis
• leprosy

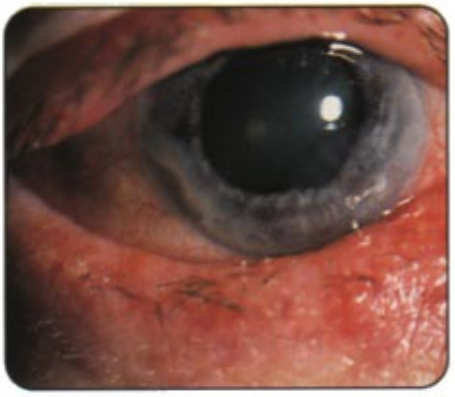


Fig. 1.8

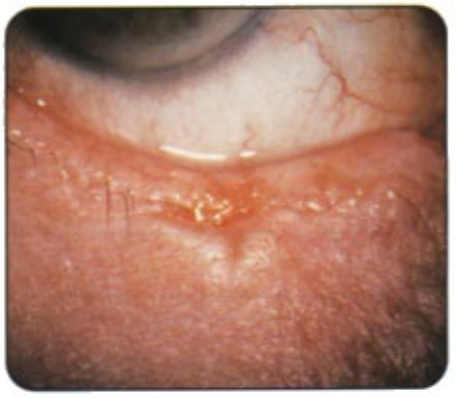


Fig. 1.9

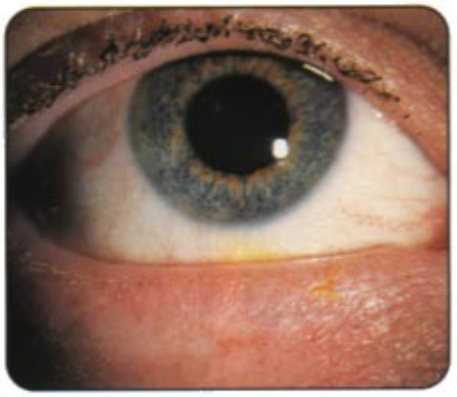


Fig. 1.10

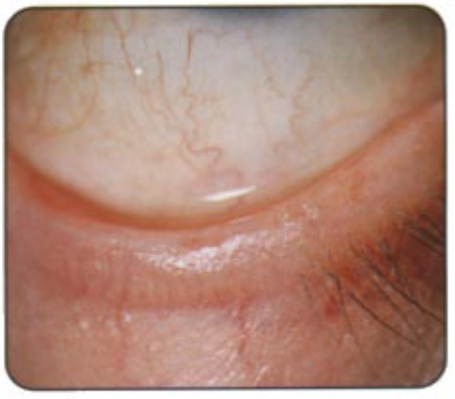


Fig. 1.11

Poliosis

Signs

- Whitening of lashes (**Fig. 1.12**).

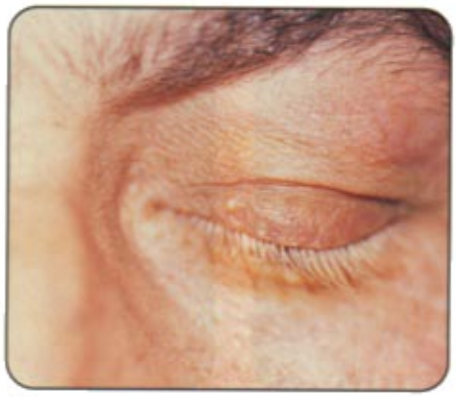


Fig. 1.12

Table 1.2 Causes of poliosis

Ocular
• chronic staphylococcal blepharitis
• sympathetic ophthalmitis
Systemic
• Vogt–Koyanagi–Harada syndrome
• Waardenburg syndrome

Phthiriasis palpebrarum

Phthiriasis palpebrarum is an uncommon infestation of lashes with crab lice (*Phthirus pubis*) and its ova (nits) which typically affects children.

Signs

- Nits stuck to the lashes (**Fig. 1.13**).

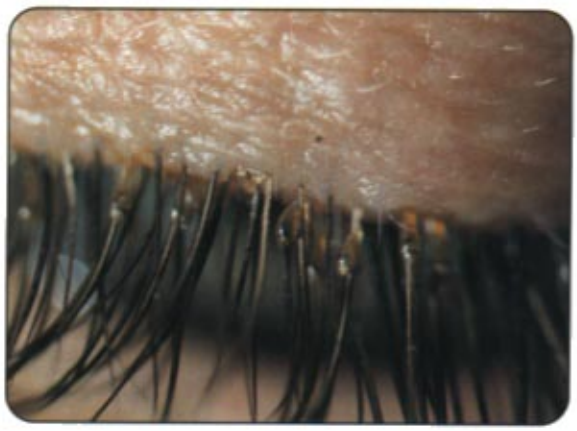


Fig. 1.13

Trichomegaly

Signs

- Excessively long and luxuriant lashes (**Fig. 1.14**).

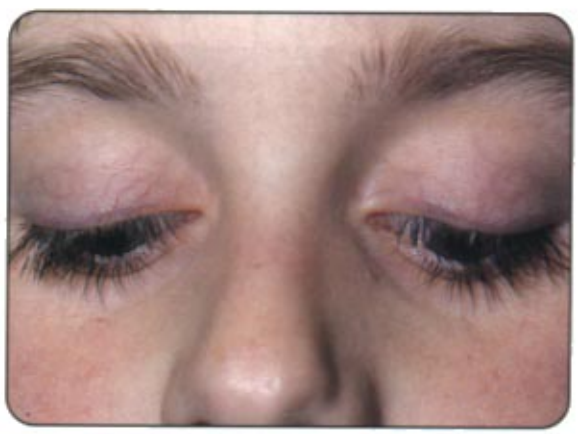


Fig. 1.14

SUPERFICIAL INFLAMMATIONS

Acute contact dermatitis

Acute contact dermatitis is a common, unilateral or bilateral condition, frequently caused by sensitivity to topical medication.

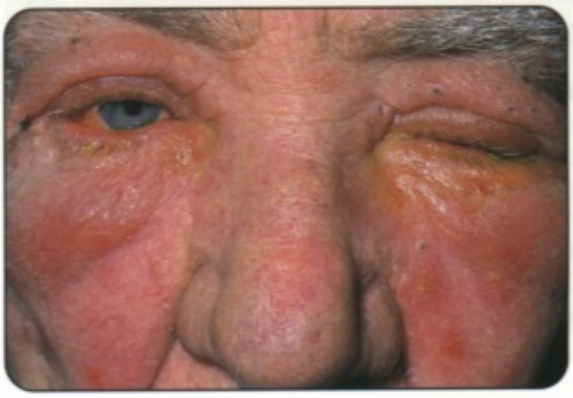


Fig. 1.15

Signs

- Erythema, edema, vesiculation, and crusting (**Fig. 1.15**).

Chronic contact dermatitis

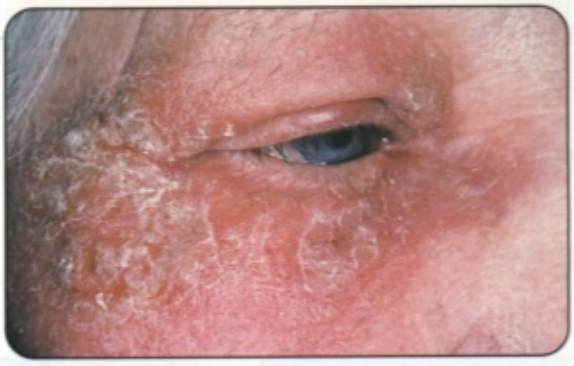


Fig. 1.16

Signs

- Thickening and crusting of skin (**Fig. 1.16**).

Atopic dermatitis

Atopic dermatitis of the lids is an uncommon, bilateral condition which may occur in patients with more generalized skin involvement.

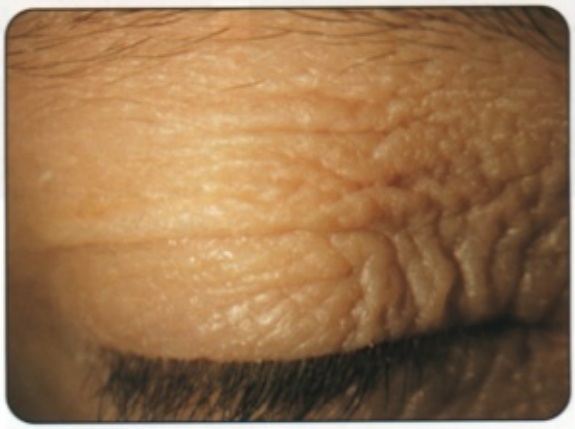


Fig. 1.17

Signs

- Thickened and fissured eyelids (**Fig. 1.17**).

Ocular associations

- Chronic staphylococcal blepharitis.
- Angular blepharitis.
- Vernal conjunctivitis in children.
- Atopic keratoconjunctivitis in adults.
- Keratoconus.
- Anterior shield-like cataract.
- Retinal detachment.

Primary herpes simplex

Primary herpes simplex of the lids is an uncommon, unilateral condition which may be severe in patients with associated atopic dermatitis or immune deficiency states.

Signs

- Crops of small vesicles (**Fig. 1.18**) which then rupture, crust, and heal without scarring after about 7 days.

Complications

- Acute follicular conjunctivitis and keratitis.

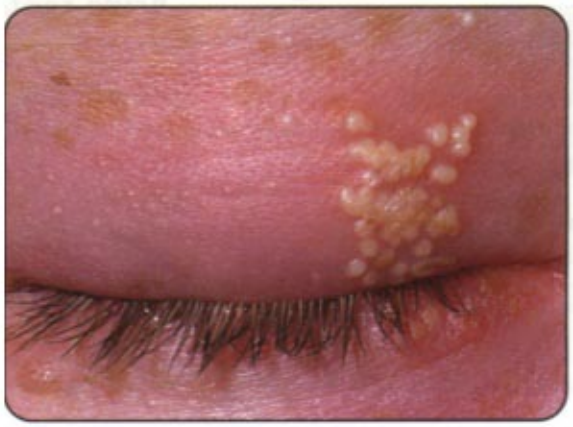


Fig. 1.18

Herpes zoster ophthalmicus

Herpes zoster ophthalmicus is a very common, unilateral condition which may be severe in patients with immune deficiency states.

Signs (in chronological order)

- Painful maculopapular rash involving the first division of the trigeminal nerve.
- Development of vesicles, pustules, and crusting ulceration (**Fig. 1.19**).
- Periorbital edema due to secondary bacterial cellulitis which in severe cases may spread to the opposite side, giving the erroneous impression that the condition is bilateral (**Fig. 1.20**).

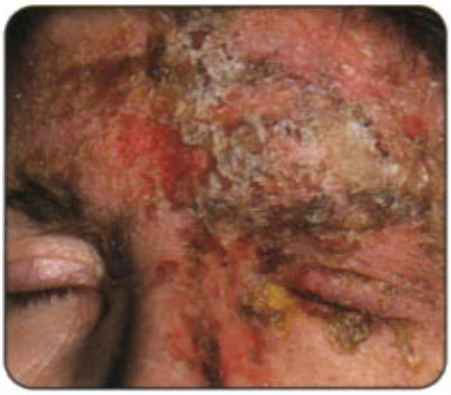


Fig. 1.19

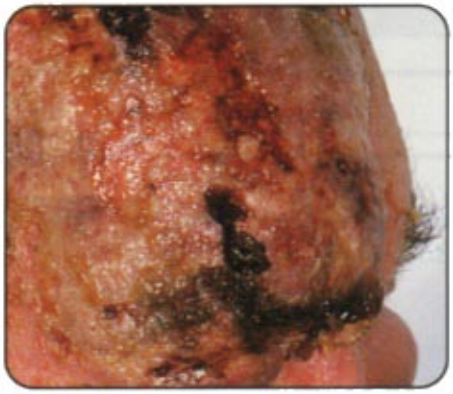


Fig. 1.20

Table 1.3 Complications of herpes zoster ophthalmicus

Anterior segment
• conjunctivitis
• episcleritis
• scleritis
• keratitis
• iritis
Other
• optic neuritis
• necrotizing retinitis
• extraocular nerve palsies

Angular blepharitis

Angular blepharitis is an uncommon, unilateral or bilateral infection caused by *Staphylococcus aureus* or *Moraxella lacunata*. It is frequently associated with atopic dermatitis.

Signs

- Erythema, fissuring, maceration, and scaling, localized to one or both canthi (**Fig. 1.21**).

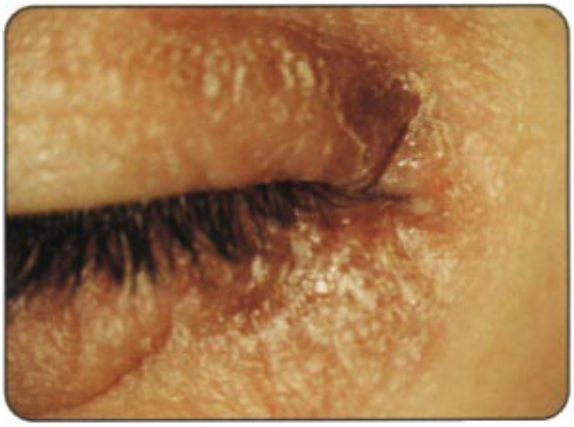


Fig. 1.21

Impetigo

Impetigo is an uncommon, bilateral condition caused by staphylococci or β-hemolytic streptococci which typically affects children.

Signs

- Initially small vesicles and bullae which on rupturing produce crusts composed of golden-yellow crystals (**Fig. 1.22**).

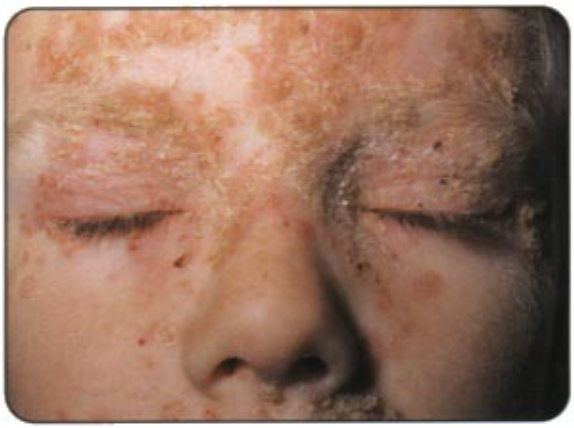


Fig. 1.22

Erysipelas

Erysipelas is an uncommon, subcutaneous cellulitis caused by entry of β-hemolytic streptococci through a site of minor skin trauma.

Signs

- Well-defined, erythematous, tender subcutaneous plaque, often with a butterfly configuration (**Fig. 1.23**).

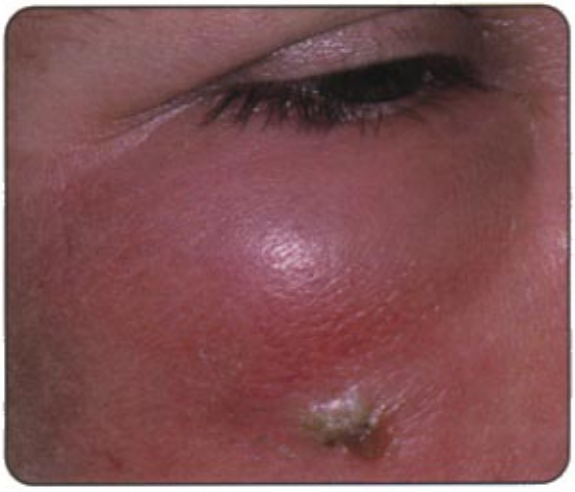


Fig. 1.23

Necrotizing fasciitis

Necrotizing fasciitis is an extremely rare but very serious condition comprising cutaneous gangrene, suppurative fasciitis, and vascular thrombosis, which usually involves the extremities and trunk.

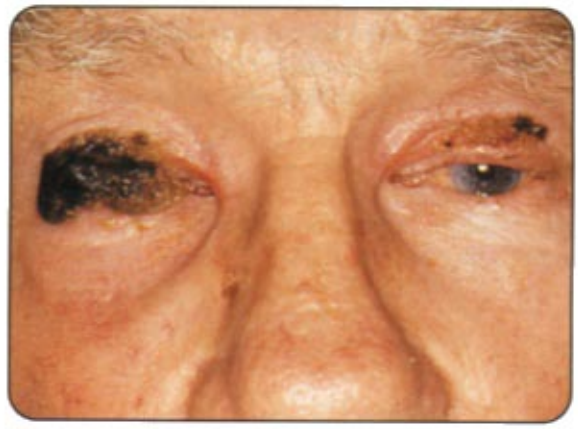


Fig. 1.24

Signs

- Bilateral eyelid necrosis (**Fig. 1.24**) is an occasional manifestation.

DIFFUSE SWELLING

Inflammatory

Associated with conjunctivitis

Adenoviral conjunctivitis in particular may be associated with severe generalized unilateral or bilateral lid edema (**Fig. 1.25**).

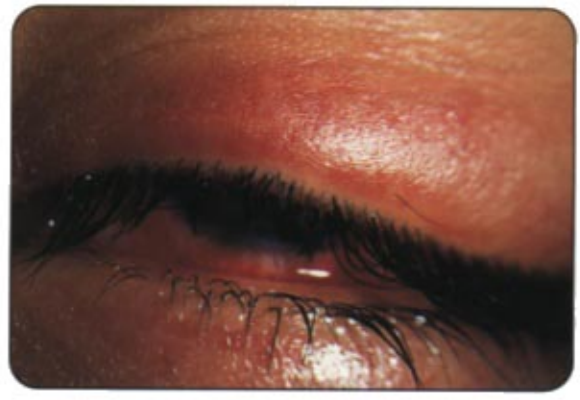


Fig. 1.25

Associated with dacryoadenitis

Signs

- Usually unilateral, tender erythema and edema involving the upper outer part of the lid, causing a characteristic S-shaped deformity (**Fig. 1.26**).

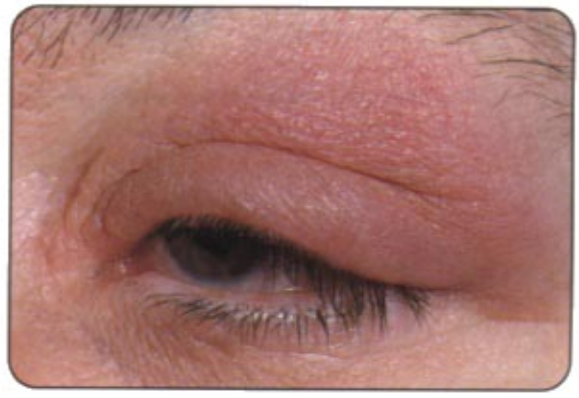


Fig. 1.26

Associated with dacryocystitis

Signs

- Very tender erythema and edema centered at the inner canthus, with variable spread to the lower and upper lids (**Fig. 1.27**)

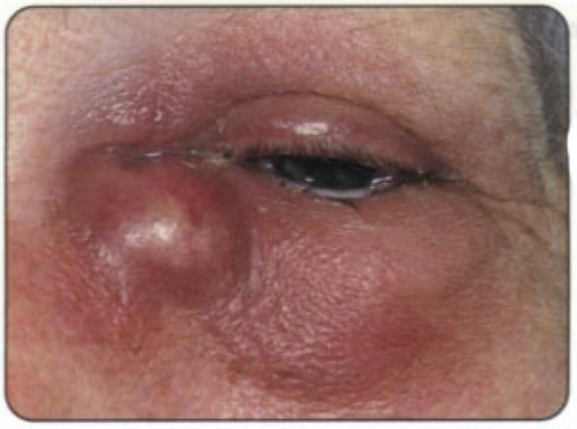


Fig. 1.27

Preseptal cellulitis

Signs

- Unilateral tender erythema and edema mainly involving the upper eyelid (**Fig. 1.28**).
- In contrast to orbital cellulitis there is no proptosis, and visual acuity, ocular motility, and pupillary reactions are all normal

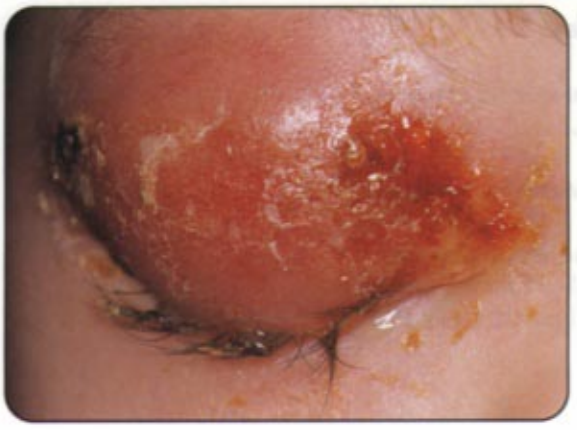


Fig. 1.28

Allergy

Signs

- Unilateral or bilateral painless edema (**Fig. 1.29**).

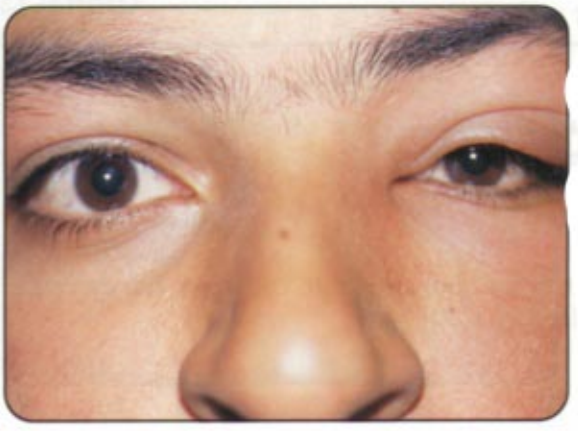


Fig. 1.29

Causes

- Insect bites.
- Angioneurotic edema.
- Urticaria.

Postsurgical

Signs

- Severe but painless lid edema (**Fig. 1.30**), usually associated with retinal detachment surgery or extensive retinal cryotherapy.

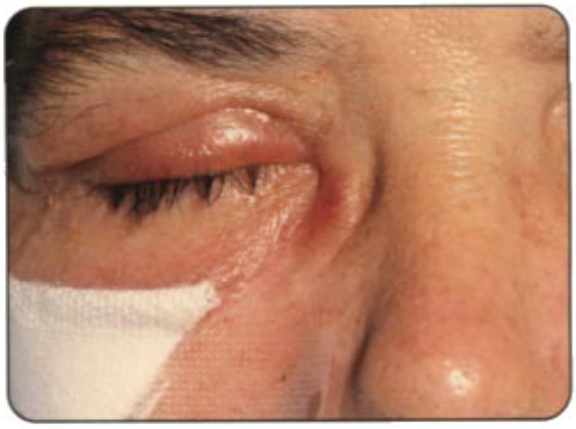


Fig. 1.30

Inflammatory orbital disease

Signs

- Painless edema associated with chemosis and proptosis (**Fig. 1.31**).

Causes

- Thyroid ophthalmopathy.
- Orbital cellulitis.
- Orbital pseudotumor.

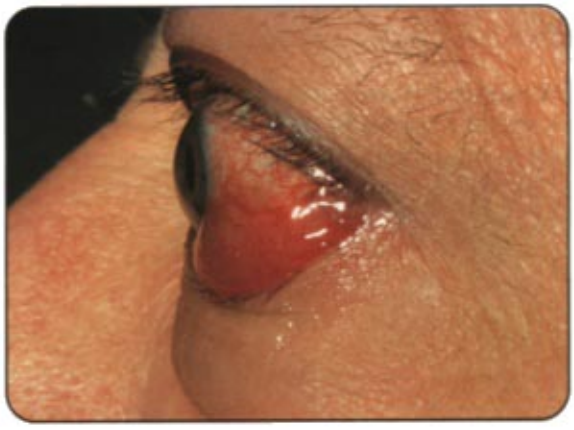


Fig. 1.31

Noninflammatory

Orbital fat herniation

Orbital fat herniation is a very common, usually bilateral, age-related condition.

Signs

- Pockets of fat herniating into the upper lid, especially medially (**Fig. 1.32**).

Fig. 1.32

Blunt trauma

Signs

- Painful edema and variable ecchymosis – 'panda bear' sign (**Fig. 1.33**).

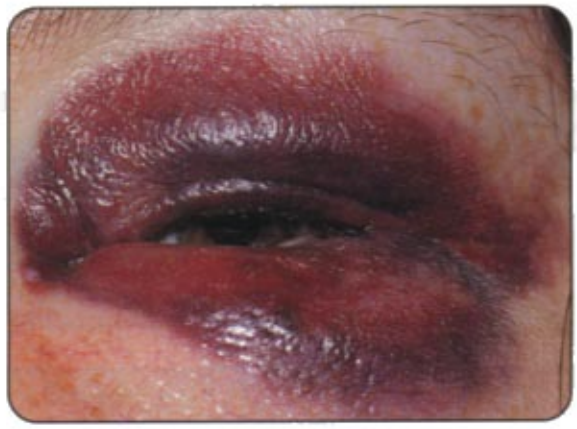


Fig. 1.33

Blepharochalasis

Blepharochalasis is an uncommon, usually bilateral condition which typically affects young individuals. It may subsequently give rise to an aponeurotic ptosis.

Signs

- Recurrent attacks of nonpitting edema of the upper (**Fig. 1.34**) – and occasionally also the lower – eyelid.

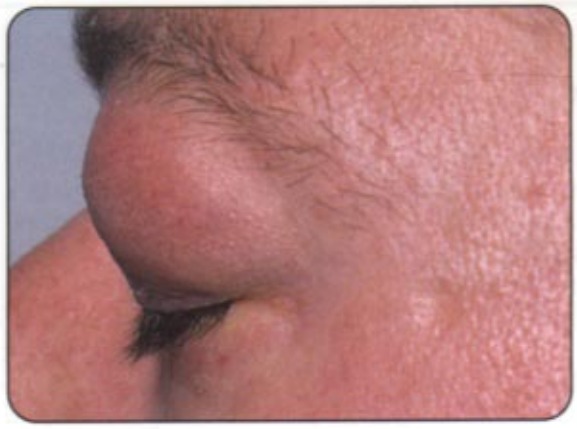


Fig. 1.34

Noninflammatory orbital disease

Causes

- Lymphomas – usually bilateral and non-Hodgkin.
- Rapidly growing tumors, especially rhabdomyosarcoma (**Fig. 1.35**) and metastatic carcinoma.
- Sphenoidal ridge meningioma may give rise to puffy eyelids due to chronic fluid stasis.
- Carotid–cavernous fistula.
- Subcutaneous emphysema associated with medial wall fracture.

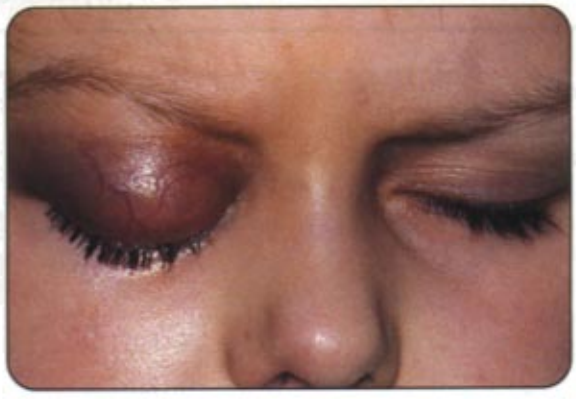


Fig. 1.35

Miscellaneous systemic disorders

All of the following conditions may be associated with bilateral periorbital swelling:
- Myxedema (**Fig. 1.36**).
- Renal disease.
- Congestive heart failure.
- Obstruction of the superior vena cava.
- Fabry disease.

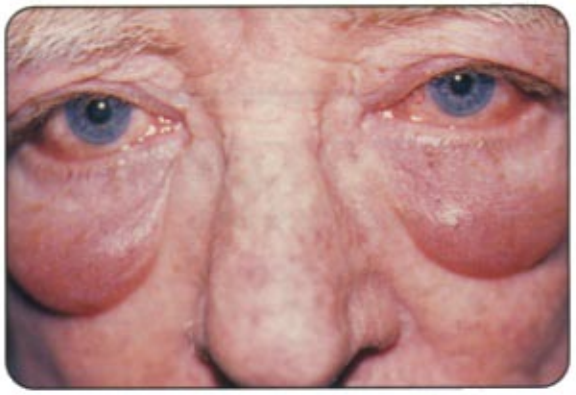


Fig. 1.36

CHRONIC MARGINAL BLEPHARITIS

Anterior blepharitis

Seborrheic blepharitis

Signs

- Shiny, waxy anterior lid margin, with mild–moderate erythema and oily lashes which may be stuck together (**Fig. 1.37**).
- The scales are soft, yellow, and greasy, and are located in between the lash roots.

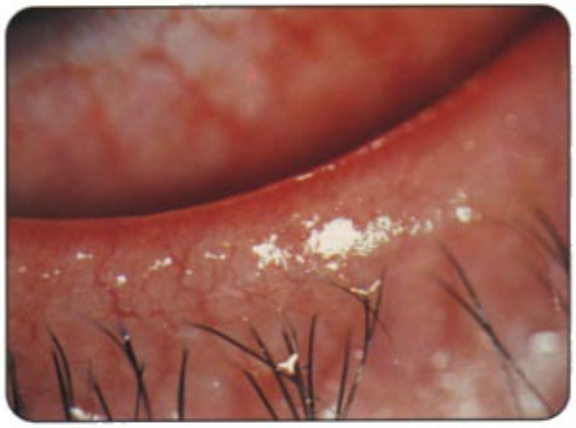


Fig. 1.37

Staphylococcal blepharitis

Signs

- Erythema and scarring of the anterior lid margin (**Fig. 1.38**).
- The scales are hard and brittle and are centered around the lash roots (collarettes) (**Fig. 1.39**).
- Misdirection of the lashes in long-standing cases (**Fig. 1.40**).

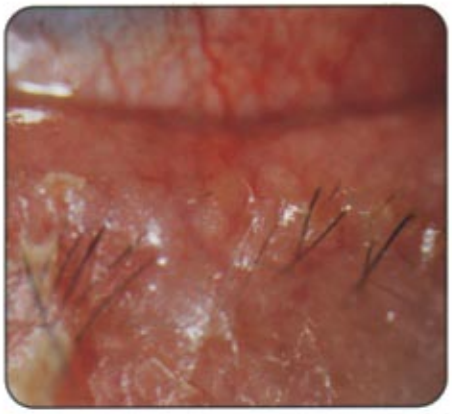


Fig. 1.38

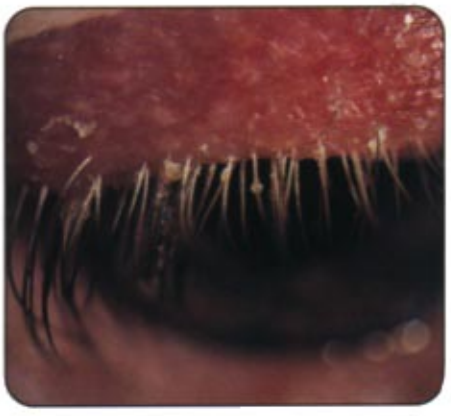


Fig. 1.39

Complications

- Acute external hordeolum (stye).
- Madarosis.
- Poliosis.
- Chronic papillary conjunctivitis.
- Punctate epitheliopathy.
- Marginal keratitis.
- Tear film instability.

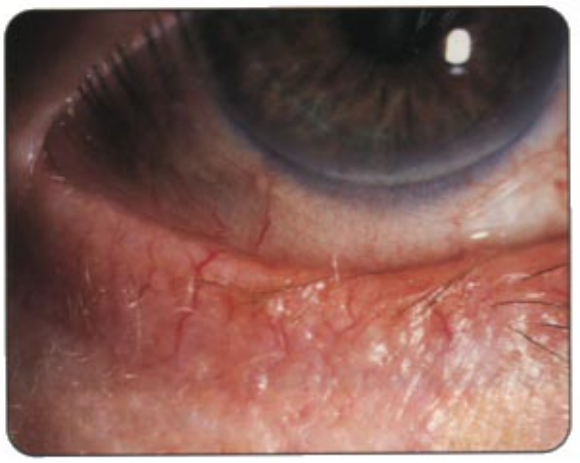


Fig. 1.40

Posterior blepharitis

Meibomian seborrhea

Signs

- Erythema of the posterior lid margin associated with an oily and foamy tear with accumulation of a frothy discharge on the lids and at the canthi (**Fig. 1.41**).
- Expressed meibomian gland secretions are profuse.

Look for

- Acne rosacea which is a common association.

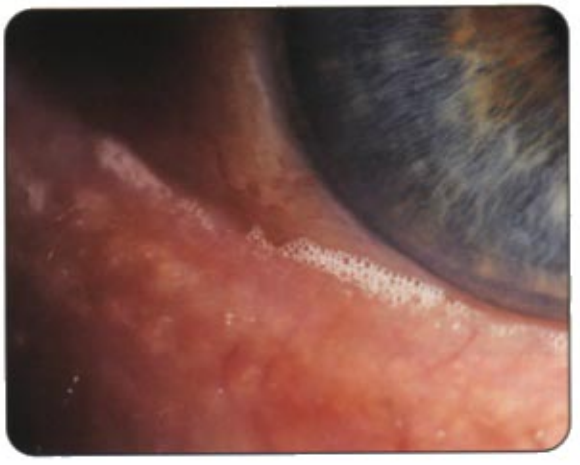


Fig. 1.41

Meibomitis

Signs

- Diffuse inflammation centered around the meibomian glands (**Fig. 1.42**).
- Expressed meibomian secretions may be turbid and have a toothpaste-like consistency (**Fig. 1.43**).
- In severe cases the meibomian gland orifices may be completely occluded (**Fig. 1.44**) and the posterior lid margin thickened, rounded, and notched.
- Meibomian cysts may also form (**Fig. 1.45**).

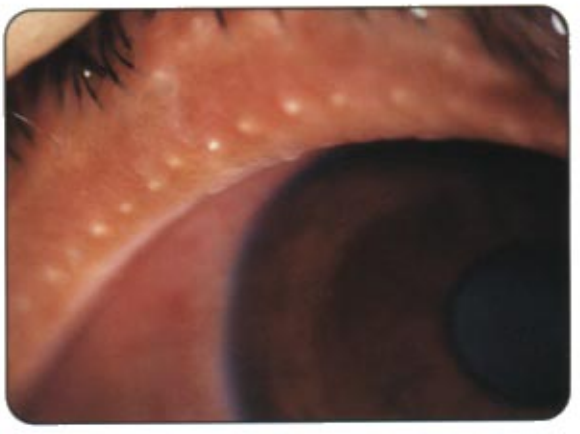


Fig. 1.42

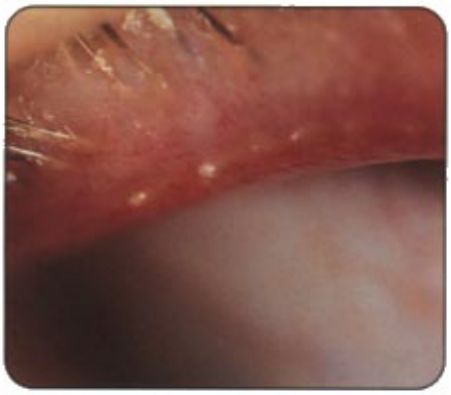


Fig. 1.43

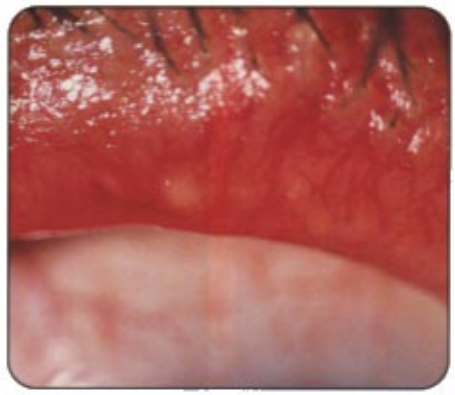


Fig. 1.44

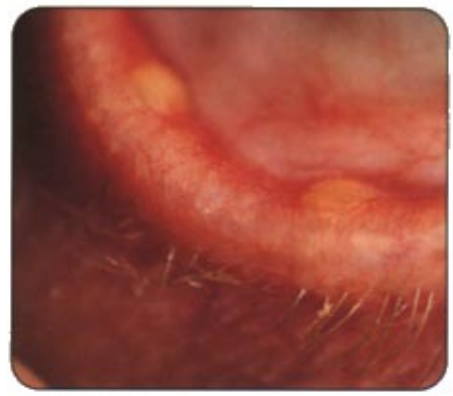


Fig. 1.45

NODULAR LESIONS

Benign

Xanthelasma

Xanthelasma is a common, frequently bilateral condition which is usually found in elderly individuals and those with hypercholesterolemia.

Signs

- Yellow subcutaneous plaque, typically occurring at the medial canthi (**Fig. 1.46**).

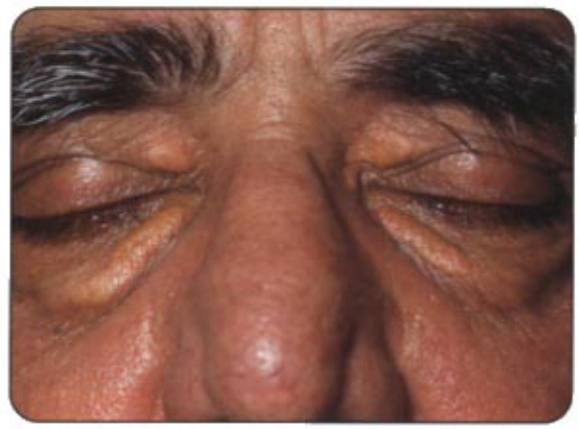


Fig. 1.46

Chalazion

Chalazion is a very common, usually unilateral, chronic lipogranulomatous inflammation of the meibomian glands or glands of Zeis.

Signs

- Painless, roundish, firm lesion within the tarsal plate (**Fig. 1.47**).
- If the cyst ruptures through the tarsal conjunctiva it may result in the formation of a polypoid mass (**Fig. 1.48**).

Look for, in recurrent cases

- Seborrheic dermatitis.
- Acne rosacea.
- Meibomian gland carcinoma.

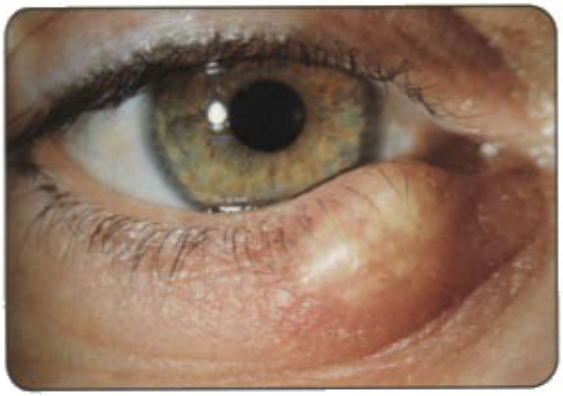


Fig. 1.47

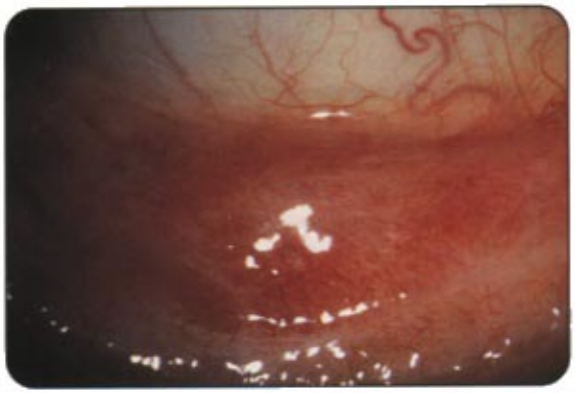


Fig. 1.48

Internal hordeolum (acute chalazion)

An internal hordeolum is a common, usually unilateral, staphylococcal infection of the meibomian glands.

Signs

- Acute, tender, diffuse, inflamed swelling within the tarsal plate (**Fig. 1.49**), resulting from a purulent infection of the meibomian glands.
- In severe cases it may be associated with preseptal cellulitis.

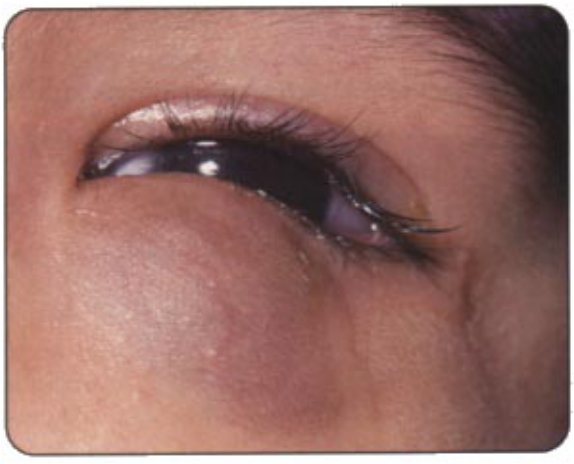


Fig. 1.49

External hordeolum (stye)

An external hordeolum is a very common, usually unilateral, acute purulent infection of a lash follicle and its associated gland of Zeis or Moll.

Signs

- Tender, localized, inflamed swelling in the lid margin, pointing anteriorly through the skin (**Fig. 1.50**).

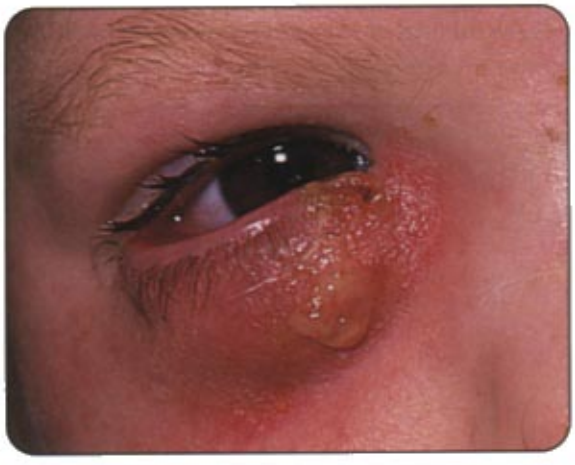


Fig. 1.50

Molluscum contagiosum

Molluscum contagiosum is an uncommon, unilateral or bilateral disease, with single or multiple lesions, caused by a poxvirus. In patients with immune deficiency states, the lesions may be more severe and even confluent.

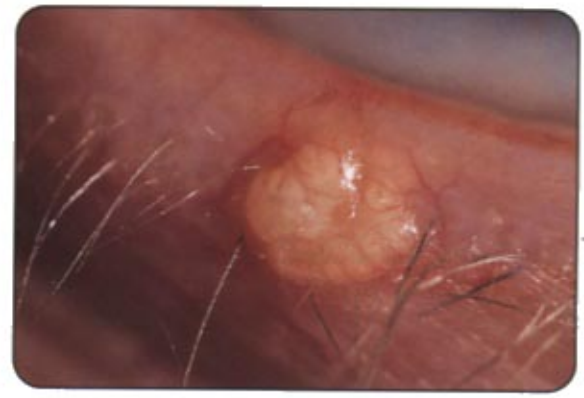


Fig. 1.51

Signs

- Small, pale, waxy, umbilicated nodule (**Fig. 1.51**).

Complications

- Chronic follicular conjunctivitis.
- Keratitis.

Malignant

Nodular basal cell carcinoma

Basal cell carcinoma is by far the most common malignant eyelid tumor. Fifty percent of tumors involve the lower lid, 30% the medial canthus, 15% the upper lid, and 5% the lateral canthus. Patients with xeroderma pigmentosum (**Fig. 1.52**) and Gorlin–Goltz (nevoid basal cell carcinoma) syndrome at at increased risk of developing basal cell carcinomas which may be multiple.

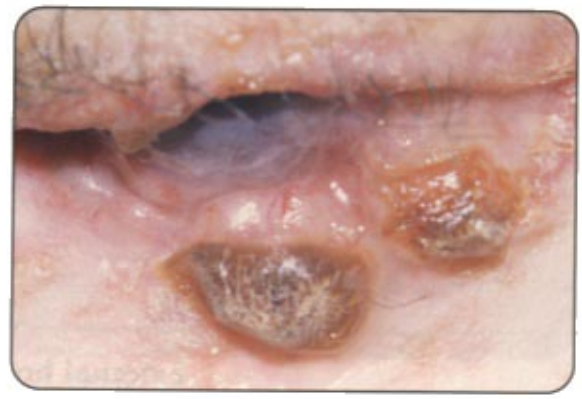


Fig. 1.52

Signs

- Slow-growing, firm, shiny, indurated nodule with fine surface telangiectasia (**Fig. 1.53**).
- Occasionally, associated hyperkeratosis may give the tumor a scaly appearance (**Figs 1.54, 1.55**).
- In some cases the tumor is pigmented.

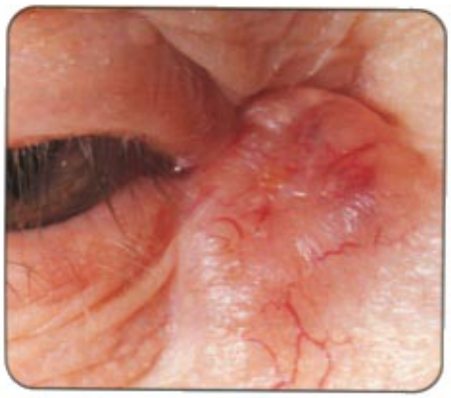


Fig. 1.53

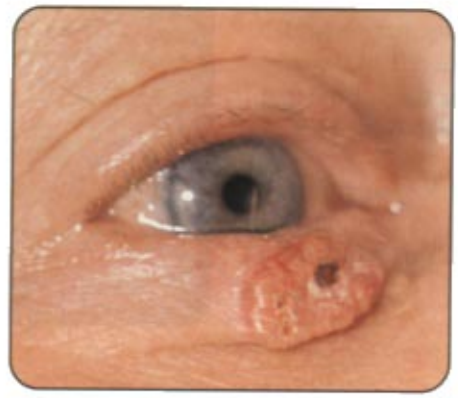


Fig. 1.54

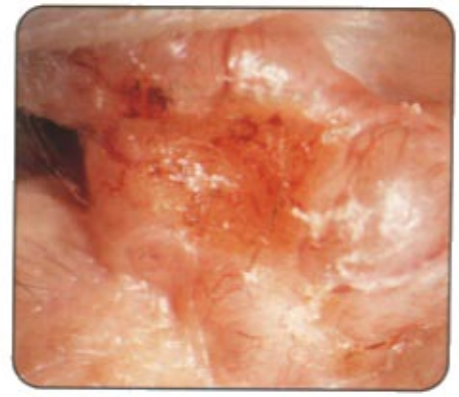


Fig. 1.55

Nodular squamous cell carcinoma

A nodular squamous cell carcinoma is much less common but faster growing than a basal cell carcinoma. It arises either *de novo* or from pre-existing actinic keratosis.

Signs

- Hyperkeratotic nodule (**Fig. 1.56**) or plaque that enlarges, and develops crusting erosions and fissures.
- In contrast to basal cell carcinoma it does not show surface telangiectasia.

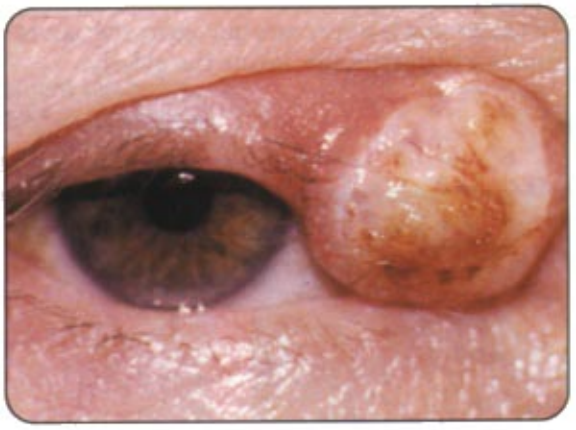


Fig. 1.56

Nodular meibomian gland carcinoma

A nodular meibomian gland carcinoma is very rare tumor which may be mistaken for recurrent chalazion.

Signs

- Hard swelling, most commonly within the upper tarsal plate (**Fig. 1.57**). A chalazion is more rubbery and localized.

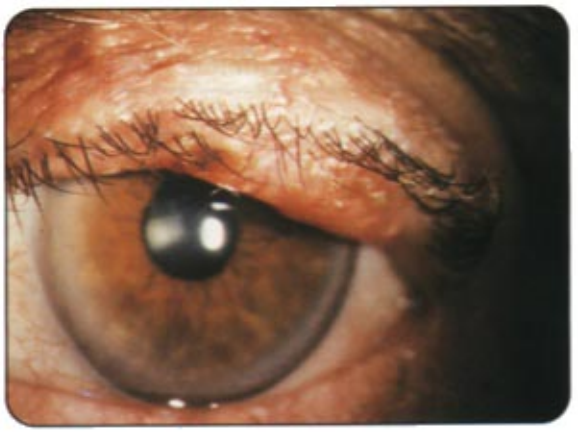


Fig. 1.57

Nodular gland-of-Zeis carcinoma

Signs

- Discrete, slow-growing, firm nodule on the lid margin (**Fig. 1.58**), which is associated with localized loss of lashes.

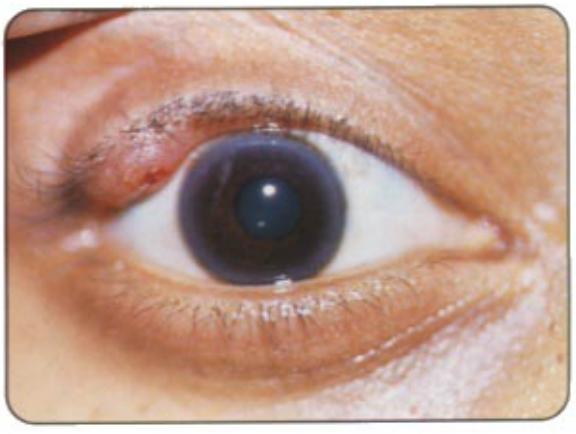


Fig. 1.58

Merkel cell carcinoma

A Merkel cell carcinoma is a very rare but aggressive neuroendocrine tumor which typically affects the elderly.

Signs

- Painless, fast-growing, red or purple nodule with overlying telangiectatic blood vessels (**Fig. 1.59**).

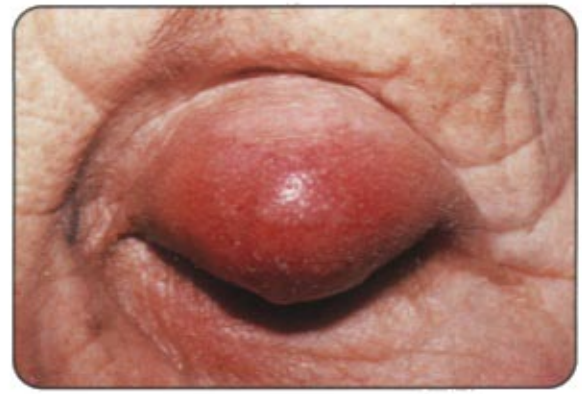


Fig. 1.59

ULCERATING TUMORS

Keratoacanthoma

A keratoancanthoma is an uncommon, fast-growing, benign tumor.

Signs

- It starts as a firm, pinkish, indurated nodule which quickly acquires a dome-shaped configuration with rolled edges and a keratin-filled crater (**Figs 1.60–1.62**).

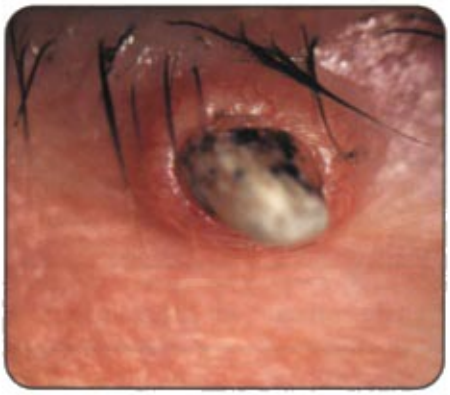


Fig. 1.60

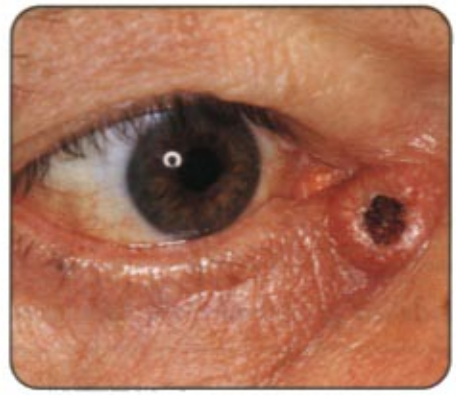


Fig. 1.61

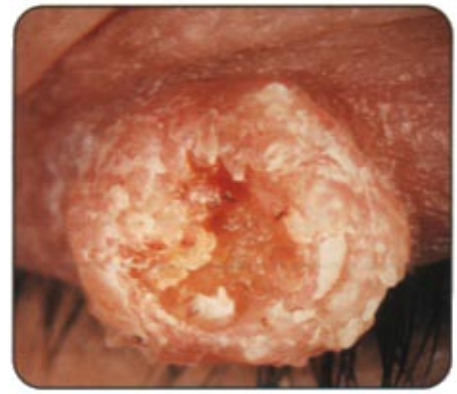


Fig. 1.62

Noduloulcerative basal cell carcinoma

Signs

- Initially it has central umbilication, followed by the development of the typical rodent ulcer with raised, rolled edges and an ulcerated center (**Figs 1.63–1.65**).

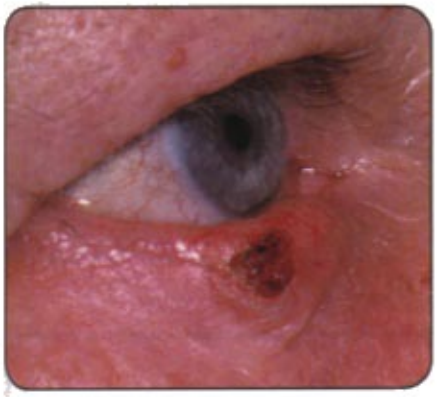


Fig. 1.63

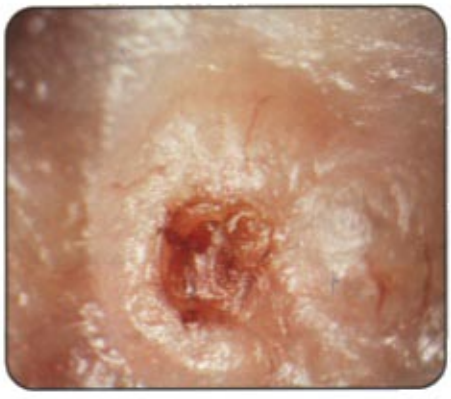


Fig. 1.64

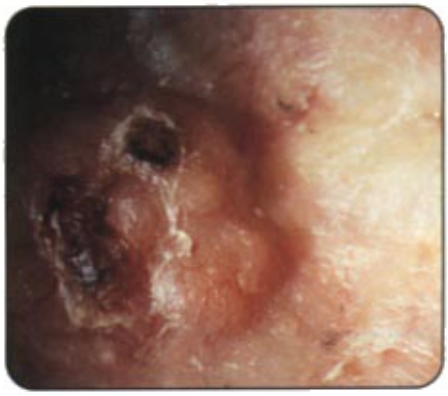


Fig. 1.65

Ulcerating squamous cell carcinoma

Signs

- The appearance (**Fig. 1.66**) is similar to that of basal cell carcinoma (BCC) but growth may be more rapid than with BCC.
- Advanced neglected tumors may invade the orbit (**Fig. 1.67**).

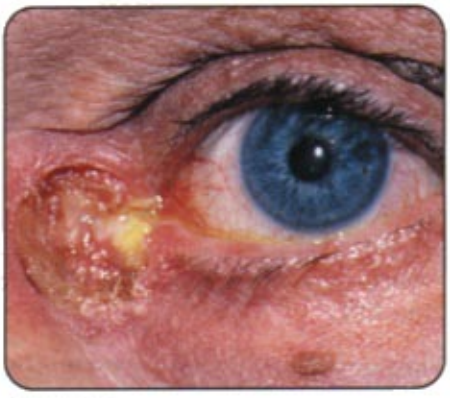


Fig. 1.66

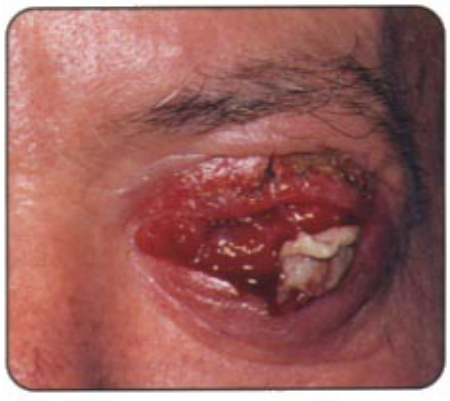


Fig. 1.67

Ulcerating gland-of-Zeis carcinoma

Signs

- Yellowish ulcerating nodule on the lid margin (**Fig. 1.68**)

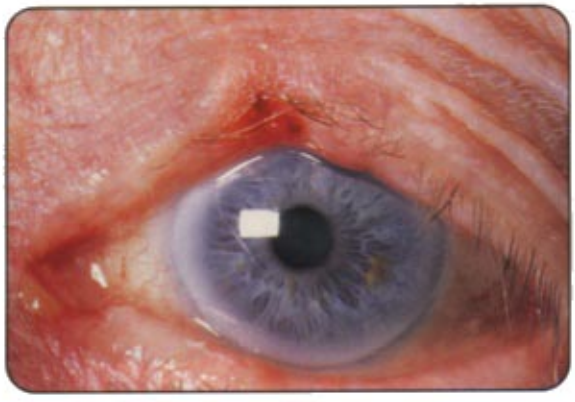


Fig. 1.68

INFILTRATING TUMORS

Sclerosing basal cell carcinoma

Signs

- Flat, indurated plaque with poorly demarcated margins and an intact epidermis, often accompanied by destruction of the overlying lashes (**Figs 1.69, 1.70**).

It should not be mistaken for unilateral chronic blepharitis.

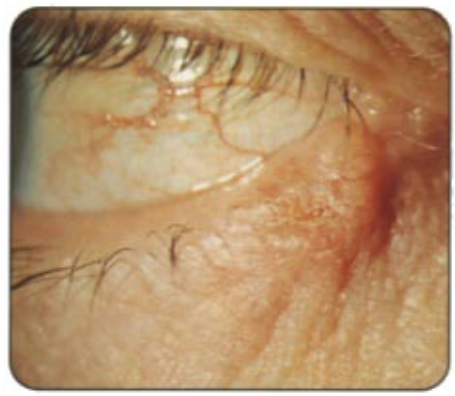


Fig. 1.69

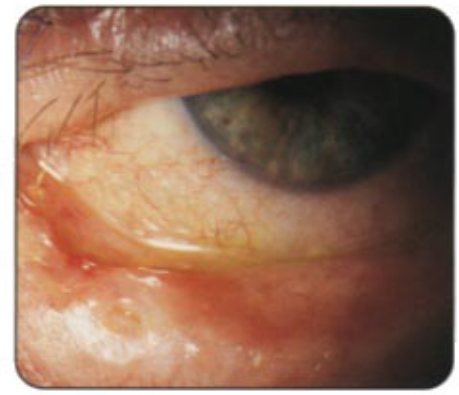


Fig. 1.70

Spreading meibomian gland carcinoma

Signs

- The appearance (**Fig. 1.71**) is similar to that of sclerosing basal cell carcinoma.
- The tumor may also spread within the epithelium of the conjunctiva (pagetoid spread) and mimic nonspecific chronic conjunctivitis (**Fig. 1.72**).

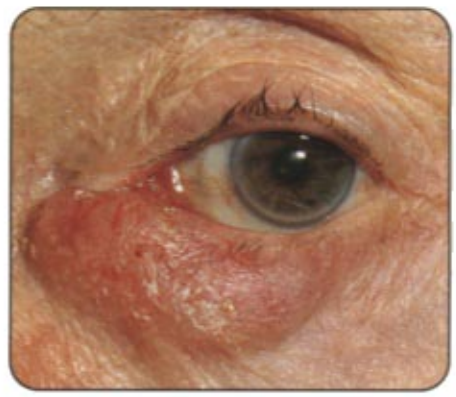


Fig. 1.71

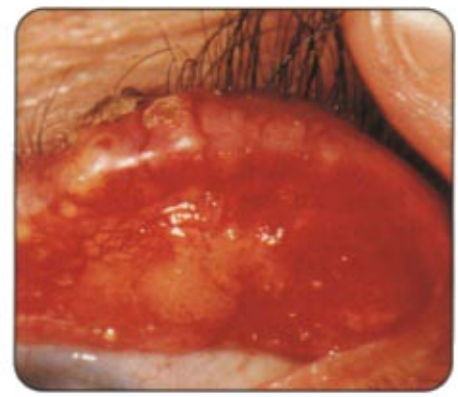


Fig. 1.72

WART-LIKE LESIONS

Viral wart (squamous cell papilloma)

Viral wart is the most common benign tumor of the eyelid.

Signs

- Pedunculated or sessile lesion with a characteristic irregular raspberry-like surface (**Figs 1.73, 1.74**).

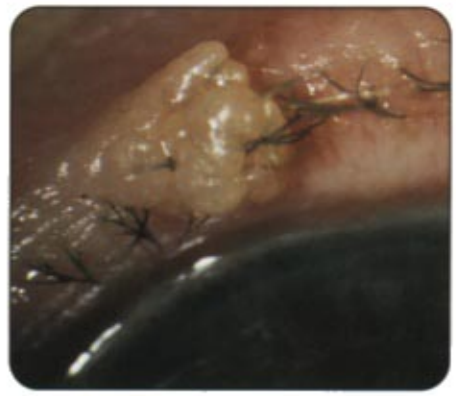


Fig. 1.73

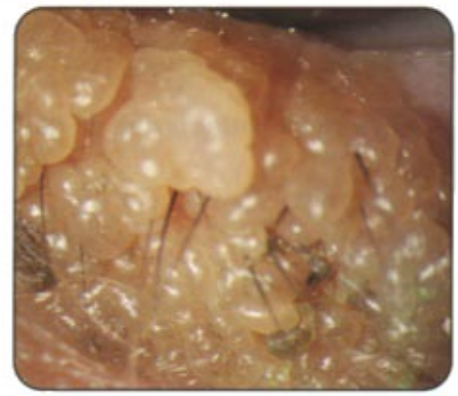


Fig. 1.74

Seborrheic keratosis (basal cell papilloma)

Seborrheic keratosis is a common condition which occurs in middle-aged and elderly patients.

Signs

- Discrete, greasy, brown, roundish lesion with friable verrucous surface and a 'stuck-on' appearance (**Fig. 1.75**).
- Occasionally it has a papillomatous configuration (**Fig. 1.76**).

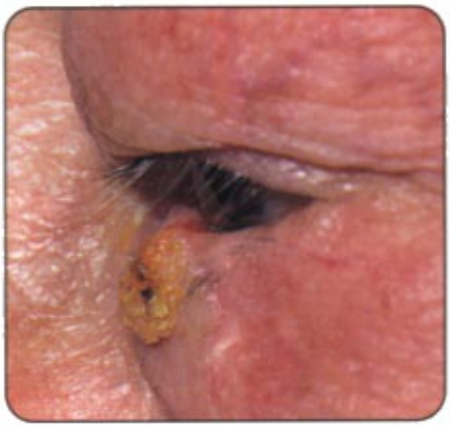


Fig. 1.75

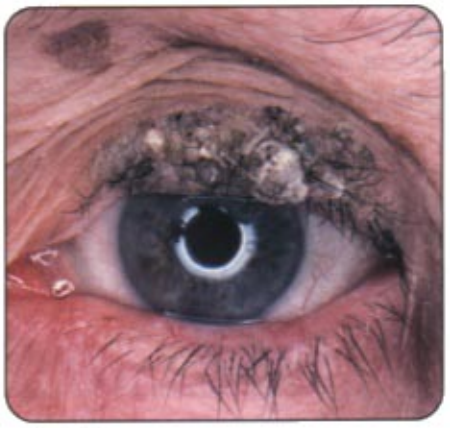


Fig. 1.76

Actinic keratosis

Actinic keratosis of the eyelids is an uncommon condition which predisposes to squamous cell carcinoma.

Signs

- Rough, dry, scaly lesion with an erythematous base (**Fig. 1.77**).

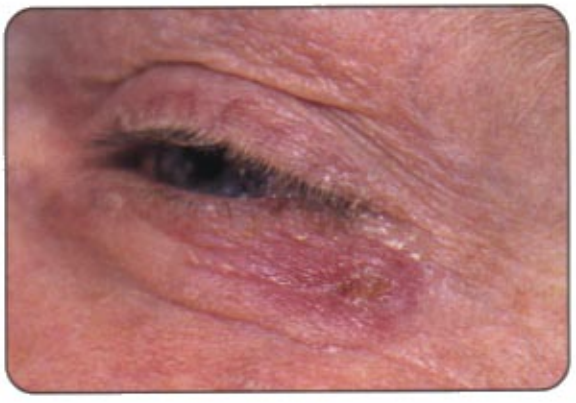


Fig. 1.77

Inverted follicular keratosis

Inverted follicular keratosis is an uncommon condition which histologically is a type of basal cell papilloma but has a different clinical appearance.

Signs

- Small, fast-growing, wart-like (**Fig. 1.78**) or nodular lesion.

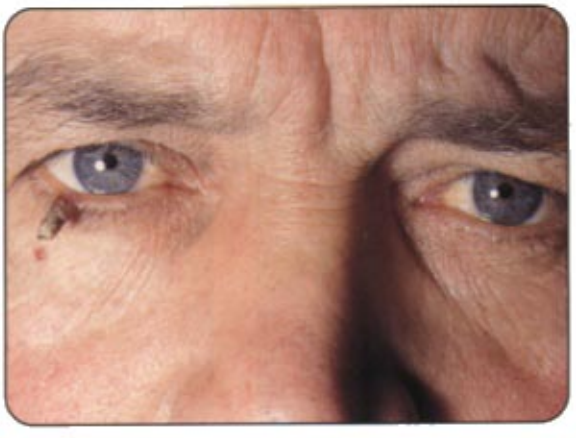


Fig. 1.78

Cutaneous horn

Cutaneous horn is a rare condition which may overlie an area of dysplastic epidermis (actinic keratosis) or malignant epidermis (squamous cell carcinoma).

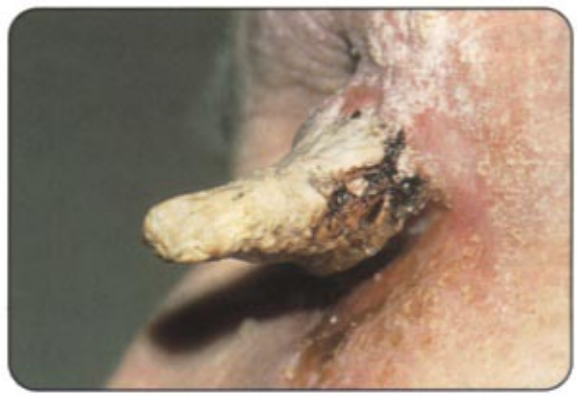


Fig. 1.79

Signs

- Hyperkeratotic, horn-like lesion protruding from the skin surface (**Fig. 1.79**).

CYSTIC LESIONS

Apocrine sweat gland hidrocystoma (cyst of Moll)

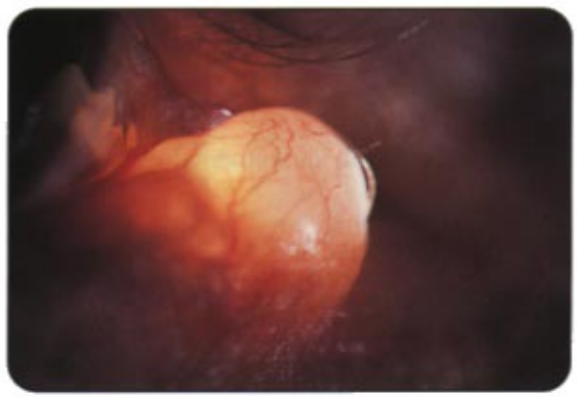


Fig. 1.80

Signs

- Very common, chronic, painless, translucent cystic nodule on the lid margin, containing serous secretions (**Fig. 1.80**).

Eccrine sweat gland hidrocystoma

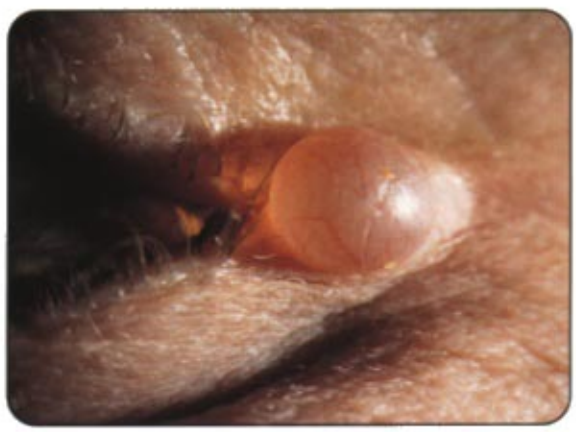


Fig. 1.81

Signs

- Less common but similar to a cyst of Moll except that it is not confined to the lid margin (**Fig. 1.81**).

Cyst of Zeis

Signs

- Very common, chronic, painless, opaque, smooth cystic nodule on the lid margin, containing oily secretions (**Fig. 1.82**).

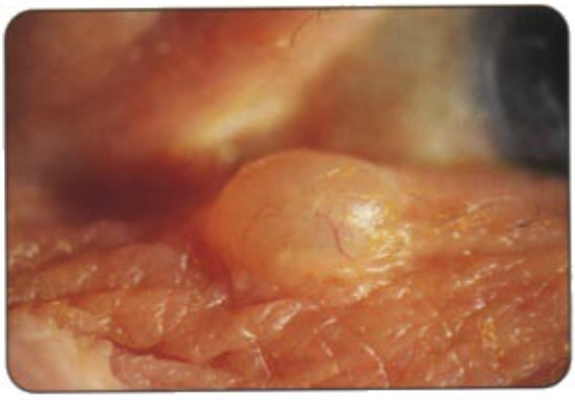


Fig. 1.82

Milia

Signs

- Small, round, superficial cysts which tend to occur in crops and may be bilateral (**Fig. 1.83**).

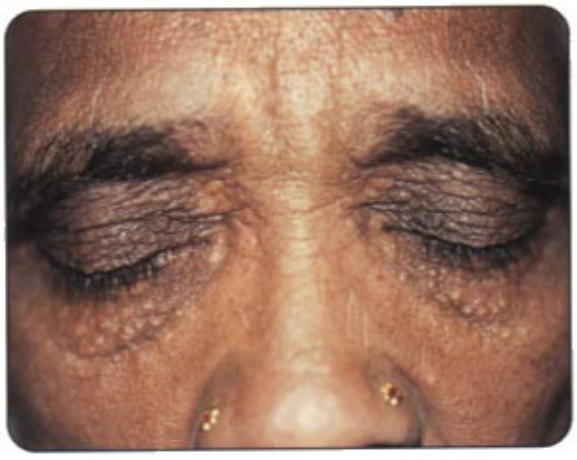


Fig. 1.83

Sebaceous cyst

Signs

- Less common but similar to a cyst of Zeis except that it usually has a central waxy punctum and it most frequently occurs at the inner canthus (**Fig. 1.84**).
- Bilateral and multiple (**Fig. 1.85**) cysts are common.

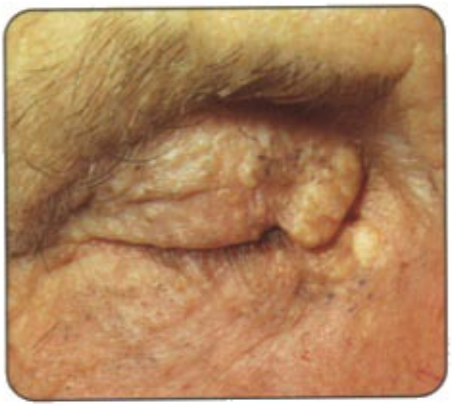


Fig. 1.84

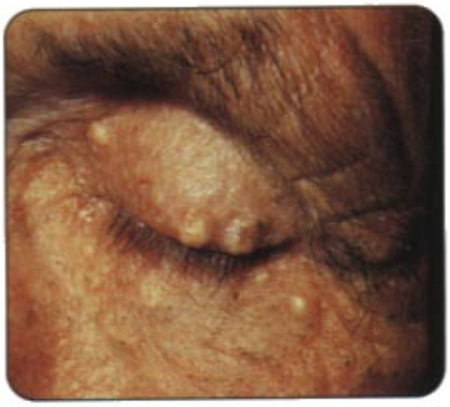


Fig. 1.85

Epidermal inclusion cyst

Epidermal inclusion cyst is an uncommon condition which is usually due to trauma or surgery but may occasionally be congenital. Multiple cysts may be found in patients with Torre syndrome and Gardner syndrome.

Signs

- Slowly progressive, firm subepithelial nodule which is usually solitary and most commonly found on the upper eyelid (**Fig. 1.86**).

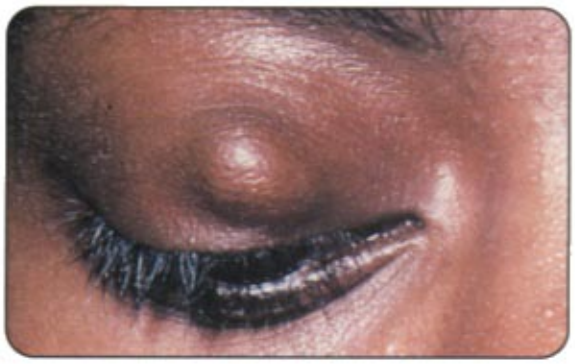


Fig. 1.86

Superficial dermoid cyst

Superficial dermoid cyst is an uncommon condition which presents in infancy.

Signs

- Smooth subcutaneous mass, most frequently located below the lateral brow (**Fig. 1.87**).

Fig. 1.87

PIGMENTED LESIONS

Benign

Oculodermal melanocytosis (nevus of Ota)

Oculodermal melanocytosis is a rare, congenital condition which is associated with an increased risk of uveal melanoma.

Signs

- Gray–blue discoloration of the skin effecting the distribution of the trigeminal nerve (**Fig. 1.88**).

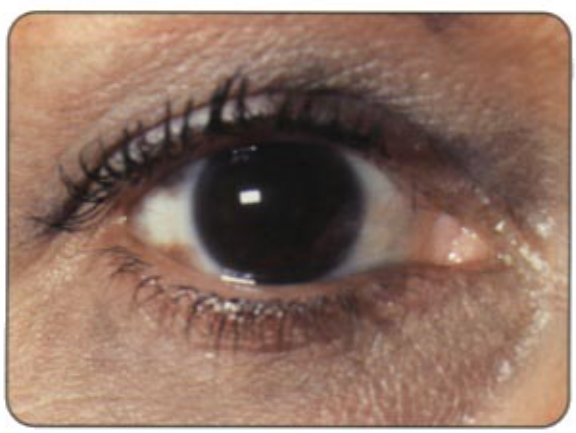


Fig. 1.88

Ocular associations

- Conjunctival subepithelial melanosis.
- Hyperpigmentation of the iris.
- Iris mammillations (uncommon).
- Trabecular hyperpigmentation.
- Glaucoma (uncommon).

Divided nevus

Signs

- Congenital melanocytic nevus which involves both the upper and lower lids (**Fig. 1.89**).

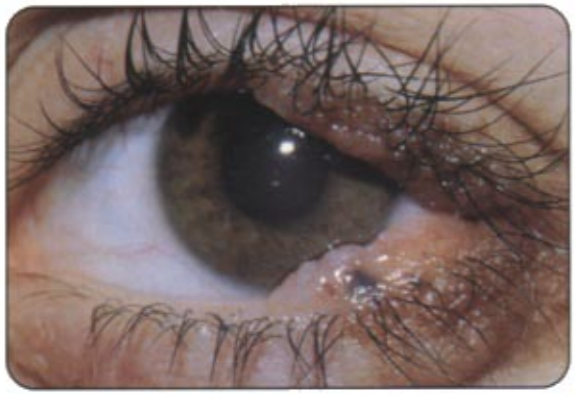


Fig. 1.89

Acquired nevi

The three main types of aquired nevi are junctional, compound, and dermal.

Signs

- Flat or elevated lesions with variable, tan to brown pigmentation (**Fig. 1.90**).
- When located on the lid margin, lashes may protrude through the lesion (**Fig. 1.91**).

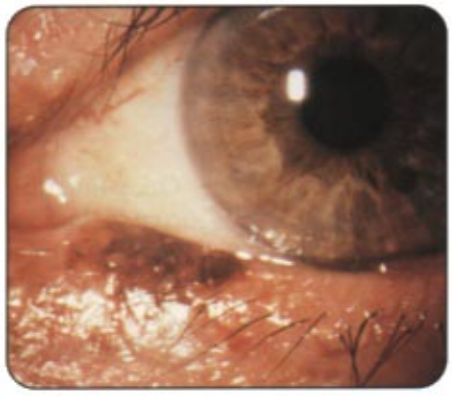


Fig. 1.90

Fig. 1.91

Malignant

Lentigo maligna (Hutchinson freckle, precancerous melanosis of Dubreuilh)

Lentigo maligna is an uncommon lesion which typically affects elderly patients. It is the preinvasive stage of melanoma.

Signs

- Slowly expanding pigmented macule (**Fig. 1.92**).
- The presence of nodular thickening is indicative of malignant transformation into a frank melanoma.

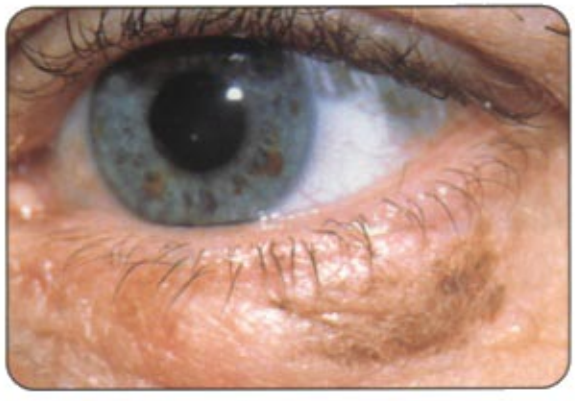


Fig. 1.92

Nodular melanoma

Signs

- Nodule with irregular pigmentation.
- It may may grow rapidly and be associated with breakdown of the overlying epidermis (**Fig. 1.93**).

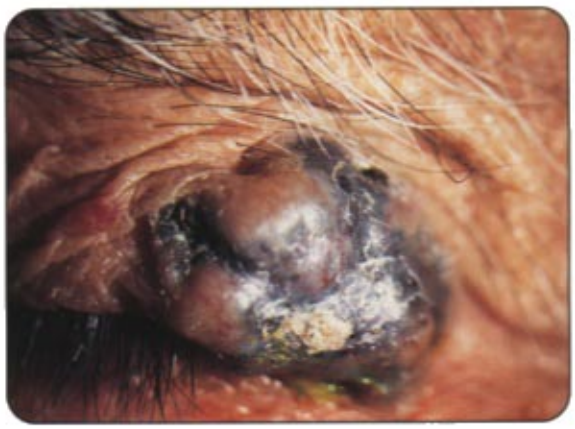


Fig. 1.93

VASCULAR LESIONS

Port-wine stain (nevus flammeus)

Port-wine stain is an uncommon, congenital, bilateral or unilateral condition.

Signs

- Sharply demarcated patch which darkens with age from red to purple (**Fig. 1.94**).
- The lesion does not blanch on pressure.
- Occasionally, the involved skin may be swollen and coarse (**Fig. 1.95**).
- Small lesions occur in isolation (**Fig. 1.96**), whereas larger lesions may have systemic implications.

Systemic associations

- Sturge–Weber syndrome.
- Klippel–Trenaunay–Weber syndrome.

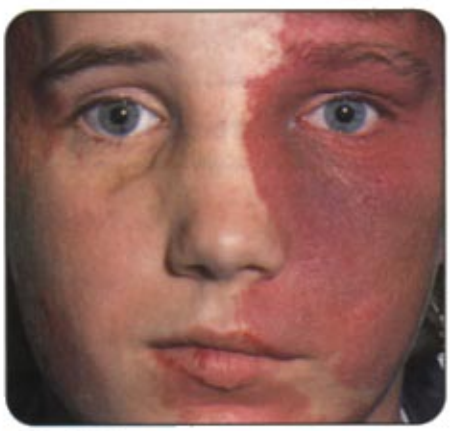


Fig. 1.94

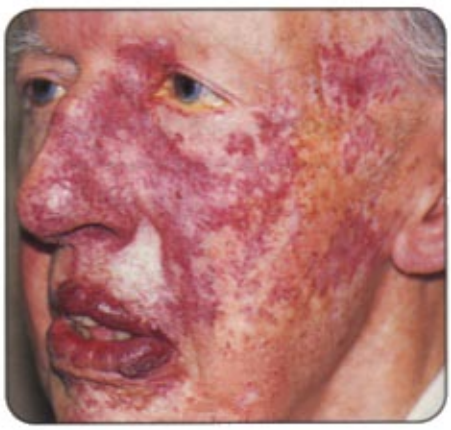


Fig. 1.95

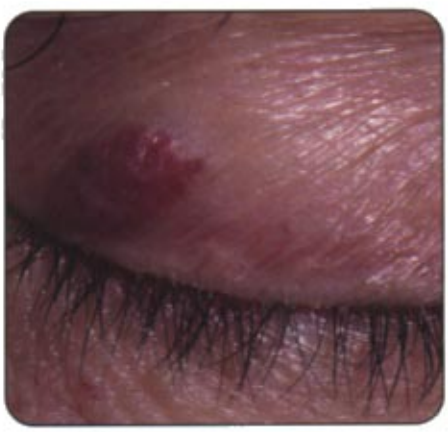


Fig. 1.96

Capillary hemangioma (strawberry nevus)

Capillary hemangioma is an uncommon tumor which develops soon after birth, grows for about 6–12 months, and then begins to spontaneously involute.

Signs

- Irregular, raised, red lesion (**Figs 1.97, 1.98**).
- The lesion blanches on pressure and may swell on crying.
- There may be intraorbital extension and, in some cases, the child may have similar skin lesions elsewhere (**Fig. 1.99**).

Systemic associations

- Thrombocytopenic purpura (Kasabach–Merritt syndrome).
- Maffucci syndrome.

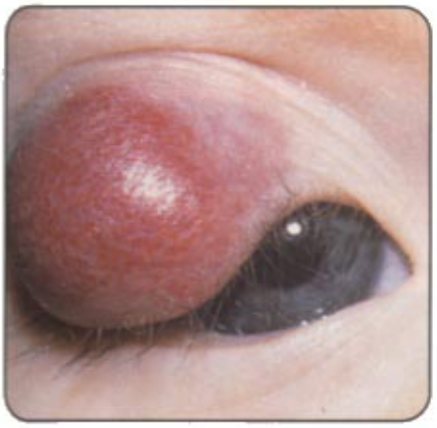


Fig. 1.97

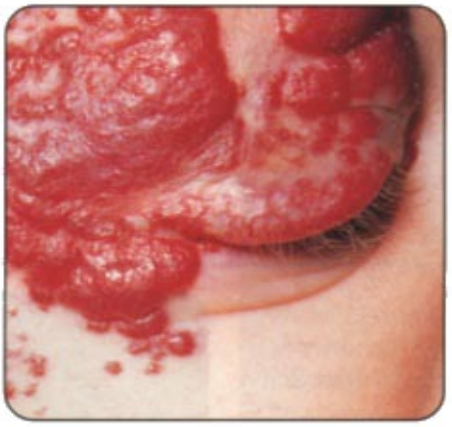


Fig. 1.98

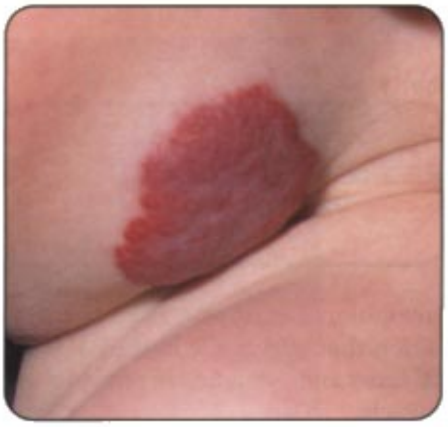


Fig. 1.99

Pyogenic granuloma

Pyogenic granuloma is an uncommon granulomatous hemangioma which is usually antedated by trauma or surgery.

Signs

- Fast-growing, pinkish, pedunculated or sessile mass (**Fig. 1.100**) which bleeds easily.

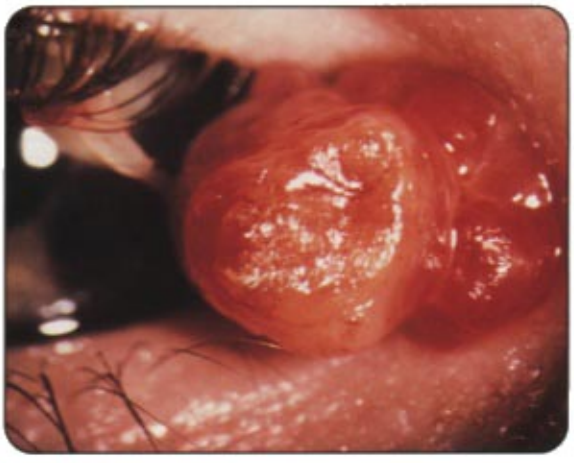


Fig. 1.100

Kaposi sarcoma

Kaposi sarcoma is an uncommon malignant tumor which typically affects patients who have AIDS.

Signs

- Starts with focal erythema, which progresses into a darker elevated lesion (**Fig. 1.101**).

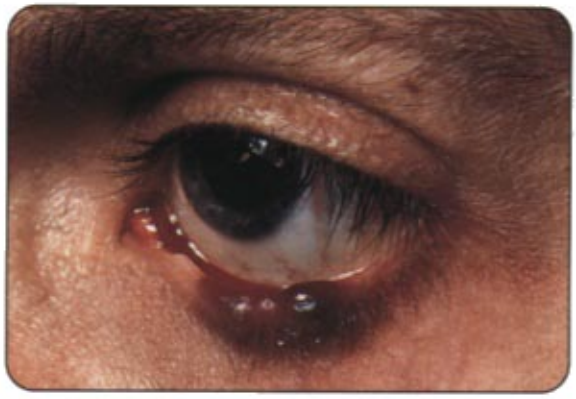


Fig. 1.101

ENTROPION

Involutional entropion

Involutional entropion is a very common age-related condition that affects the lower lid. It is caused by horizontal lid laxity and overriding of preseptal over pretarsal orbicularis muscle.

Signs

- Inturning of the lower eyelid (**Fig. 1.102**), which is easily everted by applying digital pressure to the inferior border of the tarsal plate.

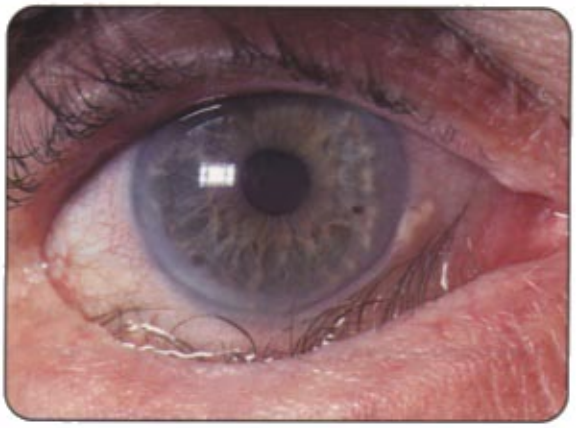


Fig. 1.102

Cicatricial entropion

Cicatricial entropion is an uncommon condition caused by conjunctival scarring.

Signs

- Inturning of usually the upper lid (**Fig. 1.103**), which cannot be corrected by digital pressure.

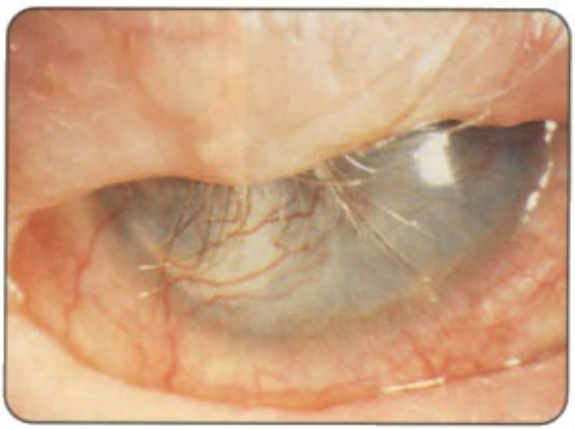


Fig. 1.103

ECTROPION

Involutional ectropion

Involutional ectropion is a very common age-related condition which affects the lower lid. It is caused by excessive horizontal eyelid length due to stretching of the medial and lateral canthal tendons and disinsertion of the lower lid retractors.

Signs

- Outward turning of the lower eyelid which may involve the lateral part more than the medial (**Fig. 1.104**).
- In some cases the medial part of the lid is involved more than the lateral (**Fig. 1.105**)
- Laxity of the medial canthal tendon is demonstrated by noting a marked temporal displacement of the lower punctum when the lid is pulled laterally.

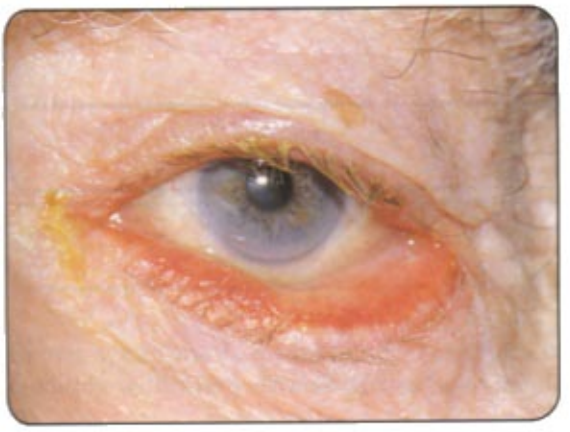


Fig. 1.104

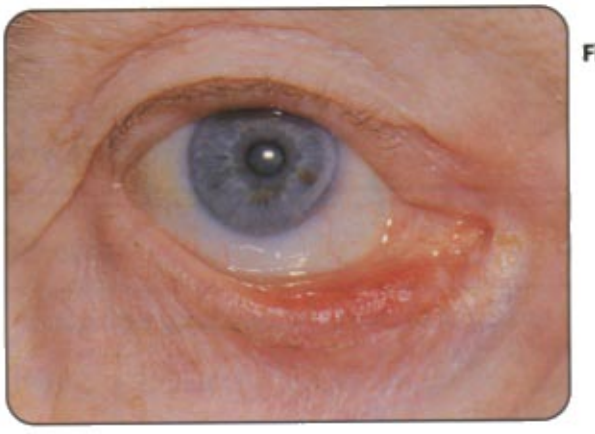


Fig. 1.105

Cicatricial ectropion

Signs

- Outward turning of the lower lid caused by contracture of skin and underlying tissues (**Fig. 1.106**).

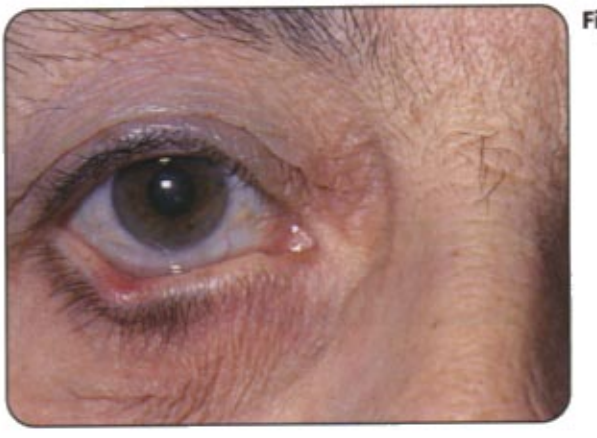


Fig. 1.106

Mechanical ectropion

Signs

- Outward turning of the lower lid caused by a large tumor (**Fig. 1.107**).

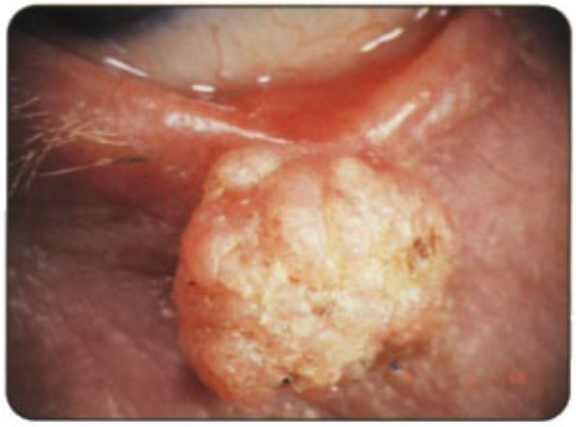


Fig. 1.107

Paralytic ectropion

Signs

- Outward turning of the lower lid associated with a facial nerve palsy (**Fig. 1.108**).
- Other signs include incomplete blinking (**Fig. 1.109**) and epiphora (**Fig. 1.110**).

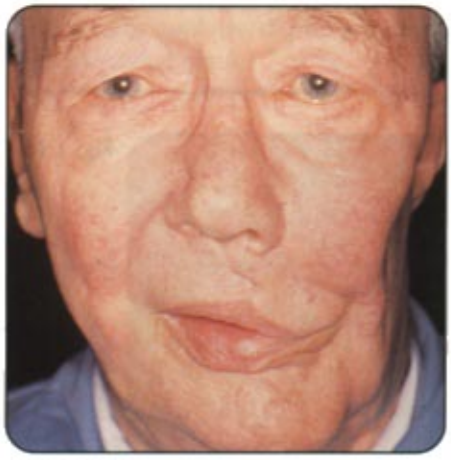


Fig. 1.108

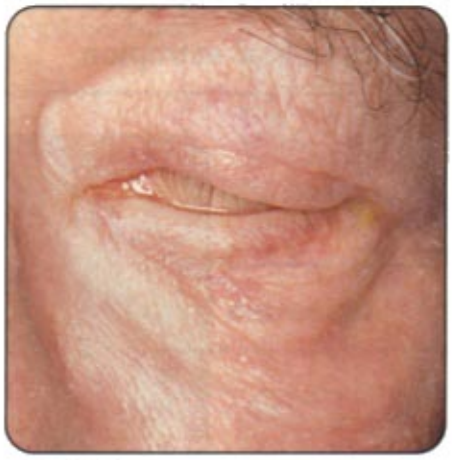


Fig. 1.109

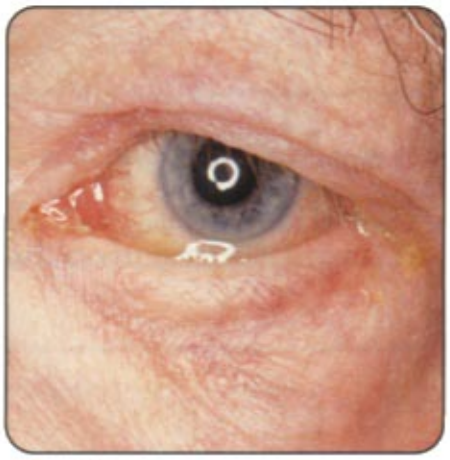


Fig. 1.110

PTOSIS

Neurogenic ptosis

Third-nerve palsy

Signs

- Usually severe, acute, unilateral ptosis associated with ophthalmoplegia (**Fig. 1.111**).

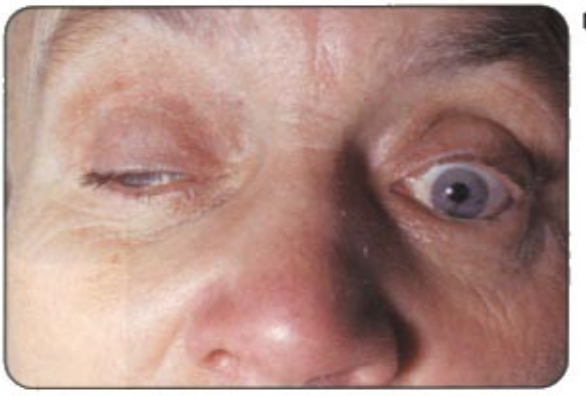


Fig. 1.111

Horner syndrome

Horner syndrome is a unilateral, congenital or acquired condition with many diverse causes (*see* Chapter 6).

Signs

- Mild ptosis associated with miosis (**Fig. 1.112**).

Look for

- Elevation of lower lid.
- Heterochromia iridis – if congenital or longstanding.
- Anhydrosis if the lesion is below the superior cervical ganglion.

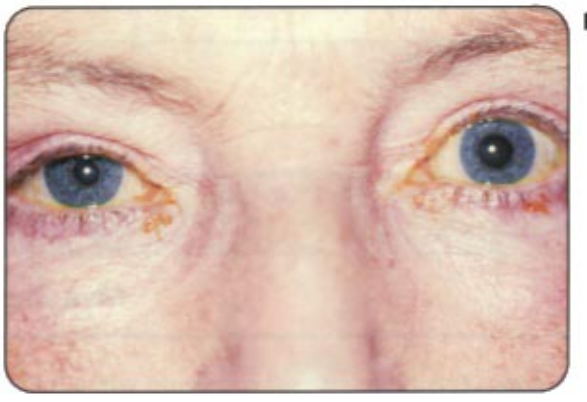


Fig. 1.112

Third-nerve misdirection

Third-nerve misdirection is a rare, acquired, unilateral condition which is usually preceded by a third-nerve palsy.

Signs

- Transient ptosis (**Fig. 1.113**) and bizarre movements of the upper lid which accompany various eye movements (**Fig. 1.114**).

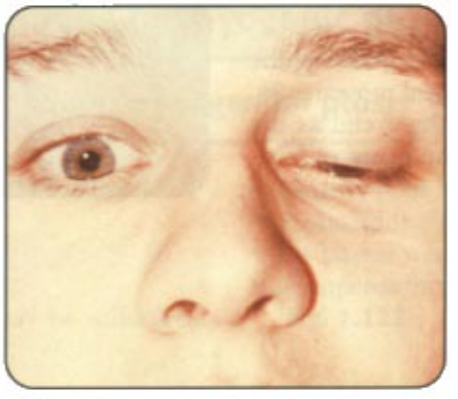


Fig. 1.113

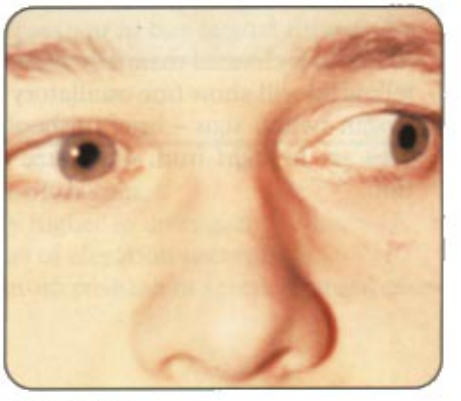


Fig. 1.114

Marcus Gunn jaw-winking syndrome

Marcus Gunn jaw-winking syndrome is an uncommon, congenital, unilateral condition.

Signs

- Ptosis (**Fig.1.115**) in which the ptotic lid retracts or 'winks' in conjunction with stimulation of the ipsilateral pterygoid muscles, as occurs when opening the mouth (**Fig. 1.116**), chewing, or moving the jaw to the contralateral side.

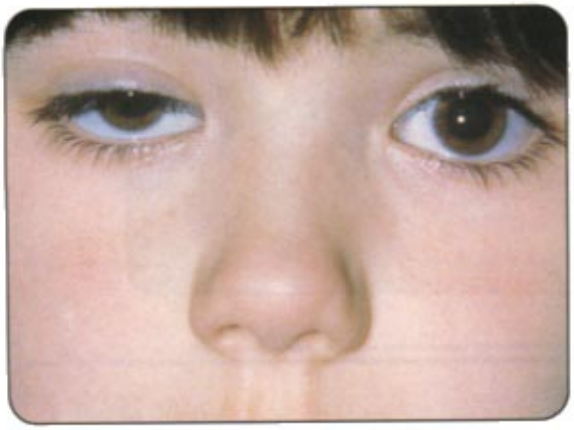


Fig. 1.115

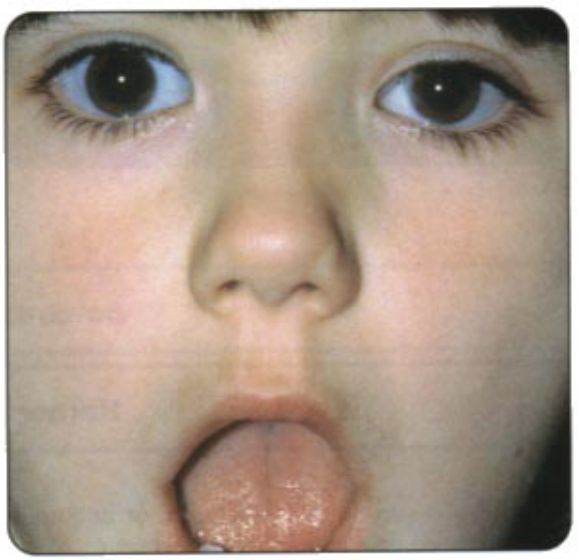


Fig. 1.116

Acquired myogenic ptosis

Myasthenia gravis

Myasthenia gravis is an uncommon condition which typically affects young females and frequently presents with ptosis and diplopia.

Ocular signs

- Insidious, bilateral but frequently asymmetric ptosis, worse with fatigue and in upgaze (**Fig. 1.117**).
- If one lid is elevated manually as the patient looks up, the fellow lid will show fine oscillatory vertical movements.
- Cogan twitch sign – brief upshoot of the eyelid as the eyes are brought from downgaze to the primary position.

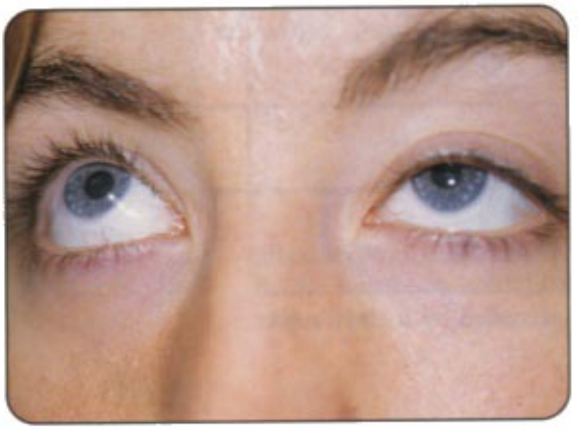


Fig. 1.117

Myotonic dystrophy

Myotonic dystrophy is a rare, dominantly inherited condition characterized by delayed relaxation of skeletal muscles after contraction, such as difficulty in relaxing grip.

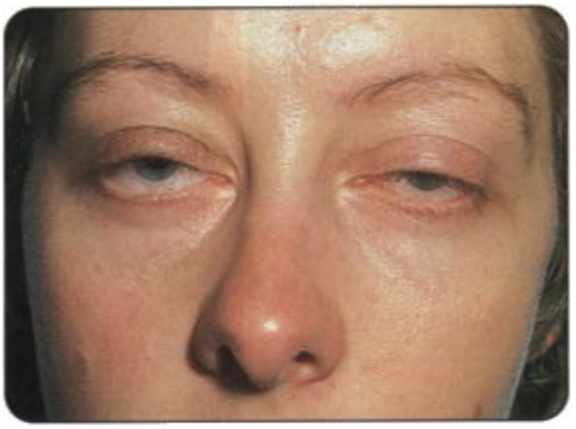


Fig. 1.118

Signs

- Bilateral ptosis and facial weakness gives rise to a mournful expression (**Fig. 1.118**).

Ocular signs

- Bilateral symmetric ptosis.
- Bilateral small pupils.
- Light-near dissociation of pupillary reactions.
- Presenile posterior stellate cataract.
- Pigmentary retinopathy.
- Symmetric ophthalmoplegia.

Ocular myopathy

Ocular myopathy is a very rare mitochondrial cytopathy which may occur in isolation or in association with oculopharyngeal dystrophy or Kearns–Sayre syndrome.

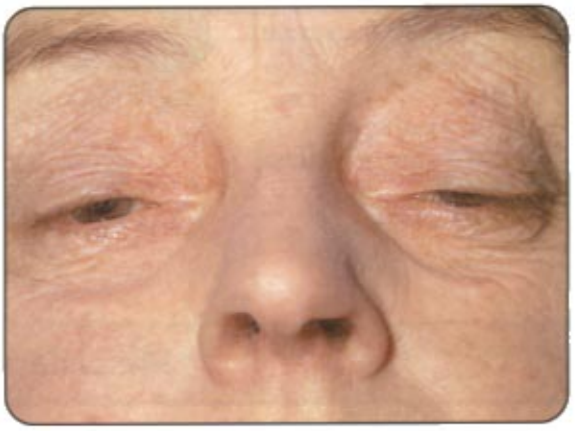


Fig. 1.119

Signs

- Slowly progressive, symmetric ptosis (**Fig. 1.119**) and symmetric external ophthalmoplegia.

Congenital myogenic ptosis

Simple congenital ptosis

This is the most common type of congenital ptosis.

Signs

Unilateral or bilateral ptosis which may be mild (**Fig. 1.120**) or severe (**Fig. 1.121**).

- Usually poor levator function.
- Ptotic lid is slightly higher in downgaze.
- Ipsilateral weakness of elevation is common.
- Compensatory chin-up position in severe bilateral cases (**Fig. 1.122**).

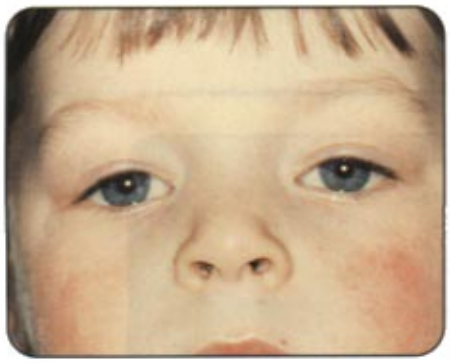


Fig. 1.120

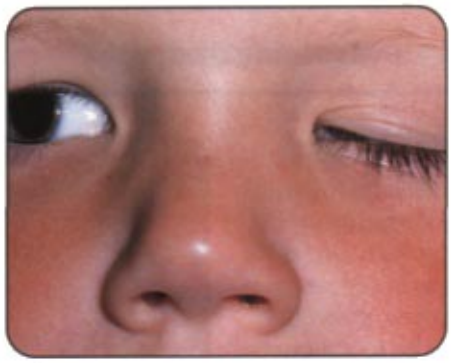


Fig. 1.121

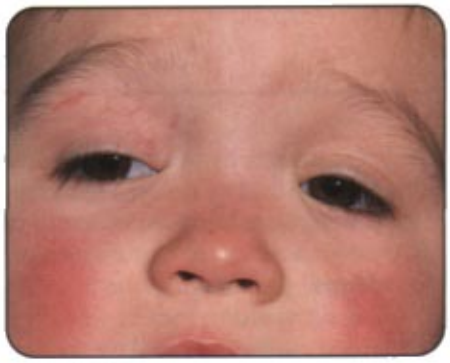


Fig. 1.122

Blepharophimosis syndrome

Blepharophimosis syndrome is a rare, dominantly inherited condition (**Fig. 1.123**).

Signs

- Moderate to severe symmetric ptosis (**Fig. 1.124**).
- Poor levator function.
- Short horizontal palpebral aperture.
- Epicanthus inversus.
- Lateral ectropion.
- Poorly developed nasal bridge.
- Hypoplasia of the superior orbital rim.
- Telecanthus.

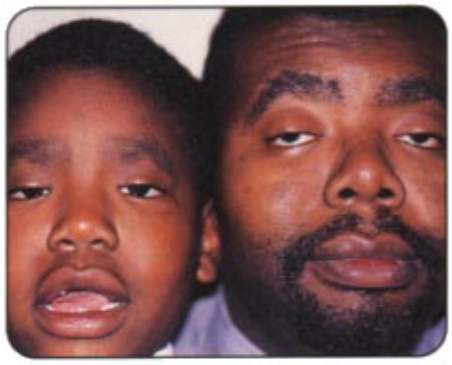


Fig. 1.123

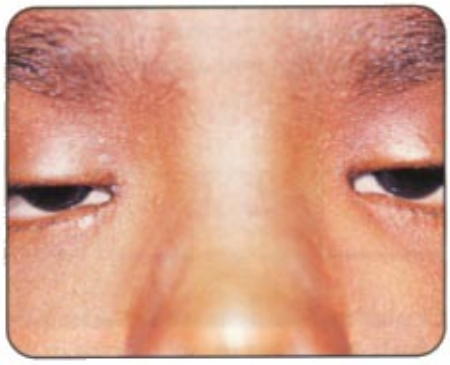


Fig. 1.124

Miscellaneous ptosis

Aponeurotic ptosis

This is a common, unilateral or bilateral ptosis caused by a defect in the aponeurosis. The latter may be involutional, or secondary to lid-swelling following surgery or blepharochalasis.

Signs

- Good levator function.
- High or absent upper-lid crease.
- Thinning of the eyelid above the tarsal plate.
- Deep upper sulcus (**Fig. 1.125**).

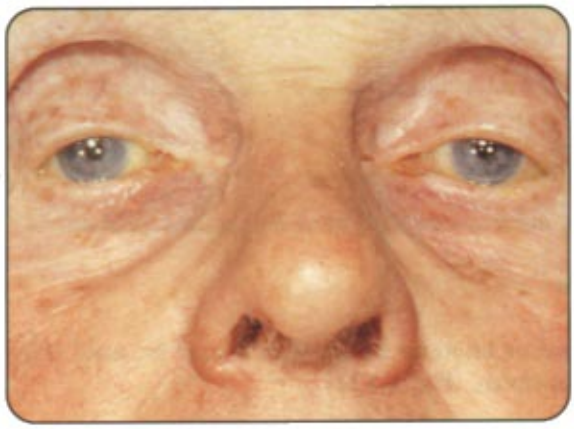


Fig. 1.125

Mechanical ptosis

Causes

- Severe lid edema (**Fig. 1.126**).
- Eyelid tumors such as plexiform neurofibromas (**Fig. 1.127**).
- Conjunctival scarring.
- Severe vernal disease.
- Orbital lesions such as cellulitis and anteriorly located tumors.
- Dermatochalasis.

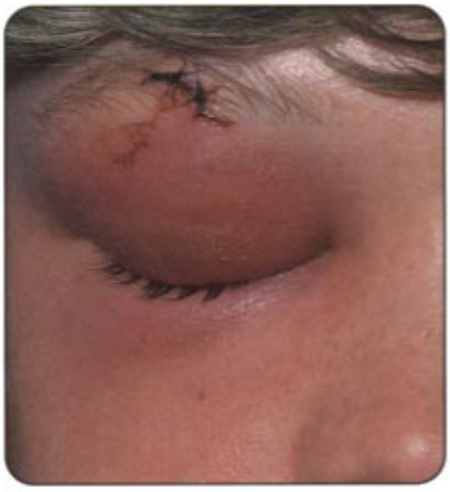


Fig. 1.126

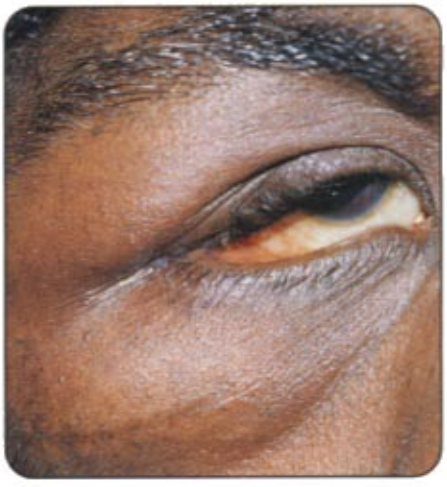


Fig. 1.127

LID RETRACTION

Thyroid eye disease

Signs

- Unilateral or bilateral lid retraction in the primary position (**Fig. 1.128**).

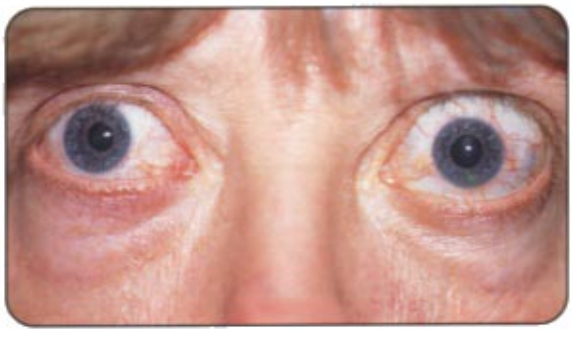


Fig. 1.128

Look for

- Lid lag in downgaze.
- Proptosis.
- Periorbital edema.
- Chemosis.
- Conjunctival hyperemia over the lateral recti.
- Superior limbic keratoconjunctivitis.
- Ocular motility defects.
- Choroidal folds.
- Optic nerve dysfunction.

Neurogenic lid retraction

Causes

- Misdirection of the third nerve.
- Marcus Gunn jaw-winking syndrome.
- Collier sign of the dorsal midbrain (Parinaud syndrome).
- Contralateral unilateral ptosis.
- Facial palsy due to unopposed action of the levator muscle.
- Sympathomimetic drops (phenylephrine, apraclonidine).
- Hydrocephalus.
- Transient 'eye popping' reflex in normal infants.

Fig. 1.129 shows right lid retraction caused by overaction of the levator and contralateral unilateral ptosis in a patient with myasthenia gravis.

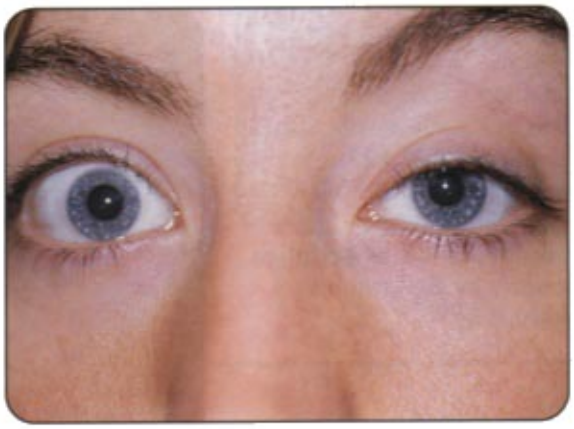


Fig. 1.129

Miscellaneous causes

- Surgical overcorrection of ptosis (**Fig. 1.130a**) which is frequently associated with lid lag in downgaze (**Fig. 1.130b**).
- Duane retraction syndrome.
- Scarring of upper-eyelid skin.
- Prominent globe (pseudo-lid-retraction).
- Uremia (Summerskill sign).
- Down syndrome.
- Congenital.

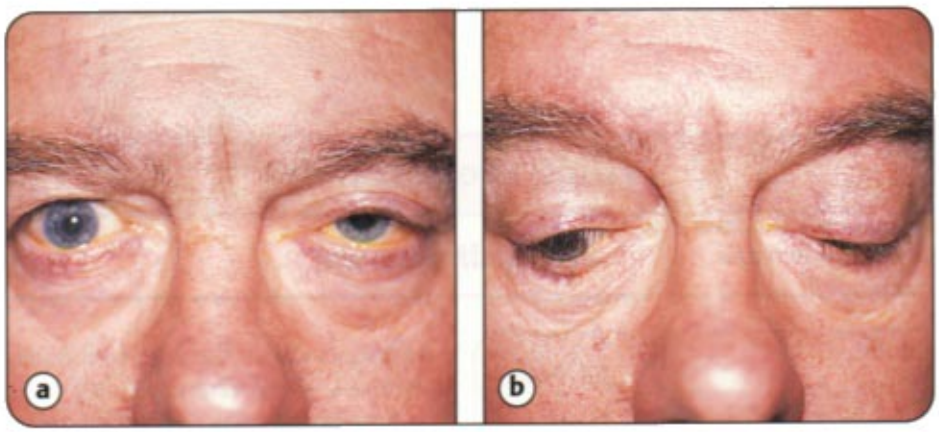


Fig. 1.130

MISCELLANEOUS DISORDERS

Acquired

Dermatochalasis

Signs

- Redundant upper-lid skin (**Fig. 1.131a**).
- It may cause a brow ptosis from a roll of skin that droops over the lid margin, concealing the true lid height (**Fig. 1.131b**).

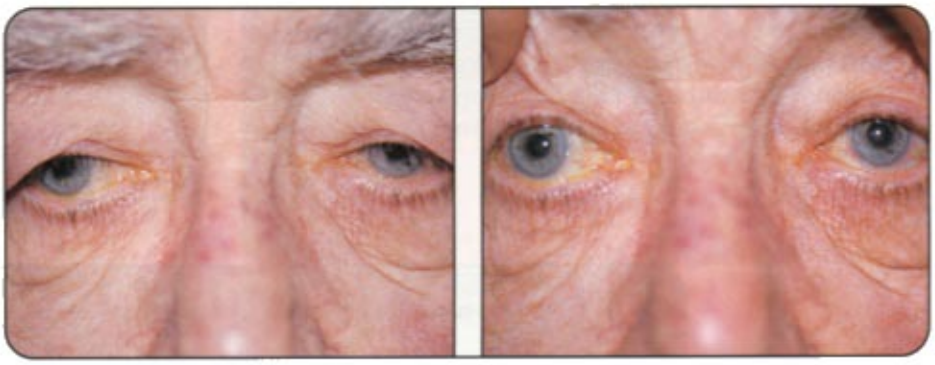


Fig. 1.131

Essential blepharospasm

Essential blepharospasm is an uncommon, idiopathic condition which usually presents during the sixth decade of life.

Signs

- Involuntary spasm of the orbicularis muscle and the upper facial muscles, which may render the patient temporarily blind (**Fig. 1.132**).

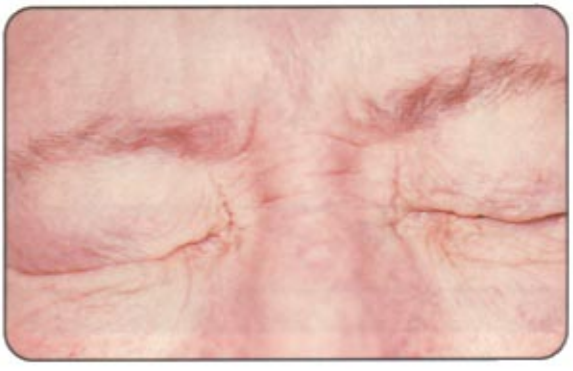


Fig. 1.132

Congenital

Epicanthic folds

Epicanthic folds is a very common condition which may give rise to a pseudoesotropia.

Signs

- Vertical folds of skin at the medial canthi (**Fig. 1.133**).

Fig. 1.133

Telecanthus

Telecanthus is an uncommon condition which may also be caused by facial trauma.

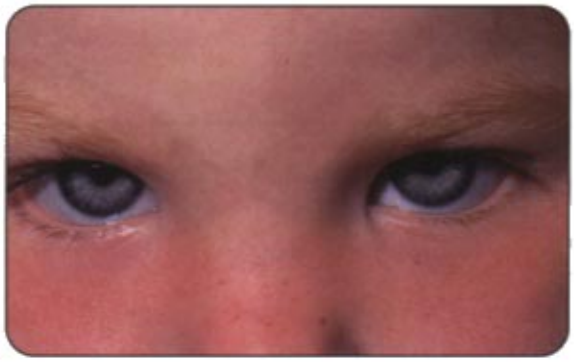


Fig. 1.134

Signs

- Increased distance between the medial canthi (**Fig. 1.134**), resulting from abnormally long medial canthal tendons. Telecanthus should not be confused with hypertelorism, in which there is wide separation of the orbits.

Table 1.4 Syndromes associated with congenital telecanthus

Noneponymous syndromes	• Möbius
• blepharophimosis	• Rubinstein–Taybi
• fetal valproate	• Treacher Collins
• craniofrontonasal dysplasia	• Saethre–Chotzen
• CHARGE	• Turner
	• von Recklinghausen neurofibromatosis
Eponymous syndromes	• Edward (trisomy 18)
• Waardenburg	• Noonan
• Freeman–Sheldon	

Coloboma

Signs

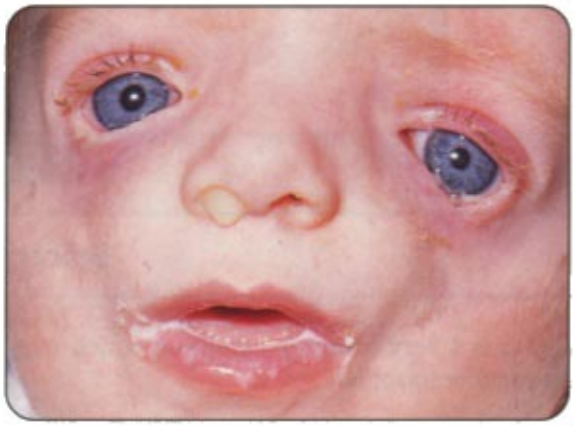


Fig. 1.135

- Coloboma is an uncommon, bilateral or unilateral, partial or full-thickness defect of the lid margin.
- Upper-lid coloboma occurs at the junction of the inner and middle thirds. It is not associated with systemic defects.
- Lower-lid coloboma occurs at the junction of the middle and outer thirds. It is frequently associated with systemic abnormalities such as the Treacher Collins syndrome (**Fig. 1.135**).

Epiblepharon

Epiblepharon is very common in orientals. It should not be confused with the much less common congenital entropion.

Signs

- Upward riding of skin and orbicularis over the inferior tarsus, with inrolling of lashes (**Fig. 1.136**). The nasal third of the lid is most commonly affected.

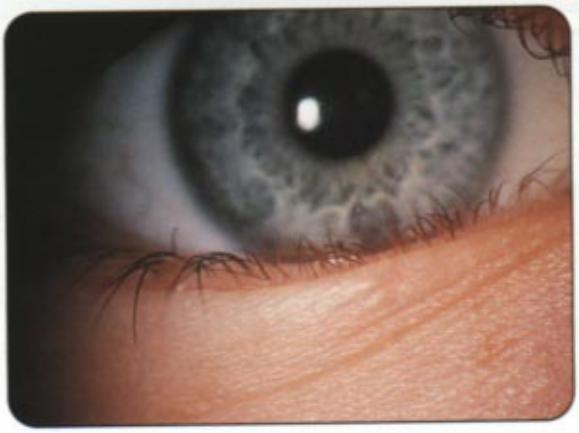


Fig. 1.136

Cryptophthalmos

Cryptophthalmos is a very rare, recessively inherited condition which may be associated with Fraser syndrome.

Signs

- Absence of formed lids resulting in eyes covered by a layer of skin (**Fig. 1.137**).
- There is no conjunctival sac present and the eyes are malformed.

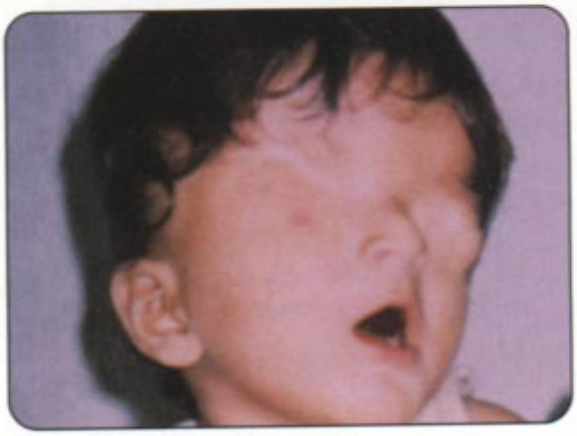


Fig. 1.137

2 The orbit

BONY ABNORMALITIES 44

Hypertelorism
Orbital dystopia

ENOPHTHALMOS 45

PROPTOSIS 46

PROPTOSIS IN NEONATES AND INFANTS 47

Tumors
- Capillary hemangioma
- Juvenile xanthogranuloma
- Other tumors

Cystic lesions
Shallow orbits

PROPTOSIS IN CHILDREN 50

Inflammatory causes
- Orbital cellulitis
- Idiopathic orbital inflammatory syndrome (pseudotumor)
- Thyroid eye disease

Benign tumors
- Lymphangioma
- Plexiform neurofibroma
- Optic nerve glioma

Malignancies
- Rhabdomyosarcoma
- Acute leukemia
- Metastatic neuroblastoma
- Other tumors

Langerhans-cell histiocytosis
Other causes

PROPTOSIS IN ADULTS 55

Congestive proptosis
- Thyroid eye disease
- Orbital cellulitis
- Idiopathic orbital inflammatory syndrome (pseudotumor)
- Carotid–cavernous fistula
- Cavernous sinus thrombosis
- Metastatic tumors

Chronic axial proptosis
- Thyroid eye disease
- Cavernous hemangioma
- Optic nerve sheath hemangioma
- Varix

Chronic nonaxial proptosis
- Lymphoid tumors
- Sphenoidal ridge meningioma
- Mucoceles
- Paget disease of the skull
- Orbital extension of sinus tumors
- Other causes

LACRIMAL ENLARGEMENT 63

Unilateral enlargement
- Dacryoadenitis
- Dacryops
- Pleomorphic adenoma (benign mixed cell tumor)
- Malignant lacrimal gland tumors

Bilateral enlargement

ORBITAL RIM LESIONS 65

DYNAMIC PROPTOSIS 66

- Intermittent proptosis
- Pulsatile proptosis

Table 2.1 Clinical signs in orbital disease

Bony	Restricted ocular motility
• hypertelorism • dystopia	• by an orbital mass • restrictive myopathy • ocular motor nerve palsy • muscle entrapment in a fracture
Soft tissue	**Posterior segment**
• lid edema • ptosis • chemosis	• disc edema • optic atrophy • chorioretinal folds • retinal vascular congestion
Globe malposition	
• proptosis • enophthalmos • displacement	

BONY ABNORMALITIES

Hypertelorism

Signs

- Wide separation of the orbits (**Fig. 2.1**), which is determined by measuring the interpupillary distance.
- Normal readings at 0–2 years of age are 45–54 mm and at 2–14 years, 54–63 mm.
- Hypertelorism may be caused by facial trauma or by the congenital abnormalities shown in **Table 2.2**.

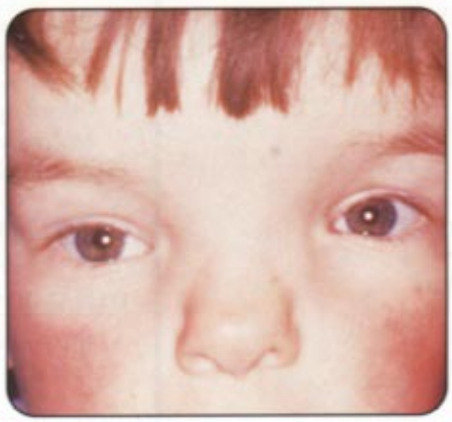


Fig. 2.1

Table 2.2 Causes of congenital hypertelorism

Craniosynostoses	Miscellaneous
• Apert • Pfeiffer • Saethre–Chotzen • Crouzon • Roberts • craniofrontal dysplasia	• frontonasal dysplasia • basal encephalocele with wide nasal bridge • idiopathic (Greig syndrome)
Syndromes	
• Noonan • Peters-plus • Patau (trisomy 13) • Gorlin–Goltz • Hurler • Meckel–Gruber • cat-eye	

Orbital dystopia

Signs

- Asymmetry of levels of the inferior orbital margins.

Figure 2.2 shows dystopia in a child with a facial clefting syndrome.

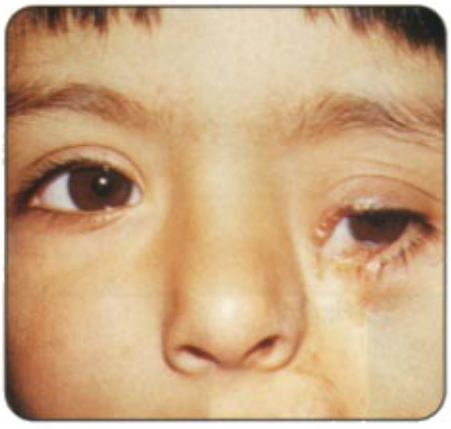


Fig. 2.2

Table 2.3 Causes of orbital dystopia

Congenital
• idiopathic
• Goldenhar syndrome
• hemifacial microsomia
• coronal craniosynostoses
• facial clefting syndromes
Acquired
• fractures of orbital floor or rim
• fibrous dysplasia
• meningioma en plaque

ENOPHTHALMOS

Enophthalmos is a condition in which the globe is recessed within the orbit.

Causes

Figure 2.3 shows Duane retraction syndrome in which there is retraction of the globe on adduction although in some cases – as in the one shown here – there may be true enophthalmos in the primary position of gaze.

Figure 2.4 shows bilateral severe enophthalmos associated with phthisis bulbi.

Figure 2.5 shows a patient with right enophthalmos associated with microphthalmos, before (**Fig. 2.5a**) and after (**Fig. 2.5b**) the insertion of a cosmetic contact lens.

Figure 2.6 shows right enophthalmos caused by a severe blowout fracture of the orbital floor.

Figure 2.7 shows a blind child with the eye-poking (oculodigital) sign, which results in atrophy of orbital fat and enophthalmos.

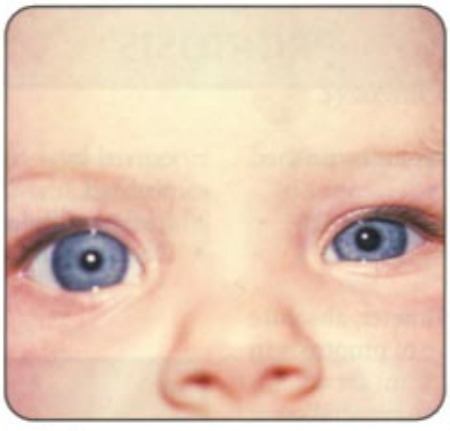


Fig. 2.3

Table 2.4 Causes of enophthalmos

Small globe
• phthisis bulbi
• microphthalmos
• nanophthalmos
Structural bony abnormalities
• after blowout fracture of orbital floor
• congenital bony defects as in neurofibromatosis type I
Atrophy of orbital contents
• after radiation
• Parry–Romberg hemifacial atrophy
• scleroderma
• chronic maxillary sinusitis
• eye poking (oculodigital sign) in blind infants
Cicatrizing orbital lesions
• metastatic scirrhous breast carcinoma
• chronic sclerosing inflammatory orbital disease
Duane retraction syndrome

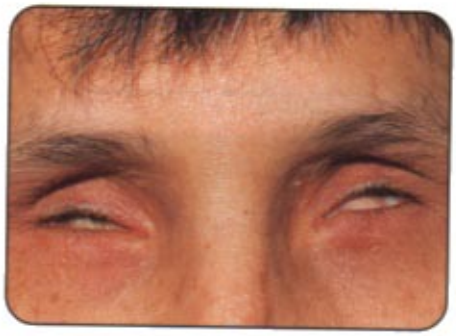


Fig. 2.4

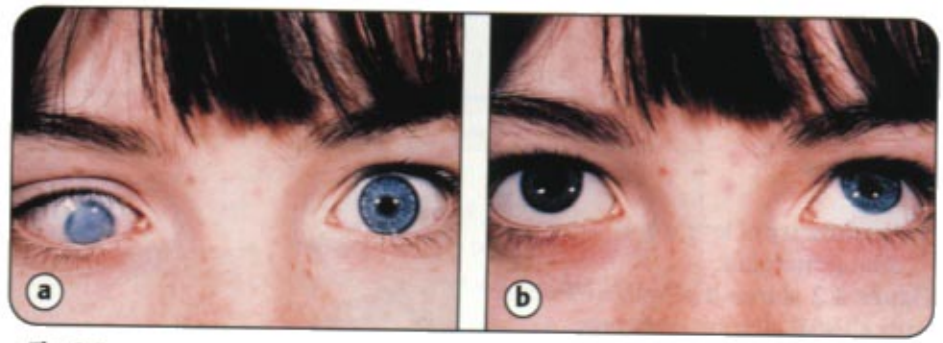


Fig. 2.5

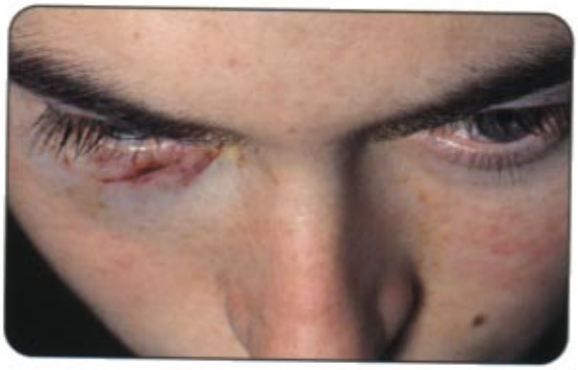


Fig. 2.6

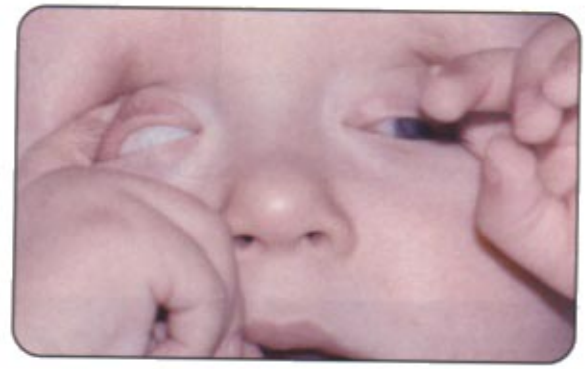


Fig. 2.7

PROPTOSIS

Proptosis is a condition in which the globe is pushed forward.

Signs

- Measured using the Hertel exophthalmometer, absolute values greater than 20 mm are indicative of proptosis in whites, but slightly higher readings (≤ 22 mm) are acceptable in blacks and orientals, who tend to have shallower orbits.
- Asymmetric readings between the two sides greater than 2 mm imply orbital disease.
- Proptosis is best detected by looking down on the patient from above (**Fig. 2.8**).

Conditions that may mimic proptosis

- Ipsilateral large globe (e.g. buphthalmos, high myopia). **Figure 2.9a–c** shows a patient with left pseudoproptosis caused by a combination of left high myopia and right phthisis.
- Ipsilateral lid retraction.
- Contralateral enophthalmos.

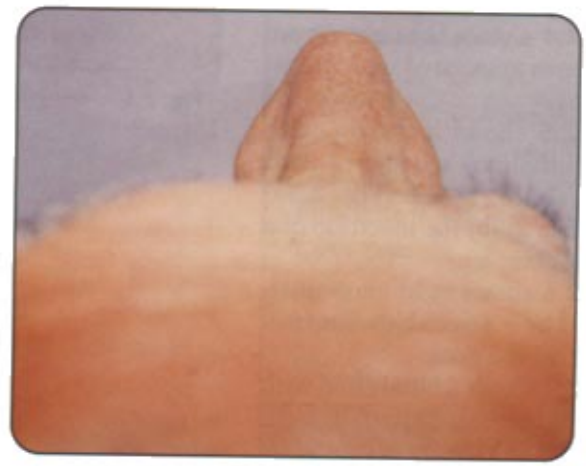


Fig. 2.8

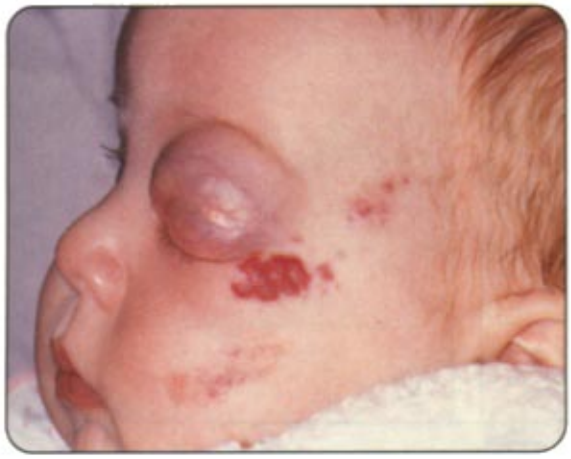


Fig. 2.13

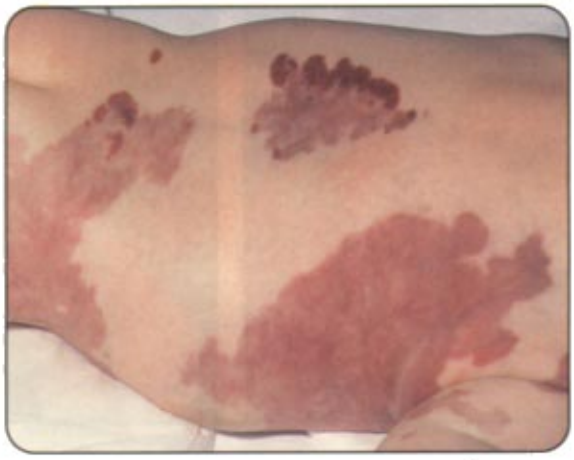


Fig. 2.14

Juvenile xanthogranuloma

Juvenile xanthogranuloma is a rare, benign condition which presents in infancy.

Signs

- Proptosis (**Fig. 2.15a**) which is often associated with strabismus (**Fig. 2.15b**).

Look for

- Skin lesions.
- Iris involvement and hypochromic heterochromia.
- Spontaneous hyphema.

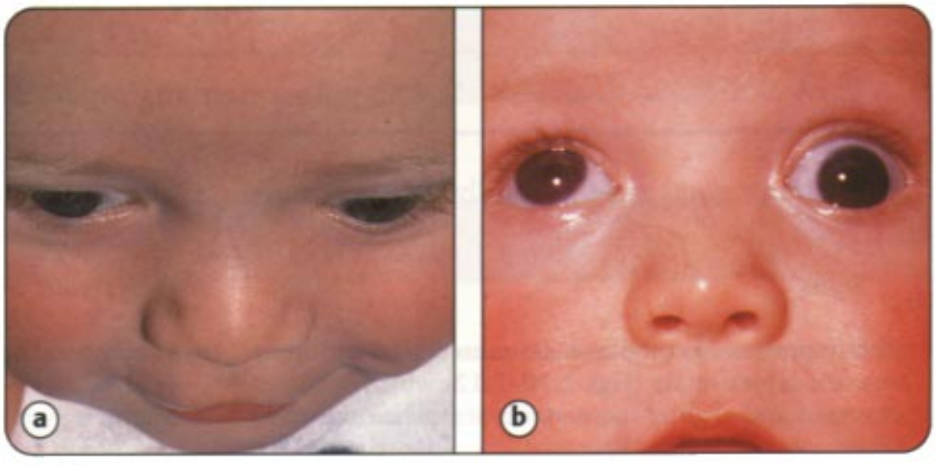


Fig. 2.15

Other tumors

- Teratoma is a very rare benign tumor which presents at birth with massive proptosis.
- Rhabdomyosarcoma may very rarely present during infancy.
- Retinoblastoma invading the orbit may very rarely present during infancy.
- Acute leukemia (**Fig. 2.16**) may very rarely present during infancy.

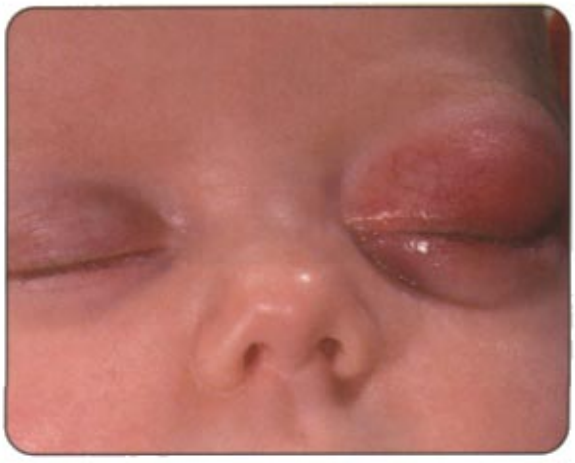


Fig. 2.16

Cystic lesions

- Microphthalmos with cyst is caused by incomplete closure of the fetal fissure leading to prolapse of cystic tissue into the orbit. This is characterized by a bluish cyst, with upward displacement of the microphthalmic eye.
- Congenital cystic eyeball (anophthalmos with cyst).
- Anterior orbital encephalocele causes a slowly progressive, pulsatile proptosis which increases in size on crying or straining. The globe is displaced laterally.
- Posterior orbital encephalocele is similar to the anterior variety except that the globe is displaced downward (**Fig. 2.17**). It may be associated with neurofibromatosis type I.

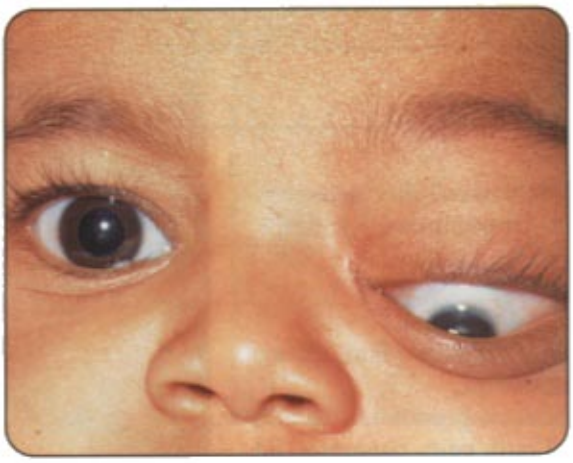


Fig. 2.17

Shallow orbits

Figure 2.18a–c shows bilateral proptosis associated with shallow orbits in a child with Roberts syndrome.

Table 2.5 Causes of shallow orbits

Craniosynostoses	Other bony defects
• Crouzon syndrome	• osteogenesis imperfecta
• Apert syndrome	• osteopetrosis
• Pfeiffer syndrome	• hypophosphatemia
• Saethre–Chotzen syndrome	**Chromosomal abnormalities**
• Roberts syndrome	• trisomy 13 (Patau syndrome)
	• trisomy 18 (Edward syndrome)

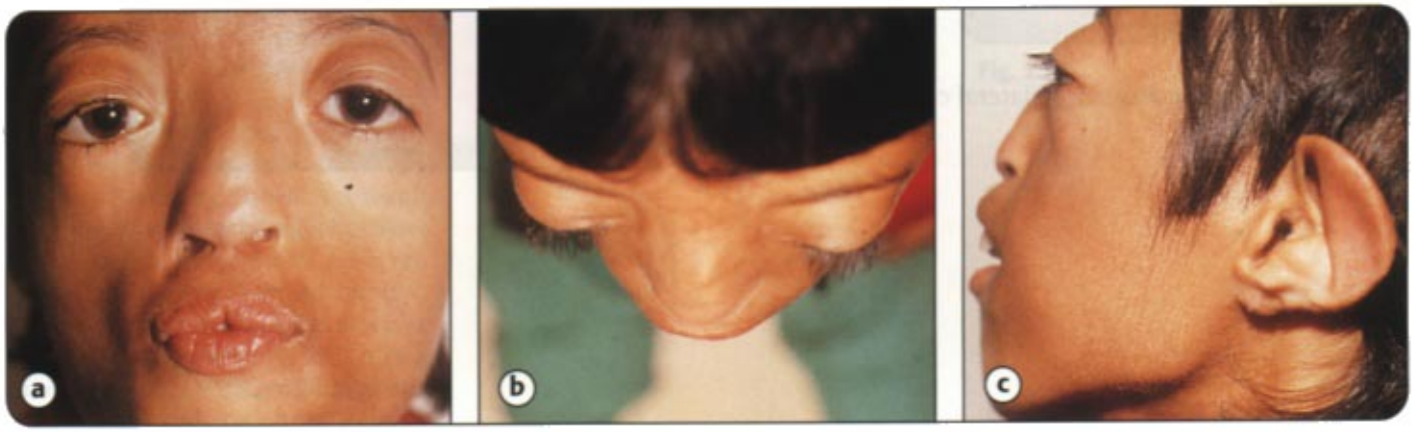


Fig. 2.18

PROPTOSIS IN CHILDREN

Inflammatory causes

Orbital cellulitis

In children, orbital cellulitis is usually secondary to ethmoiditis.

Signs

- The child is very unwell and has a high temperature.
- Rapid onset of proptosis (usually down and out), pain, chemosis, lid edema (**Fig. 2.19**), and ocular motility restriction. **Figure 2.20** shows a patient after drainage of an orbital abscess.
- In severe cases there may be signs of optic nerve dysfunction.

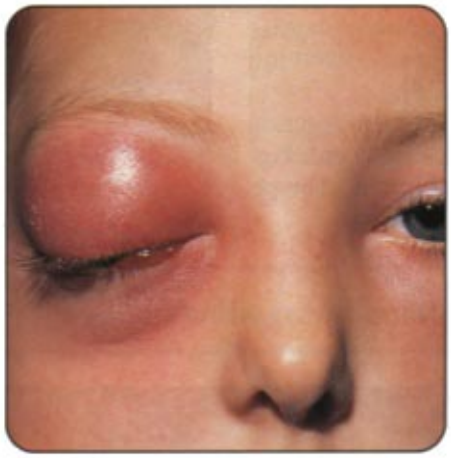


Fig. 2.19

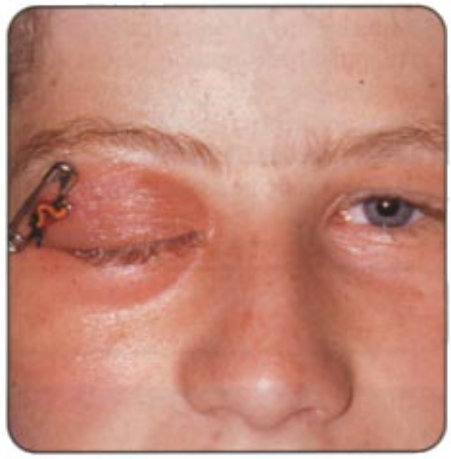


Fig. 2.20

Idiopathic orbital inflammatory syndrome (pseudotumor)

Presentation of idiopathic orbital inflammatory syndrome is usually between the ages of 6 and 14 years. About one-third of patients have bilateral involvement.

Signs

- Subacute-onset axial proptosis associated with chemosis and lid edema (**Fig. 2.21**).

Look for

- Wegener granulomatosis in bilateral cases.

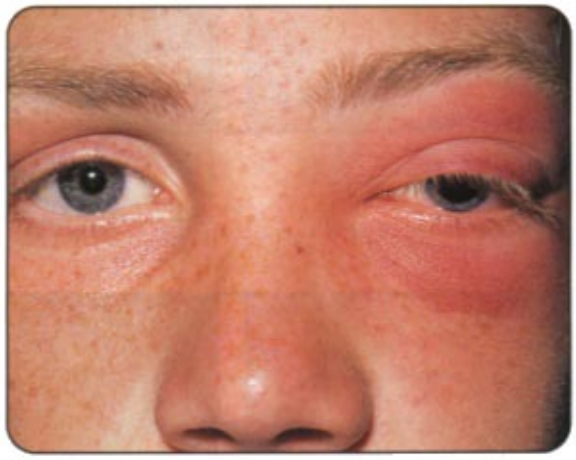


Fig. 2.21

Thyroid eye disease

Thyroid eye disease may rarely present in children as young as 10 years and cause either unilateral or bilateral proptosis (**Fig. 2.22a**) and lid retraction (**Fig. 2.22b**).

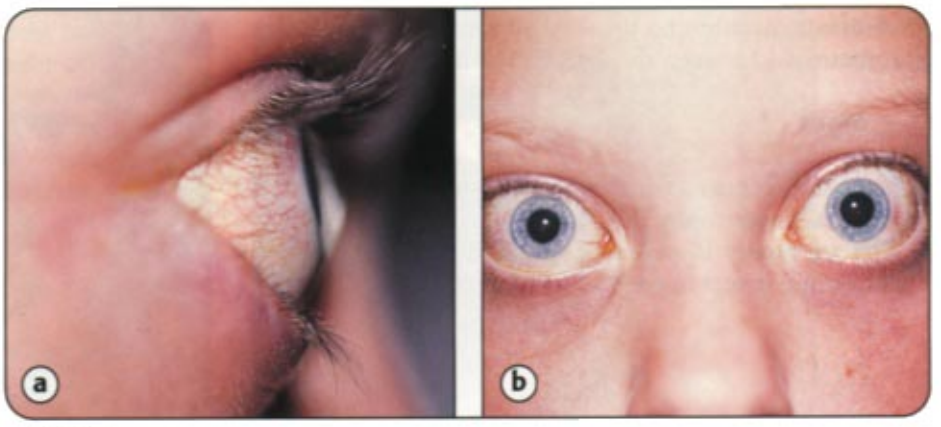


Fig. 2.22

Benign tumors

Lymphangioma

Presentation of lymphangioma is between the ages of 1 and 15 years. The lesion may be limited to the conjunctiva or eyelid, lie deep within the orbit, or be a combination of the two. It may also involve the face, sinuses, and oropharynx (**Fig. 2.23**).

Signs

- Soft bluish mass, most commonly located in the superior orbit (**Fig. 2.24**).
- The lesion may remain stationary for long periods of time or it may suddenly enlarge either as a result of spontaneous bleeding (chocolate cyst) or in association with an upper respiratory tract infection.
- Visual compromise or extraocular motility impairment are much more common with lymphangioma than with capillary hemangioma.

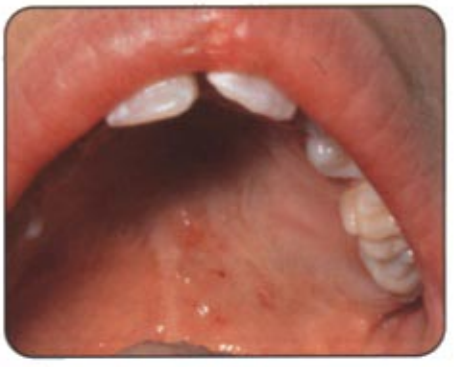


Fig. 2.23

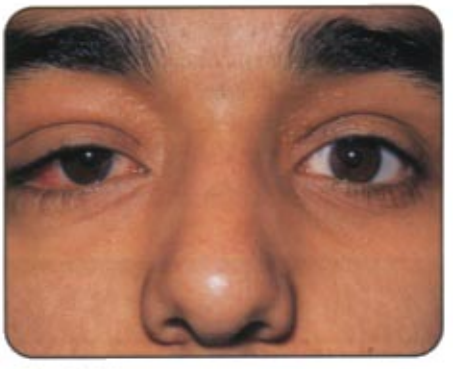


Fig. 2.24

Plexiform neurofibroma

Plexiform neurofibroma presents in patients with neurofibromatosis-I between the ages of 2 and 5 years.

Signs

- Nonaxial proptosis which may be associated with eyelid or facial hypertrophy (**Fig. 2.25**).
- The proptosis is pulsatile if there is an associated congenital defect in the sphenoid bone.

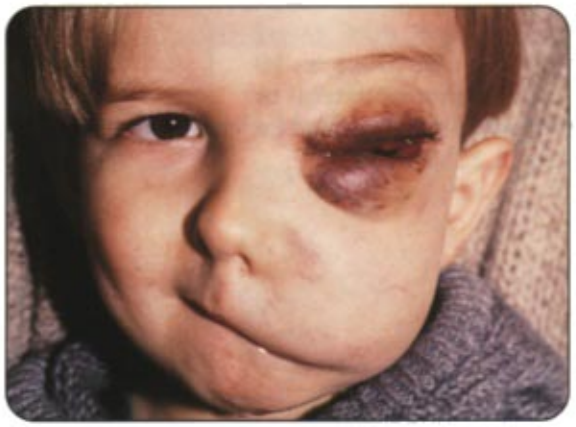


Fig. 2.25

Optic nerve glioma

Presentation of optic nerve glioma is between the ages of 2 and 7 years. Neurofibromatosis-I is present in 50% of patients with unilateral tumors and 100% of those with bilateral tumors.

Signs

- Slowly progressive axial proptosis (**Fig. 2.26**) associated with decreased visual acuity and an afferent pupillary conduction defect.
- The optic disc may show swelling, atrophy, or the presence of opticociliary shunts.
- There is a disproportionate loss of visual acuity compared to proptosis.

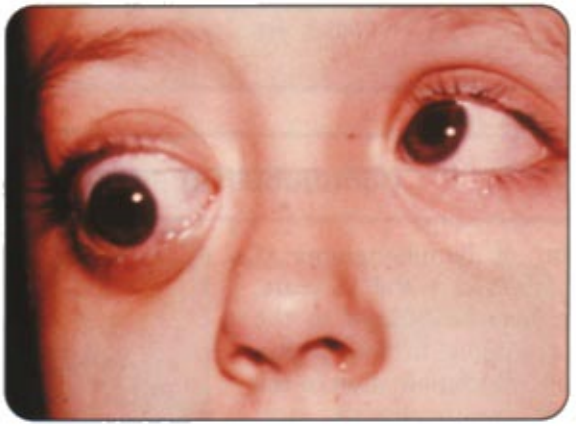


Fig. 2.26

Malignancies

Rhabdomyosarcoma

Presentation of rhabdomyosarcoma is most frequently at about age 7 years. The tumor is more common in boys than in girls.

Signs

- Rapid onset of progressive painful proptosis associated with chemosis and lid edema.
- The most frequent location of the tumor is retrobulbar, followed by superior (**Fig. 2.27**) and inferior.

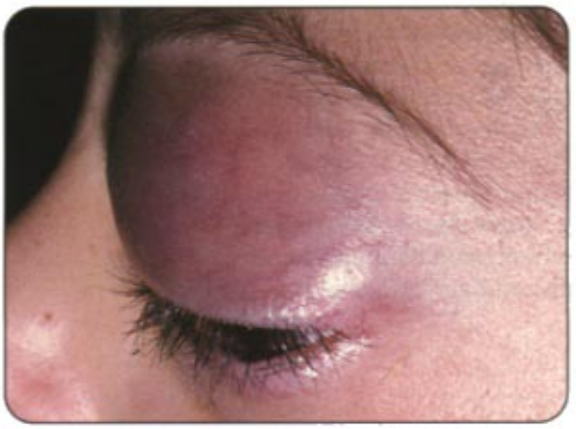


Fig. 2.27

Acute leukemia

Presentation of acute leukemia occurs most frequently at about the age of 7 years. Orbital involvement may occur in chloroma, which affects soft tissues and is not associated with abnormal peripheral blood, as well as in leukemia with established systemic disease and an abnormal blood film. Chloroma has a predilection for Asian and African children.

Signs

- Rapidly progressive proptosis which may be associated with ecchymosis and lid edema (**Fig. 2.28**).

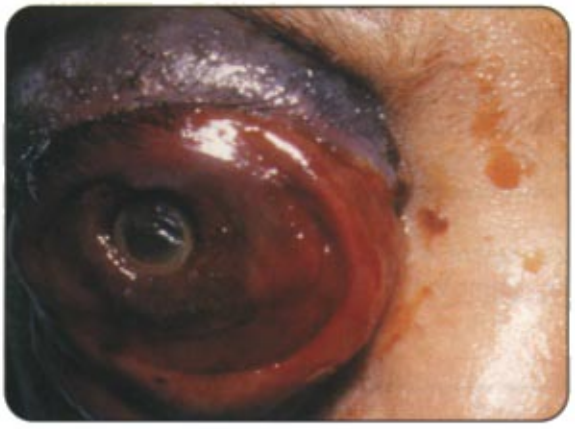


Fig. 2.28

Metastatic neuroblastoma

Metastatic neuroblastoma presents during the first 5 years of life. The primary tumor usually develops in the abdomen. Tumors arising from the neck or mediastinum may cause Horner syndrome. Forty percent of metastases involve both orbits.

Signs

- Sudden onset of rapidly progressive proptosis, frequently associated with ecchymosis and a superolateral orbital mass (**Fig. 2.29**).

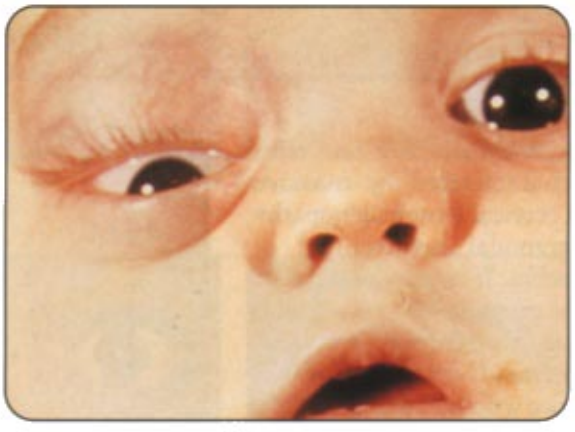


Fig. 2.29

Other tumors

- Neglected retinoblastoma may occasionally present with orbital involvement (**Fig. 2.30**).
- Metastatic Ewing sarcoma – rapid proptosis, ecchymosis, and chemosis.
- Metastatic Wilm tumor – which may be associated with aniridia.
- Fibrosarcoma – typically occurs in children who have previously received radiotherapy to the orbit for retinoblastoma.
- Sinus tumors invading the orbit – such as Burkitt lymphoma, osteogenic sarcoma, and embryonal rhabdomyosarcoma.

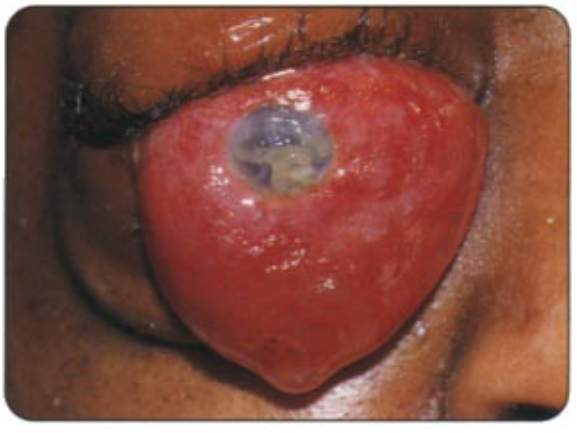


Fig. 2.30

Langerhans-cell histiocytosis

Langerhans-cell histiocytosis is a rare multisystem disorder consisting of three related and overlapping conditions:

- Eosinophilic granuloma – lesions confined to bone.
- Hand–Schüller–Christian disease – a triad of (a) diabetes insipidus, (b) proptosis, and (c) bony skull defects.
- Letterer–Siwe disease – aggressive visceral involvement.

Orbital involvement occurs in 25% of cases with either eosinophilic granuloma or Hand–Schüller–Christian disease.

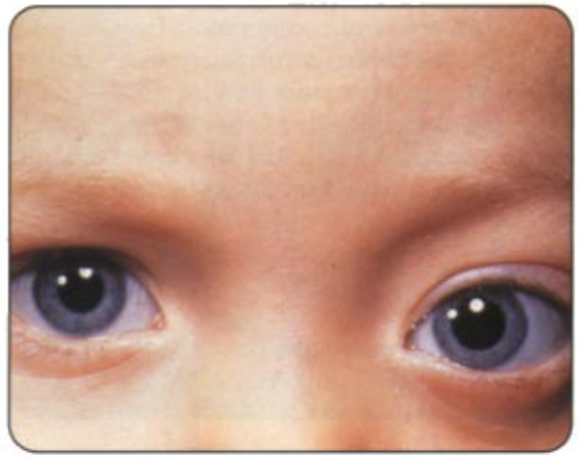


Fig. 2.31

Signs

- Bilateral or unilateral bony lysis and soft-tissue growth which typically involves the superolateral orbit (**Fig. 2.31**).

Other causes

- Sinus histiocytosis is a rare condition characterized by massive painless cervical lymphadenopathy and extranodal involvement of other viscera. It causes bilateral or unilateral proptosis associated with soft-tissue involvement of the orbit.
- Deep dermoid cyst presents in adolescence with nonaxial proptosis.
- Fibrous dysplasia typically involves the orbital roof and causes proptosis with downward displacement of the globe. It may be associated with McCune–Albright syndrome.
- Juvenile ossifying fibroma arises from either the roof or the ethmoids. It causes chronic painless proptosis with downward or lateral globe displacement.
- Osteopetrosis is an hereditary condition characterized by increased density and thickness of bone and increased susceptibility to fracture. It causes bilateral proptosis (**Fig. 2.32**) and optic atrophy.

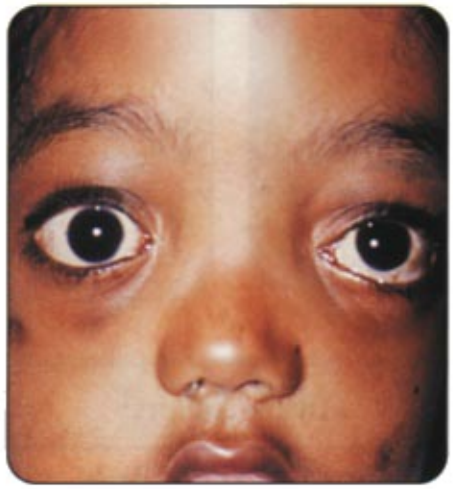


Fig. 2.32

Table 2.6 Causes of rapid proptosis in children

Nonmalignant
• orbital cellulitis
• ruptured deep dermoid cyst
• chocolate cyst with lymphangioma
• pseudotumor
Malignant
• rhabdomyosarcoma
• acute leukemia
• metastases

PROPTOSIS IN ADULTS

Congestive proptosis

Congestive proptosis is a group of disorders usually characterized by a rapid onset of variable lid edema, chemosis, conjunctival congestion, ocular motility restriction and, frequently, pain.

Thyroid eye disease

Thyroid eye disease is the most common cause of proptosis in adults.

Signs

- Axial proptosis which may be unilateral (**Fig. 2.33a and b**) or bilateral (**Fig. 2.34a and b**).

Look for

Lid retraction (**Fig. 2.35**) and lid lag.
Injection over horizontal recti (**Fig. 2.36**).
Superior limbic keratoconjunctivitis (**Fig. 2.37**).
Restrictive ocular motility defects.
Signs of optic nerve compression.
Chorioretinal folds (**Fig. 2.38**).

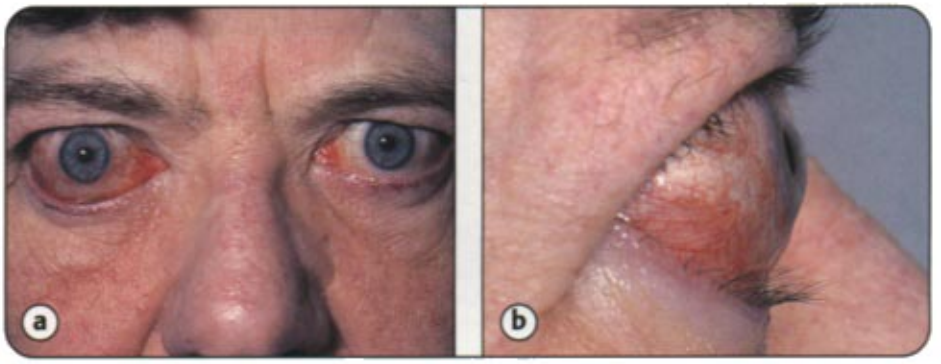


Fig. 2.33

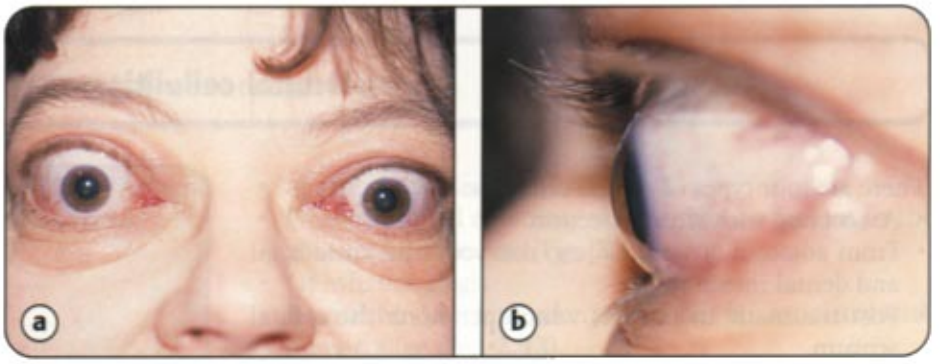


Fig. 2.34

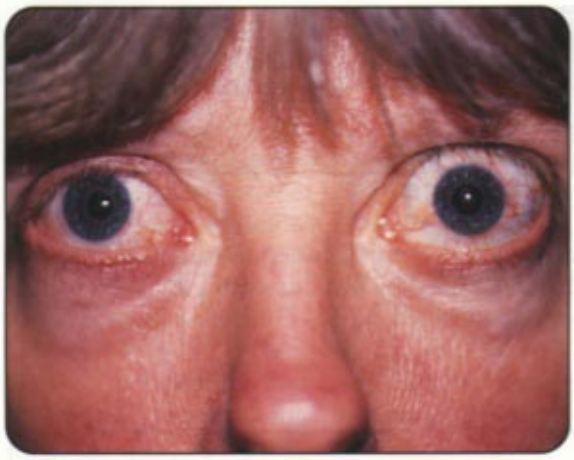


Fig. 2.35

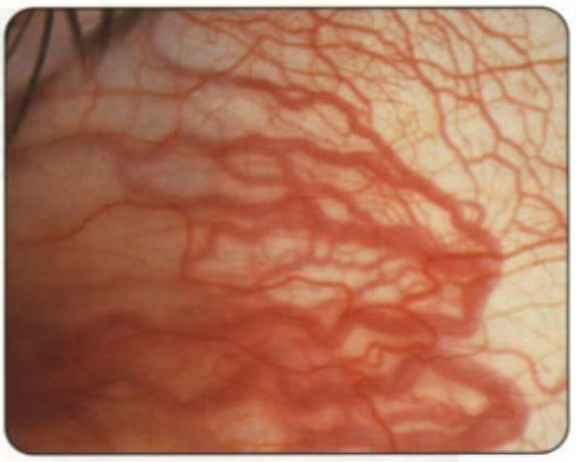


Fig. 2.36

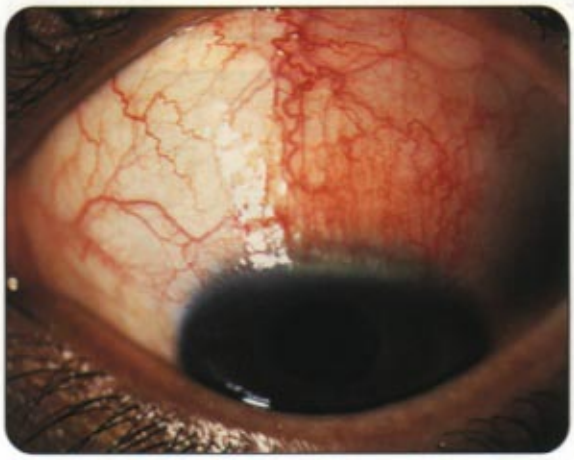


Fig. 2.37

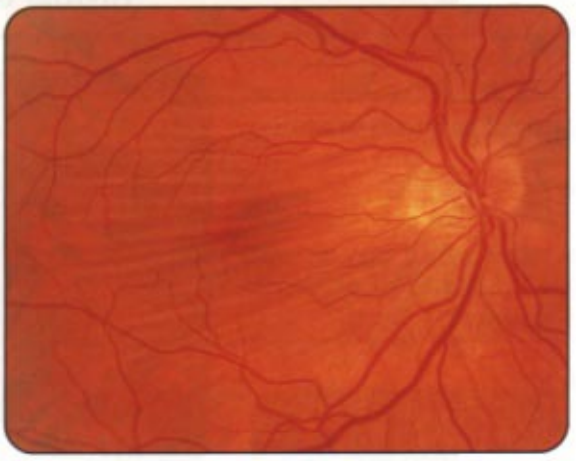


Fig. 2.38

Orbital cellulitis

There are four types of orbital cellulitis:

- Associated with sinus infection.
- From adjacent structures (e.g. dacryocystitis, midfacial and dental infection).
- Post-traumatic in injuries which penetrate the orbital septum.
- Postsurgical (e.g. retinal detachment, strabismus, lacrimal, and orbital surgery).

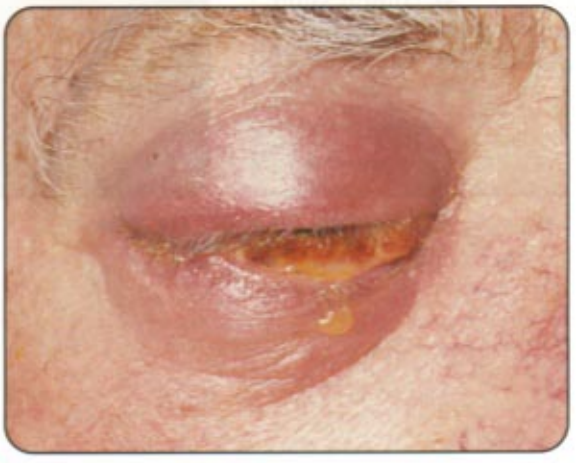


Fig. 2.39

Signs

- Unilateral painful proptosis and severe lid edema (**Fig. 2.39**) in a very unwell patient.
- Ophthalmoplegia.

Look for

- Signs of optic nerve compression.

Idiopathic orbital inflammatory syndrome (pseudotumor)

Idiopathic orbital inflammatory syndrome presents usually between the ages of 20 and 50 years. Acute orbital myositis is a type of pseudotumor which primarily affects one or more of the extraocular muscles.

Signs

- Usually unilateral axial proptosis associated with variable chemosis (**Fig. 2.40a and b**).
- Ophthalmoplegia and diplopia. In patients with acute orbital myositis, pain and diplopia are increased on attempted gaze into the field of the affected muscle.

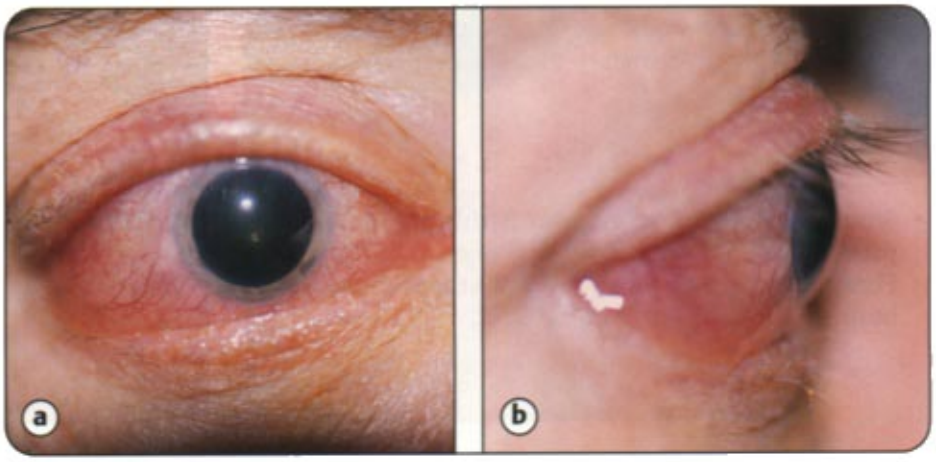


Fig. 2.40

Look for, in bilateral cases

- Wegener granulomatosis.
- Polyarteritis nodosa.
- Sarcoidosis.
- Tuberculosis.
- Waldenström macroglobulinemia.

Carotid–cavernous fistula

A carotid–cavernous fistula may be caused by either head trauma or a spontaneous rupture of an intracavernous aneurysm.

Signs

- Unilateral painful pulsatile proptosis associated with a bruit.
- Severe chemosis (**Fig. 2.41**).
- Grossly dilated epibulbar vessels may be present in the absence of chemosis (**Fig. 2.42**).
- Ophthalmoplegia.
- Retinal venous congestion which may be associated with hemorrhage (**Fig. 2.43**).

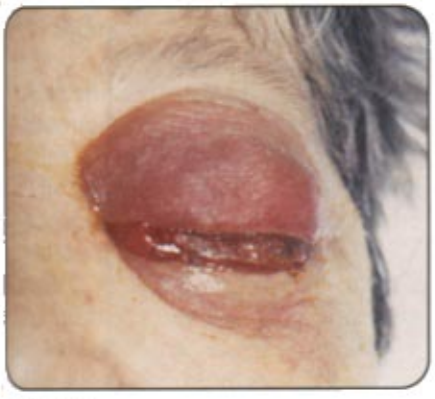


Fig. 2.41

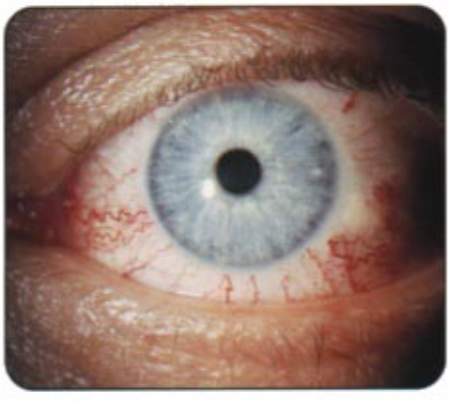


Fig. 2.42

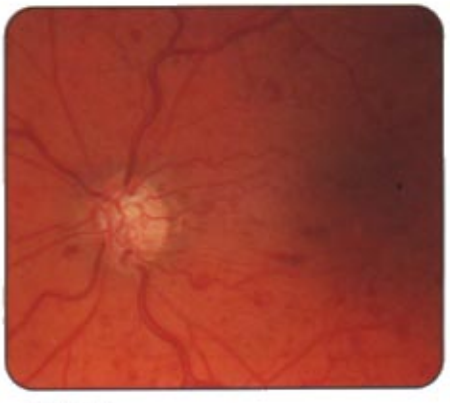


Fig. 2.43

Cavernous sinus thrombosis

Cavernous sinus thrombosis is a serious condition which is most commonly secondary to skin or paranasal sinus infection.

Signs

- Similar to a carotid–cavernous fistula except that the patient is usually more ill because of systemic infection. **Figure 2.44** shows a patient with cavernous sinus thrombosis following drainage of the orbit.

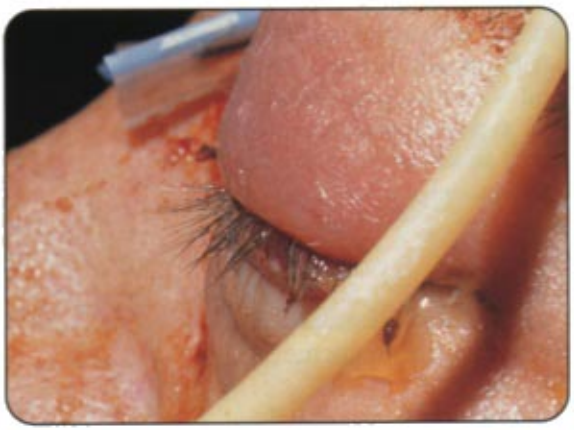


Fig. 2.44

Metastatic tumors

Presentation of metastatic tumors is usually with a rapid onset of diplopia, lid edema, and pain.

Signs

- Although most metastases from tumors cause proptosis (**Fig. 2.45**), secondaries from scirrhous breast carcinoma may give rise to enophthalmos.

Other common primary sites include the bronchus, gastrointestinal tract, and prostate.

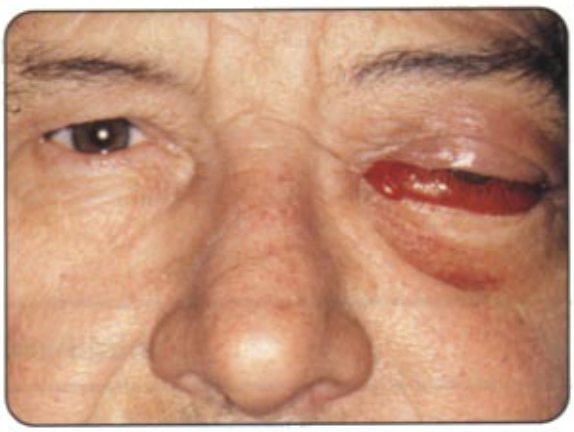


Fig. 2.45

Chronic axial proptosis

Thyroid eye disease

Thyroid eye disease may also cause unilateral or bilateral chronic axial proptosis unassociated with signs of congestion (**Fig. 2.46**).

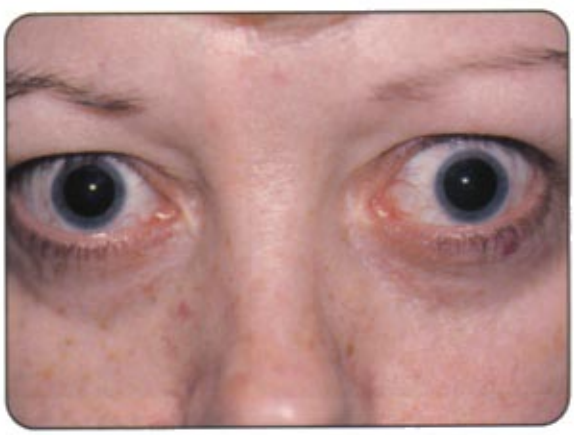


Fig. 2.46

Cavernous haemangioma

Cavernous hemangioma presents during the fourth and fifth decades of life. It is the most commonly encountered benign orbital tumor in adults.

Signs

- Unilateral, usually axial, slowly progressive proptosis which may be quite subtle (**Fig. 2.47**).
- Optic nerve compression is uncommon unless the tumor is located near the orbital apex.

Look for

- Acquired hypermetropia and chorioretinal folds.

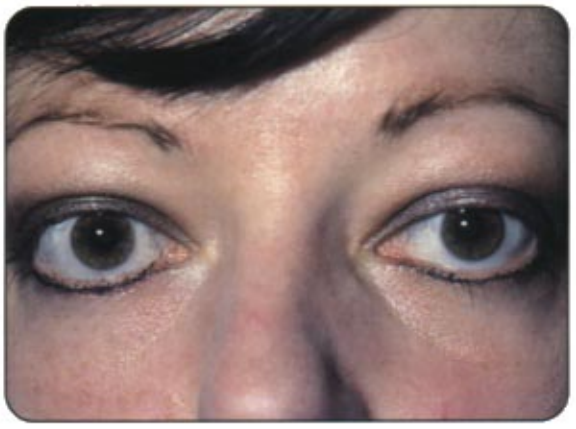


Fig. 2.47

Optic nerve sheath meningioma

Optic nerve sheath meningioma presents usually in middle age. The female to male ratio is 3:1.

Signs

- Unilateral, slowly progressive axial proptosis with early decrease in visual acuity.

Look for

- Optic atrophy and opticociliary shunts (**Fig. 2.48**).

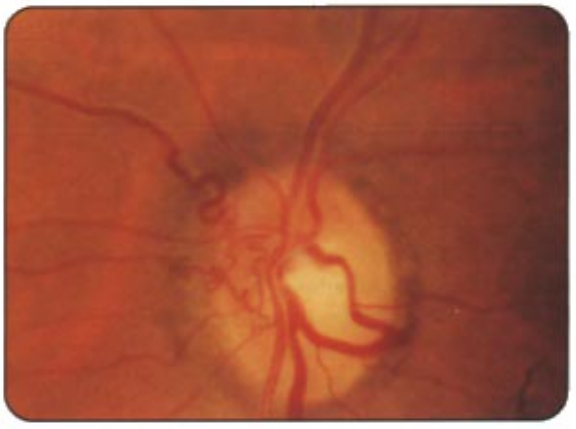


Fig. 2.48

Varix

Presentation of a varix can be at any age.

Signs

- Intermittent, nonpulsatile axial proptosis which is not associated with a bruit.
- The proptosis may be induced or accentuated by performing the Valsalva maneuver.

Figure 2.49a shows a patient with a right orbital varix before performing the Valsalva maneuver and **Figure 2.49b** shows the increase in proptosis after the Valsalva maneuver.

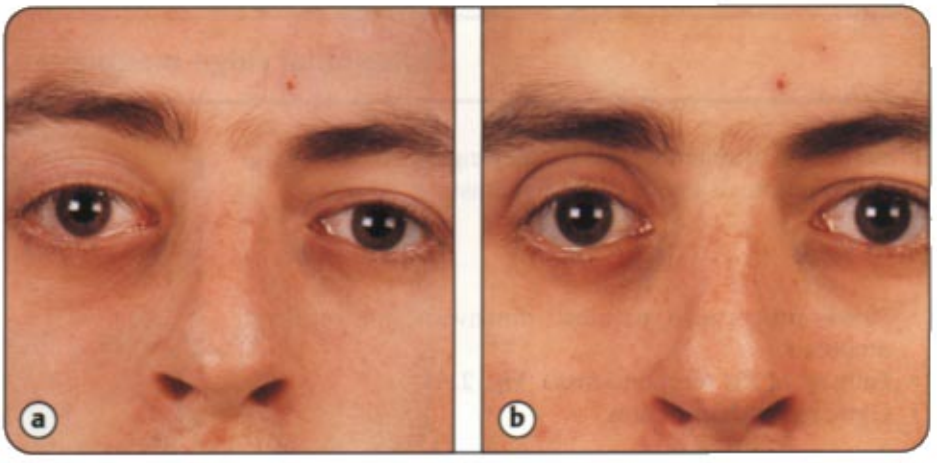


Fig. 2.49

Look for

- Vascular lesions in the eyelids (**Fig. 2.50**) and conjunctiva (**Fig. 2.51**).

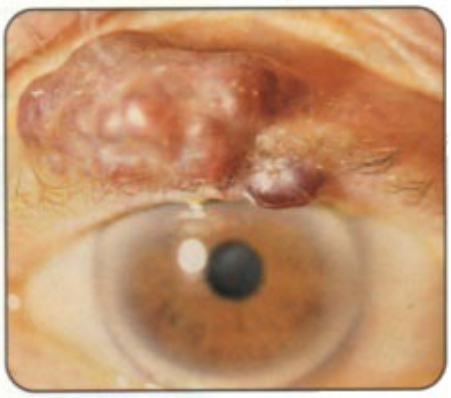


Fig. 2.50

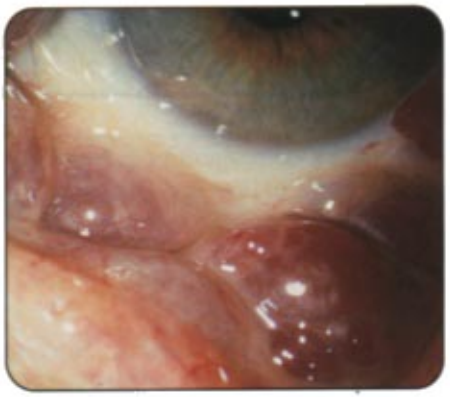


Fig. 2.51

Chronic nonaxial proptosis

Nonaxial proptosis is caused by anterior orbital disorders which displace the globe away from the lesion.

Lymphoid tumors

Presentation of lymphoid tumors is usually in old age.

Signs

- Unilateral or bilateral involvement which may be associated with periorbital puffiness (**Fig. 2.52**).
- It may be possible to palpate an anteriorly located lesion which has a rubbery consistency.
- Conjunctival extension may be present.

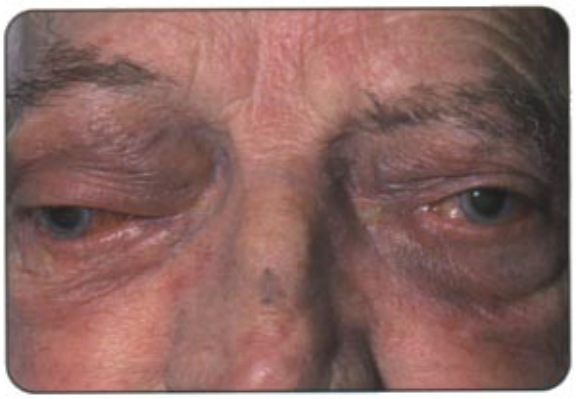


Fig. 2.52

Sphenoidal ridge meningioma

Presentation of sphenoidal ridge meningioma is in middle age with proptosis and reactive hyperostosis.

Signs

- Slowly progressive, painless, downward and outward proptosis.
- Fullness of the temporal fossa (**Fig. 2.53**).
- Optic nerve dysfunction.

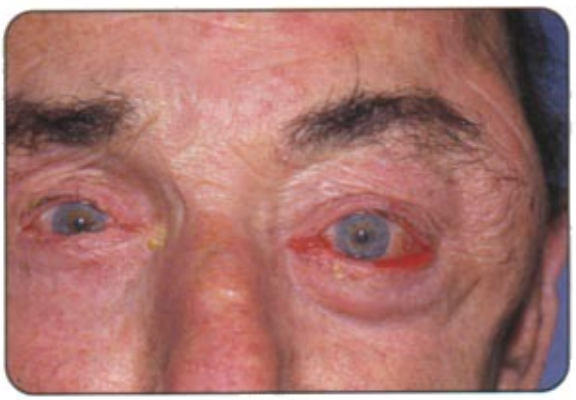


Fig. 2.53

Mucoceles

Mucoceles present with a combination of ptosis, proptosis, and globe displacement. The proptosis may fluctuate when the walls of the mucocele become inflamed.

- Frontal mucocele displaces the globe downward. It may be associated with eyelid inflammation if there is associated acute sinusitis.

Figure 2.54a–d shows a patient with a right frontal mucocele.

- Ethmoidal mucocele causes lateral displacement which may be associated with proptosis (**Fig. 2.55**).

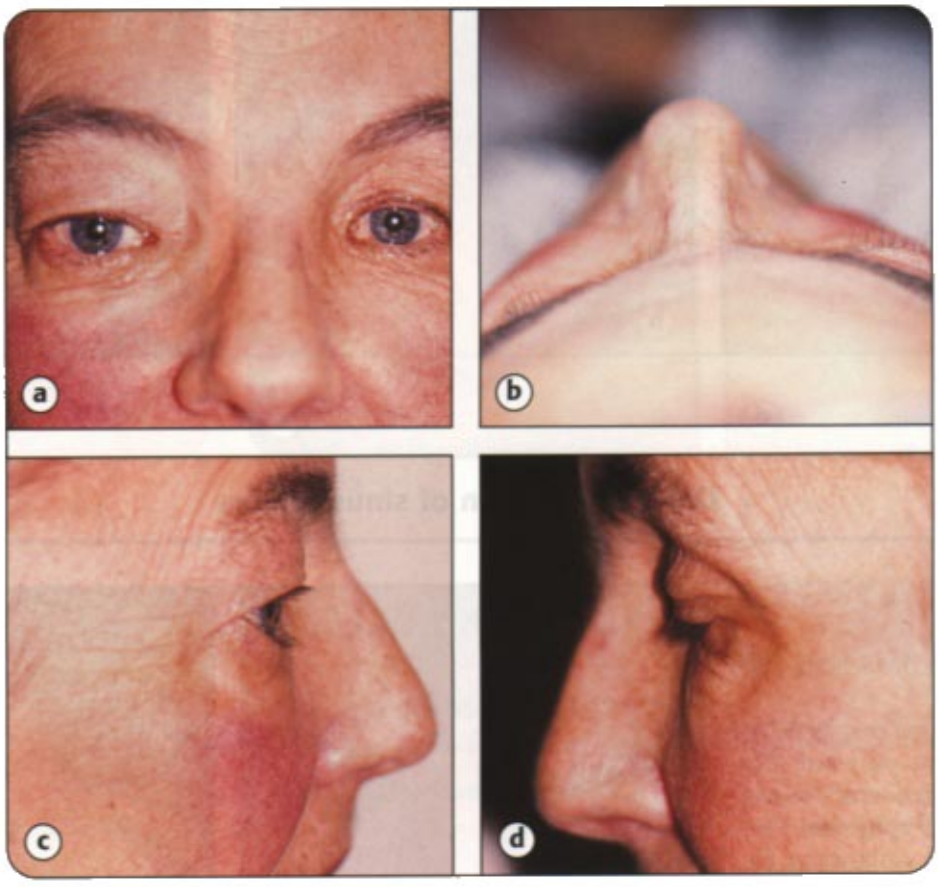


Fig. 2.54

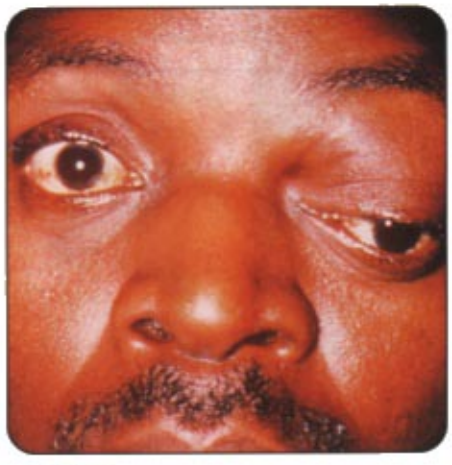


Fig. 2.55

Paget disease of the skull

Paget disease of the skull may cause slowly progressive unilateral or bilateral proptosis. Occasionally, patients with Paget disease develop osteosarcoma. **Figure 2.56a–c** shows a patient with Paget disease and an osteosarcoma of the left orbital roof.

Look for

- Optic nerve compression.

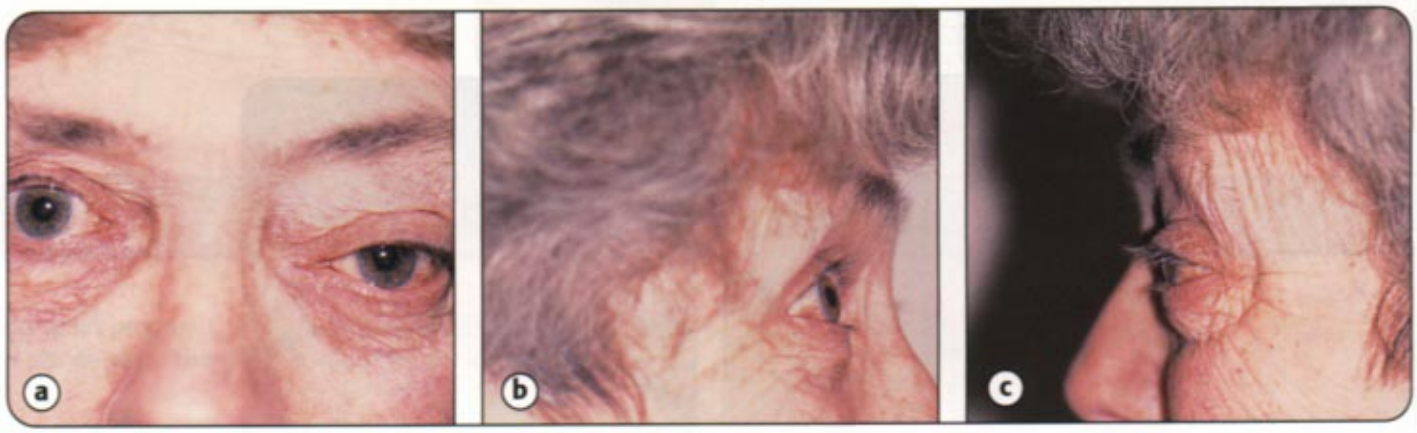


Fig. 2.56

Orbital extension of sinus tumors

Signs

The signs depend on the origin of the tumor:

- Maxillary carcinoma displaces the globe upward and outward [**Fig. 2.57a–d**] and is frequently associated with pain and epiphora.
- Ethmoidal/frontal carcinoma displaces the globe downward and outward.
- Osteosarcoma unassociated with Paget disease typically occurs in young adults. It may be associated with the inherited form of rhabdomyosarcoma with or without previous orbital irradiation.
- Ivory osteoma arises from the frontal or ethmoidal sinuses and causes a slowly progressive proptosis and displacement of the globe.

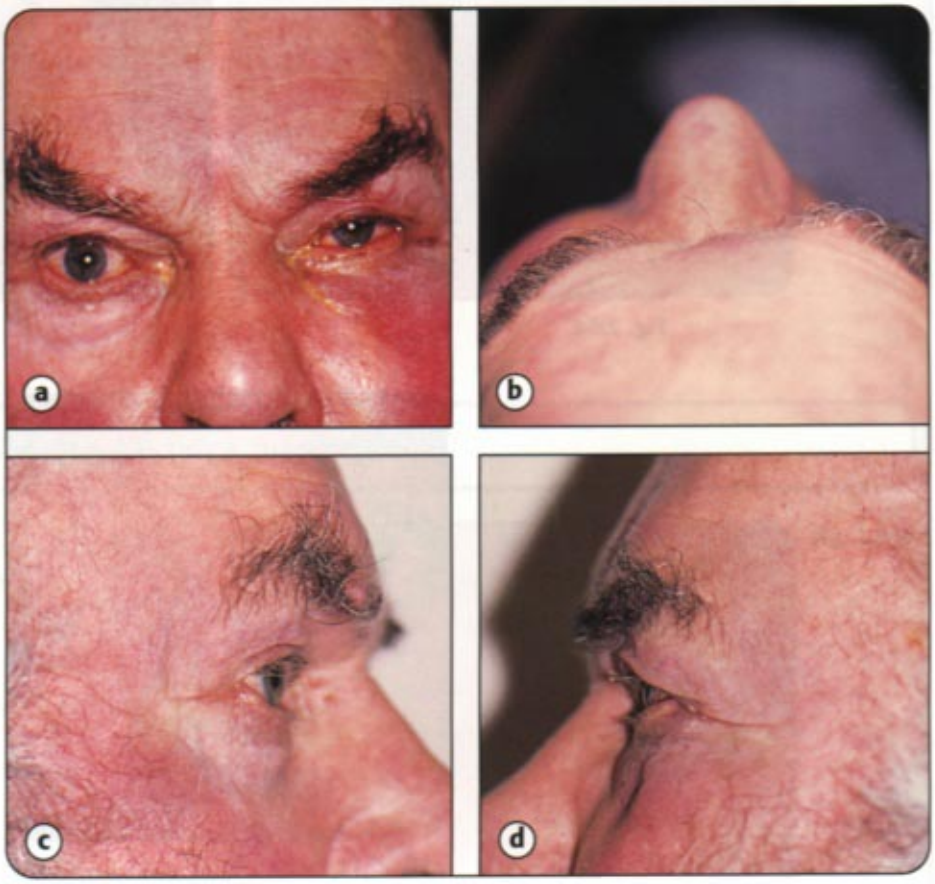


Fig. 2.57

Other causes

- Eyelid tumors such as advanced basal cell carcinoma, squamous cell carcinoma (**Fig. 2.58**), and sebaceous gland carcinoma may invade the orbit.
- Nasopharyngeal carcinoma may invade the orbit through the ethmoidal sinus and displaces the globe downward and outward (**Fig. 2.59a and b**).
- Myeloma may involve the orbit as a solitary bony deposit (plasmocytoma) or as part of generalized disease.
- Leukemic orbital involvement in adults is usually seen as part of established systemic disease.
- Lacrimal gland lesions.

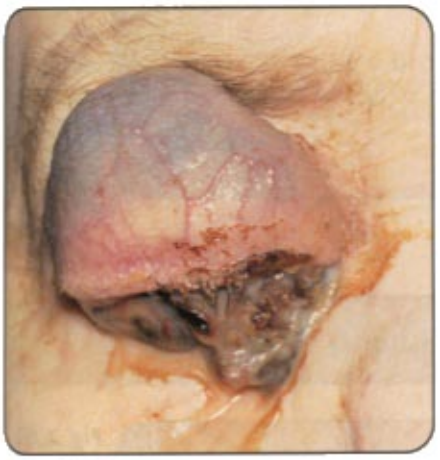


Fig. 2.58

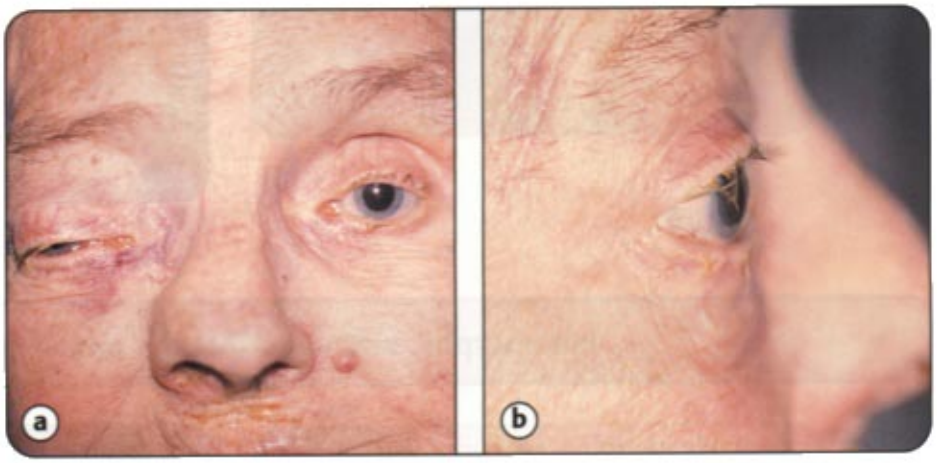


Fig. 2.59

LACRIMAL ENLARGEMENT

Unilateral enlargement

Dacryoadenitis

Dacryoadenitis is caused by infection or in association with pseudotumor.

Signs

- The gland has a gritty feeling and the upper lid has a typical 'S'-shaped curve (**Fig. 2.60**).

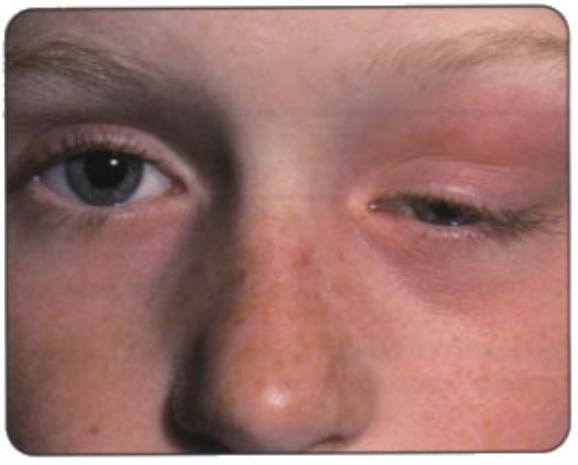


Fig. 2.60

Dacryops

Dacryops is caused by obstruction and dilatation of major lacrimal ducts.

Signs

- On lid eversion, dacryops manifests as a blue-domed cyst (**Fig. 2.61**).

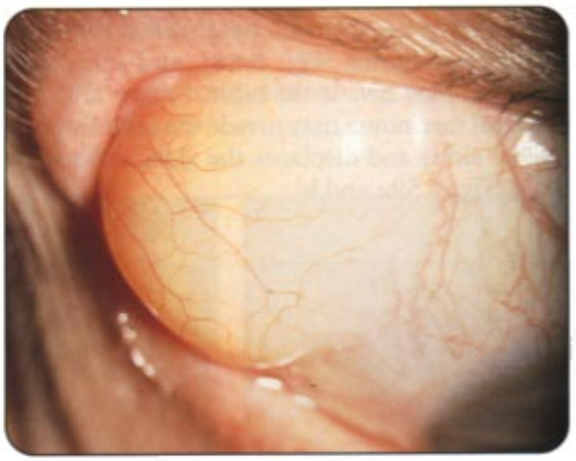


Fig. 2.61

Pleomorphic adenoma (benign mixed cell tumor)

Presentation of pleomorphic adenoma is usually in middle age.

Signs

- Chronic, painless fullness of the eyelid and displacement of the globe downward and inward (**Fig. 2.62**) (superotemporal orbital mass syndrome).
- Diplopia may occur when gaze is directed toward the lesion (i.e. upward and outward).

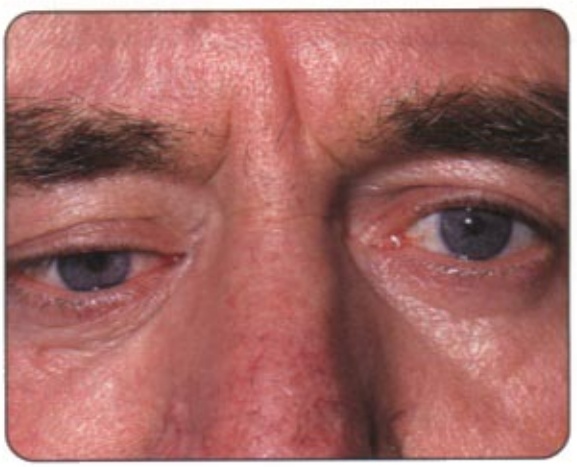


Fig. 2.62

Malignant lacrimal gland tumors

Malignant lacrimal gland tumors have a shorter history than benign tumors and are associated with pain and diplopia.

- Malignant mixed cell tumor (pleomorphic adenocarcinoma) – typically occurs in old age.
- Adenoid cystic carcinoma – typically affects women around age 40 years.
- Other carcinomas – mucoepidermoid and squamous cell.

Bilateral enlargement

Causes

- Physiological enlargement associated with shallow orbits.
- Thyroid eye disease – during the late stages.
- Sarcoidosis may cause bilateral firm enlarged lacrimal glands (**Fig. 2.63**).
- Acute leukemia – in children, a particular form of acute myeloid leukemia (chloroma) may infiltrate one or both lacrimal glands.
- Lymphomas typically cause a firm or rubbery enlargement which may be unilateral or bilateral.
- Amyloidosis.

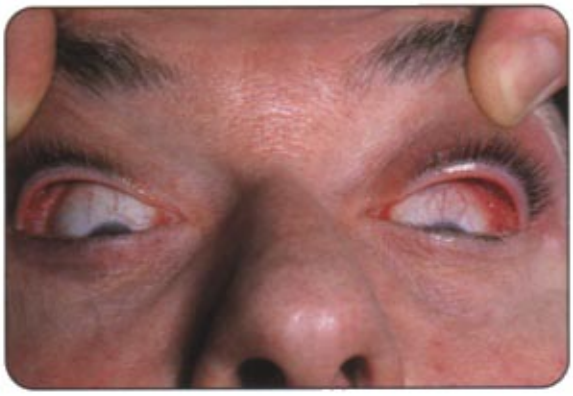


Fig. 2.63

ORBITAL RIM LESIONS

Orbital rim lesions are almost always unilateral and may occur anywhere around the orbital rim.

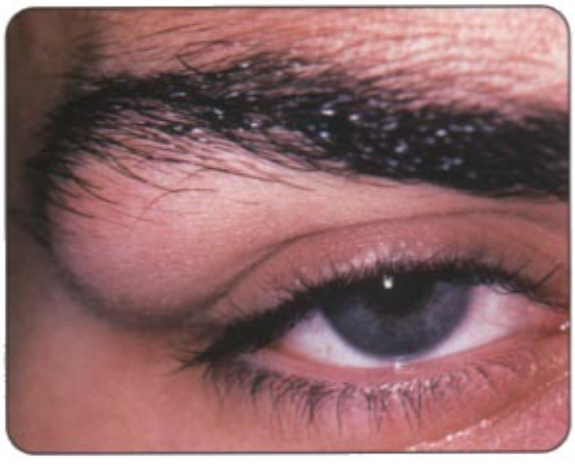


Fig. 2.64

Causes

- A superficial dermoid cyst is an asymptomatic firm round lesion, most frequently located in the upper outer quadrant of the orbital margin (**Fig. 2.64**).
- Subperiosteal hematoma.
- Subperiosteal abscess is very painful and is associated with eyelid inflammation.
- Subperiosteal cyst may be associated with proptosis.
- Necrobiotic xanthogranuloma is characterized by subcutaneous periocular indurated nodules occurring during the sixth decade of life which are associated with systemic monoclonal gammopathies and plasma cell tumors.

DYNAMIC PROPTOSIS

Intermittent proptosis

- Orbital varices cause a nonpulsatile proptosis which may be be accentuated by the Valsalva maneuver.
- Mucocele may result in proptosis which fluctuates when its walls become inflamed.
- Capillary hemangioma causes an increase in proptosis on crying.
- Lymphangioma may cause a proptosis which increases during or after an upper respiratory tract infection.

Pulsatile proptosis

Pulsatile proptosis caused by transmitted CSF pulsation is not associated with a bruit. The two main causes are:

- Encephalocele in which the pulsation is transmitted to the orbit through a defect in the sphenoid bone.
- Fracture of the orbital roof.

Pulsatile proptosis caused by arterial pulsation may be associated with a bruit. The two main causes are:

- Carotid–cavernous fistula.
- Congenital arteriovenous communication which may occur in isolation or as part of Wyburn–Mason syndrome.

3 The conjunctiva

ACUTE AND SUBACUTE CONJUNCTIVITIS 68

Bacterial conjunctivitis
Allergic conjunctivitis
Viral conjunctivitis
Adult chlamydial conjunctivitis
Conjunctivitis in Reiter syndrome

CHRONIC CONJUNCTIVITIS 71

Chronic papillary conjunctivitis
Vernal keratoconjunctivitis
Atopic keratoconjunctivitis
Giant papillary conjunctivitis
Superior limbic keratoconjunctivitis
Chronic follicular conjunctivitis
Chronic viral conjunctivitis
Chronic toxic conjunctivitis
Pernaud oculoglandular syndrome

NEONATAL CONJUNCTIVITIS 76

CICATRIZING CONJUNCTIVITIS 77

Trachoma
Mucocutaneous disorders
Stevens–Johnson syndrome
Cicatricial pemphigoid
Toxic epidermal necrolysis (Lyell disease)
Epidermolysis bullosa
Pemphigus vulgaris
Linear IgA bullous dermatosis
Cutaneous disorders
Atopic dermatitis
Xeroderma pigmentosum
Scleroderma
Toxic causes

SECONDARY CHRONIC 'CONJUNCTIVITIS' 82

Chronic blepharitis
Keratoconjunctivitis sicca
Rosacea
Chronic canaliculitis
Chronic dacryocystitis
Floppy eyelid syndrome
Mucus fishing syndrome
Lid inbrication syndrome
Masquerade syndrome

PIGMENTED LESIONS 85

Flat superficial pigmentation
Common, small, congenital focal lesions
Benign epithelial (racial) melanosis
Primary acquired melanosis
Secondary acquired melanosis
Flat deep pigmentation
Miscellaneous flat discoloration
Red
Other types of flat discoloration
Pigmented nodules
Nevus
Melanocytoma
Primary melanoma
Melanoma developing in primary acquired melanosis

NONPIGMENTED LESIONS 91

Small nodules
Granulomas
Pinguecula
Phlycten
Large nodules
Pseudoepitheliomatous hyperplasia
Epibulbar dermoid
Nonpigmented melanoma
Choristomas
Plaque-like lesions
Pterygium
Bitot spot
Conjunctival intraepithelial neoplasia
Invasive squamous cell carcinoma
Other carcinomas
Diffusely elevated lesions
Lipodermoid
Non-Hodgkin lymphoma
Reactive lymphoid hyperplasia
Papillary lesions
Pedunculated squamous papilloma
Sessile squamous papilloma
Other papillary lesions
Cystic lesions
Primary conjunctival cyst
Cyst of gland of Wolfring
Iatrogenic cysts

VASCULAR LESIONS 100

Telangiectasias
Metabolic disorders
Hematological disorders
Hemorrhagic lymphangiectasia
Sturge–Weber syndrome (encephalotrigeminal angiomatosis)
Louis-Bar syndrome (ataxia telangiectasia)
Rendu–Osler–Weber disease (hereditary hemorrhagic telangiectasia)
Vascular tumors
Capillary hemangioma
Lymphangioma
Kaposi sarcoma

ACUTE AND SUBACUTE CONJUNCTIVITIS

Bacterial conjunctivitis

Bacterial conjunctivitis is a very common, usually bilateral condition.

Most frequent causative organisms

- *Staphylococcus aureus.*
- *Streptococcus pneumoniae.*
- *Haemophilus influenzae.*

Signs

- Purulent or mucopurulent discharge.
- Conjunctival hyperemia, maximal in the fornices (**Fig. 3.1**).
- Pseudomembranes (**Fig. 3.2**) may be present in infections caused by β-hemolytic streptococci, gonococcus, and *Corynebacterium diphtheriae*.
- Conjunctival membranes consist of coagulated exudate adherent to inflamed conjunctival epithelium. Clinically, a true membrane causes bleeding on attempted removal and a pseudomembrane does not, but this rule is not infallible. The causes of true membranes and pseudomembranes are essential similar.

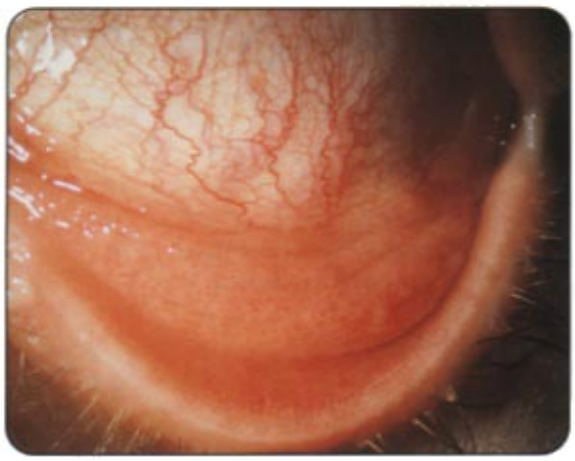


Fig. 3.1

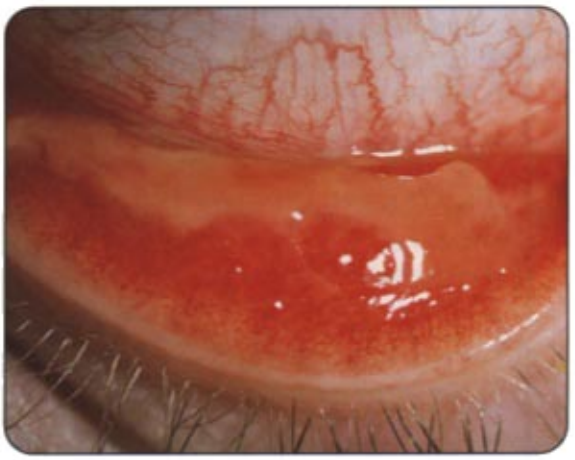


Fig. 3.2

Allergic conjunctivitis

Allergic conjunctivitis is a common, bilateral condition which may be seasonal (hay fever) or perennial.

Signs

- Lid edema.
- Watery discharge.
- Conjunctival hyperemia with a mild papillary response (**Fig. 3.3**).
- Chemosis (**Fig. 3.4**).

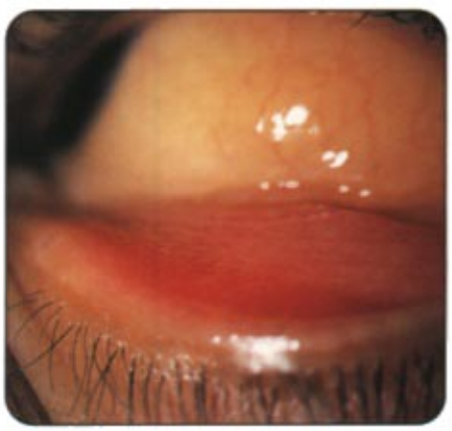


Fig. 3.3

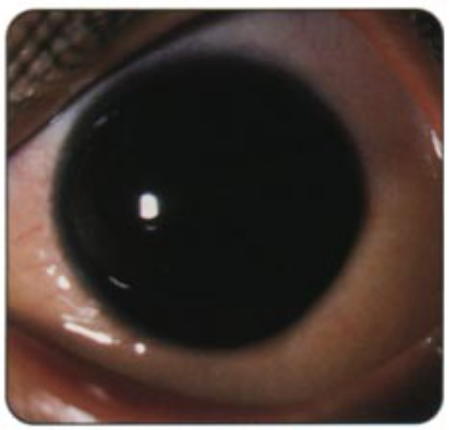


Fig. 3.4

Viral conjunctivitis

Viral conjunctivitis is a common, very contagious, usually bilateral condition.

Causative viruses

- Adenoviruses.
- Herpes simplex.
- Enteroviruses.
- Newcastle disease virus.
- Epstein–Barr virus.

Signs of adenoviral conjunctivitis

- Lid edema.
- Watery discharge.
- Generalized conjunctival hyperemia (**Fig. 3.5**).
- Conjunctival follicles, which are frequently most apparent in the inferior fornices (**Fig. 3.6**).
- Subconjunctival hemorrhages (**Fig. 3.7**).
- Pseudomembranes in severe cases (**Fig. 3.8**).
- Central punctate epithelial keratitis (**Fig. 3.9**).
- Severe preauricular lymphadenopathy may be present.

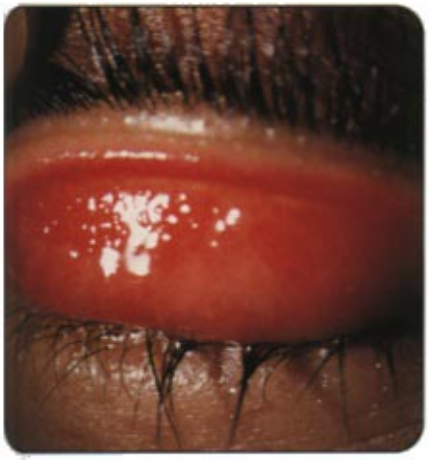


Fig. 3.5

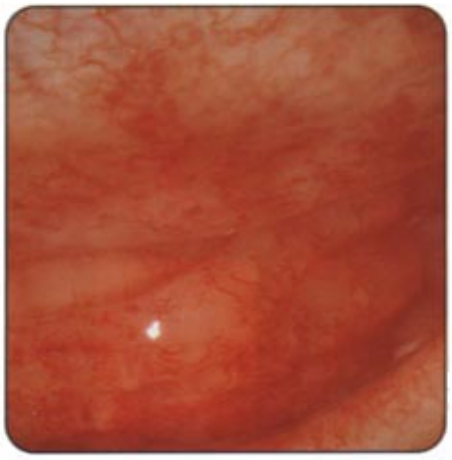


Fig. 3.6

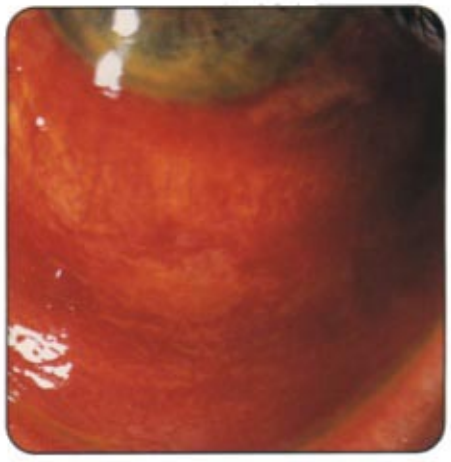


Fig. 3.7

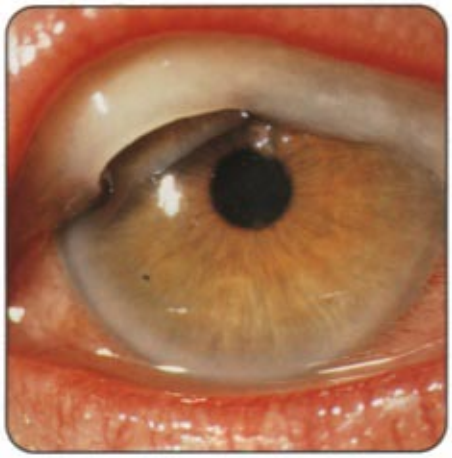


Fig. 3.8

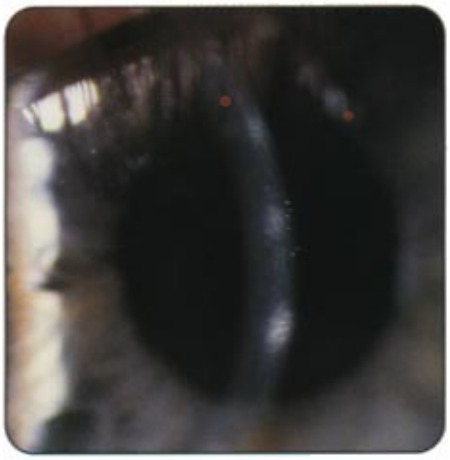


Fig. 3.9

Adult chlamydial conjunctivitis

Adult chlamydial conjunctivitis is a common, usually unilateral condition caused by *Chlamydia trachomatis* serotypes D–K.

Signs

- Mucopurulent discharge.
- Large follicles in the inferior fornices (**Fig. 3.10**).
- Superior tarsal follicles (**Fig. 3.11**).
- Occasionally, follicles at the limbus.
- Superior limbal infiltrates (**Fig. 3.12**).
- Mild preauricular lymphadenopathy may be present.

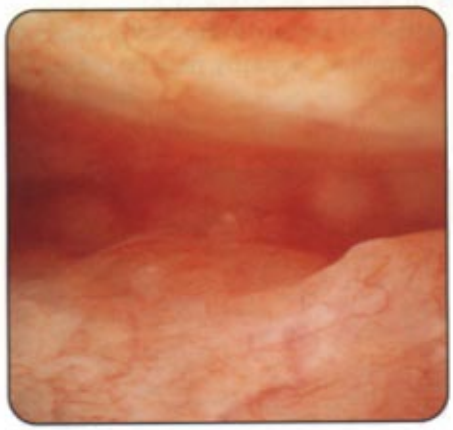


Fig. 3.10

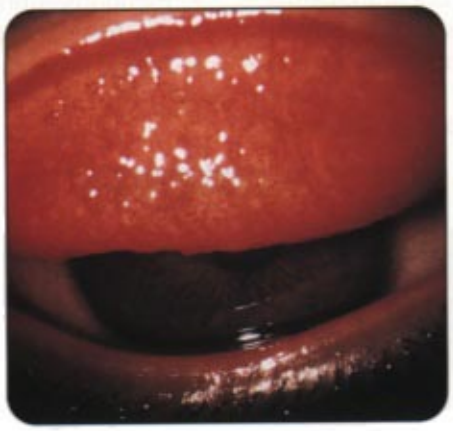


Fig. 3.11

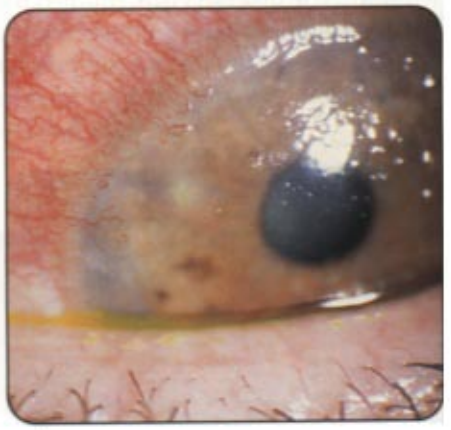


Fig. 3.12

Conjunctivitis in Reiter syndrome

Reiter syndrome is an uncommon condition which typically affects young men.

Clinical triad

- Acute, transient, bilateral, papillary mucopurulent conjunctivitis (**Fig. 3.13**).
- Nonspecific urethritis.
- Acute 'seronegative' arthritis which typically affects the lower limbs.

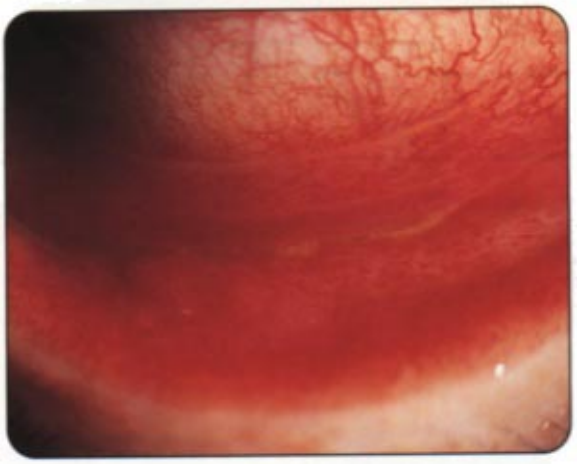


Fig. 3.13

Look for

- Acute iritis.
- Punctate subepithelial keratitis.

CHRONIC CONJUNCTIVITIS

Chronic papillary conjunctivitis

Papillae consist of hyperemic, flat-topped elevations separated by pale outlines which occur on the palpebral tarsal conjunctiva and occasionally on the perilimbal conjunctiva. The clinical appearance of papillae varies greatly according to size and number and they can be arbitrarily graded from 0 to 4.

- Grade 0 is a normal tarsal conjunctiva (**Fig. 3.14**).
- Grade 1 consists of multiple small papillae which give rise to a smooth velvety appearance (**Fig. 3.15**).
- Grade 2 consists of macropapillae, each with a diameter of 0.3–1.0 mm (**Fig. 3.16**).
- Grade 3 consists of giant papillae, each with a diameter of over 1 mm (**Fig. 3.17**).
- Grade 4 consists of enormous protruding papillae (**Fig. 3.18**).

It should be emphasized that the presence of papillae along the medial aspect of the upper tarsal plate is a normal finding.

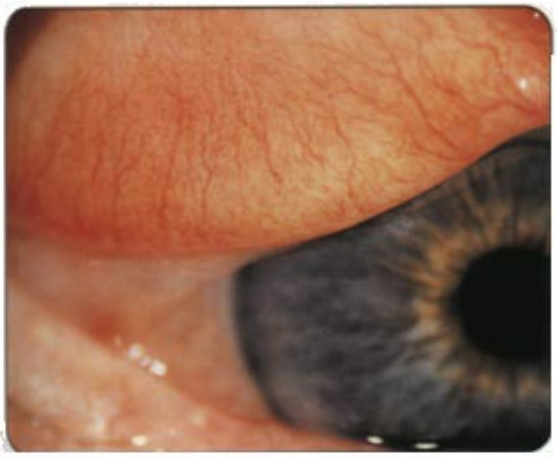


Fig. 3.14

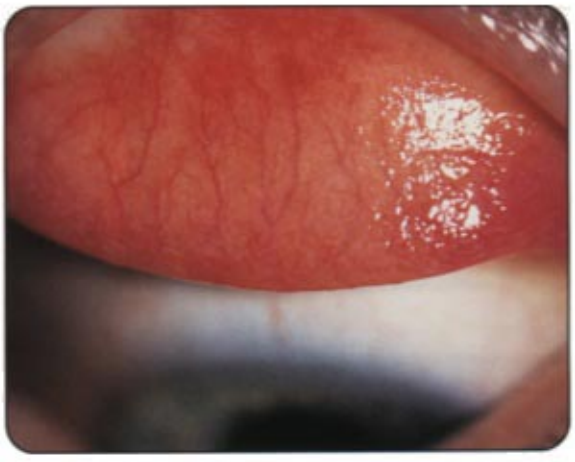


Fig. 3.15

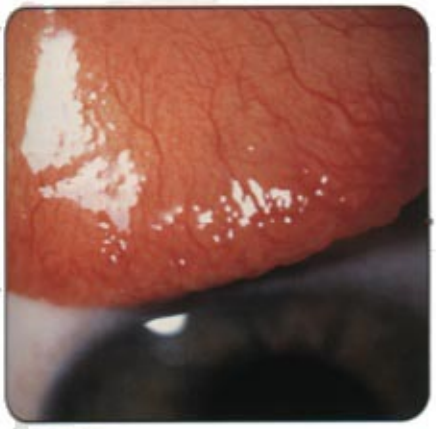


Fig. 3.16

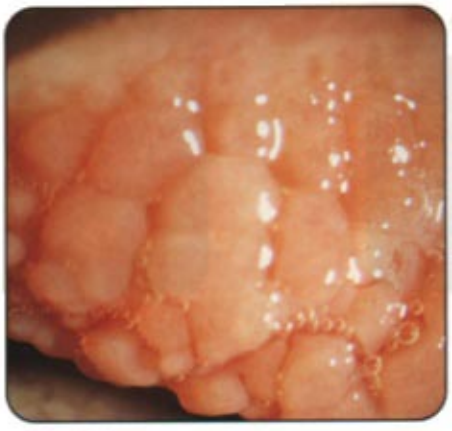


Fig. 3.17

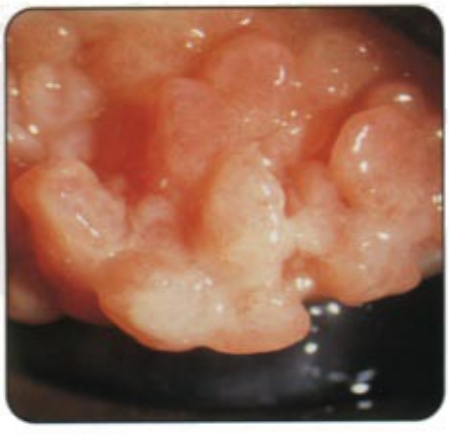


Fig. 3.18

Vernal keratoconjunctivitis

Vernal keratoconjunctivitis is a common, recurrent, bilateral condition which affects children and young adults.

Signs

- Mucoid discharge.
- Milky-white pseudomembranes.
- Superior tarsal papillae of medium to giant size (*see* **Figs 3.16–3.18**).
- Occasionally, limbal papillae (limbitis) (**Fig. 3.19**) which may be associated with small white (Trantas) dots (**Fig. 3.20**).

Complications

- Superior corneal punctate epithelial erosions.
- Micropannus (**Fig. 3.21**).
- Corneal ulceration and scarring (**Fig. 3.22**).

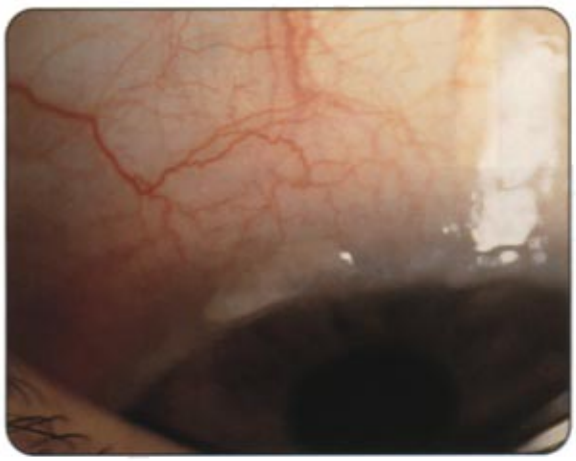


Fig. 3.19

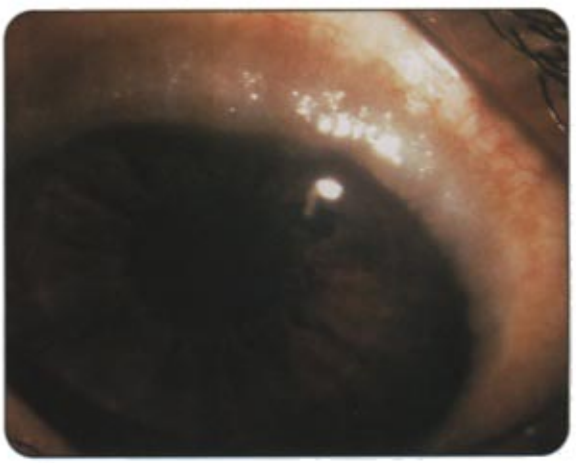


Fig. 3.20

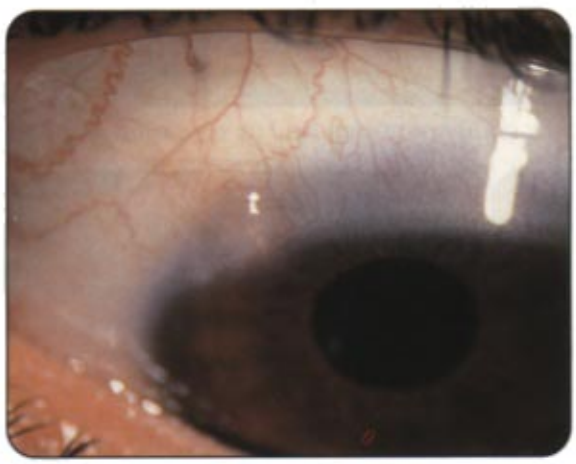


Fig. 3.21

Fig. 3.22

Atopic keratoconjunctivitis

Atopic keratoconjunctivitis is an uncommon, bilateral condition which occurs in adults with atopic dermatitis.

Signs

- Lid eczema and staphylococcal blepharitis.
- Mucoid discharge.
- Small papillae most intense on the inferior palpebral conjunctiva (**Figs 3.23, 3.24**).

Complications

- Forniceal conjunctival scarring and shortening (**Figs 3.25, 3.26**).
- Symblepharon formation (**Fig. 3.27**) in advanced cases.
- Corneal punctate epithelial erosions.
- Corneal vascularization (**Fig. 3.28**).
- Marginal corneal ulceration.

Differs from vernal conjunctivitis because

- Presents in adult life.
- Papillae are small and inferior.
- Absence of limbitis and Trantas dots.
- May cause cicatrization.

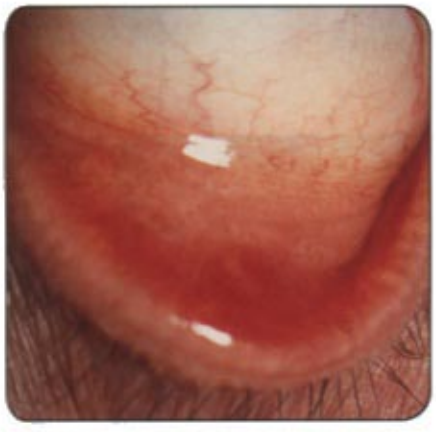


Fig. 3.23

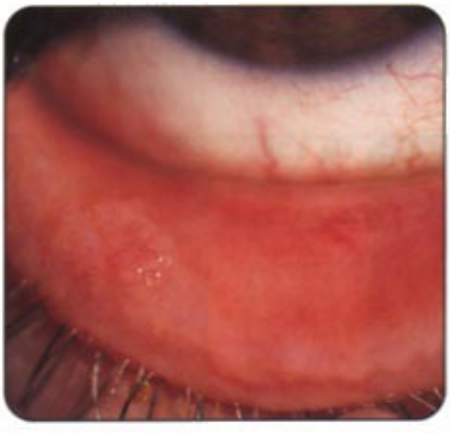


Fig. 3.24

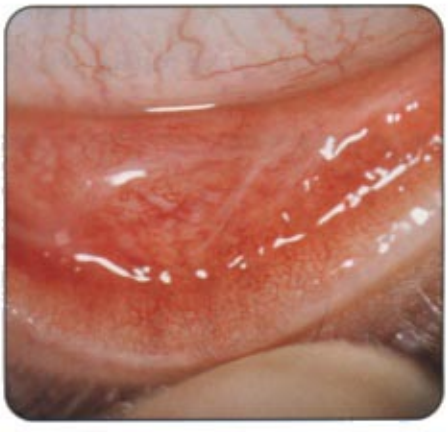


Fig. 3.25

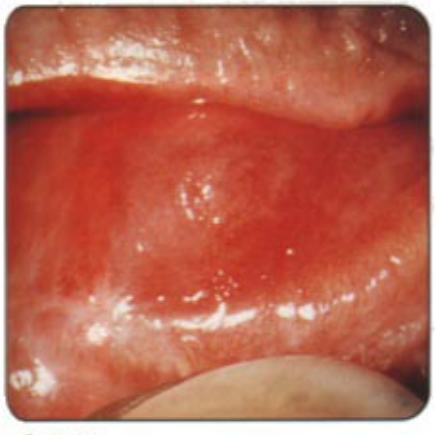


Fig. 3.26

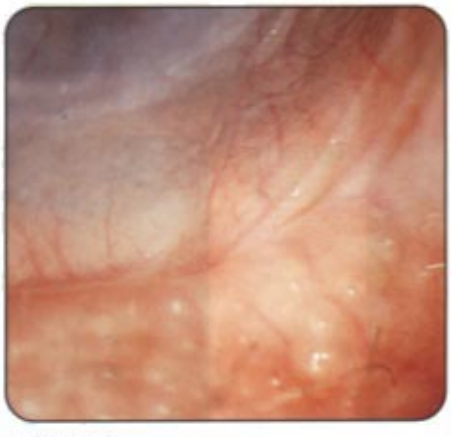


Fig. 3.27

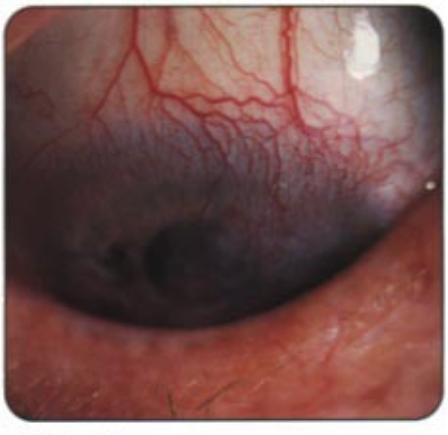


Fig. 3.28

Giant papillary conjunctivitis

Giant papillary conjunctivitis is a common condition characterized by small–medium superior tarsal papillae (*see* **Figs 3.15, 3.16**). It can be unilateral or bilateral, depending on the cause.

Causes

- Inappropriate contact lens wear, particularly soft.
- Ocular prosthetics.
- Protruding sutures following surgery.

Superior limbic keratoconjunctivitis

Superior limbic keratoconjunctivitis is an uncommon, usually bilateral condition which is frequently associated with thyroid dysfunction.

Signs

- Fine superior tarsal papillae (**Fig. 3.29**).
- Hyperemia and thickening of superior bulbar conjunctiva (**Fig. 3.30**).
- Staining of superior bulbar conjunctiva with rose bengal (**Fig. 3.31**).
- Superior corneal filaments (**Fig. 3.32**).

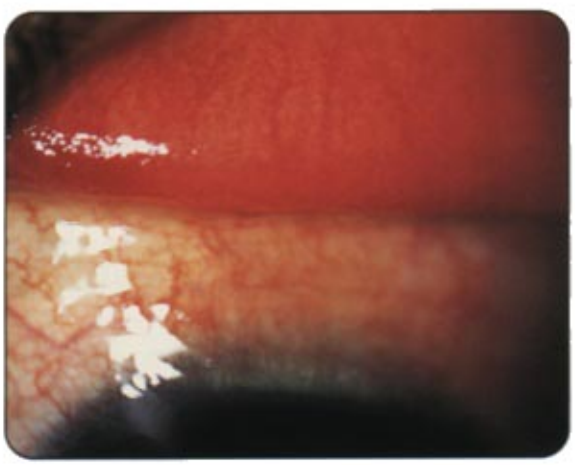


Fig. 3.29

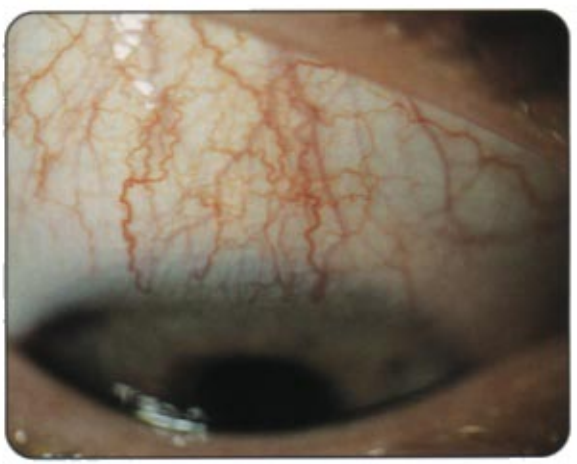


Fig. 3.30

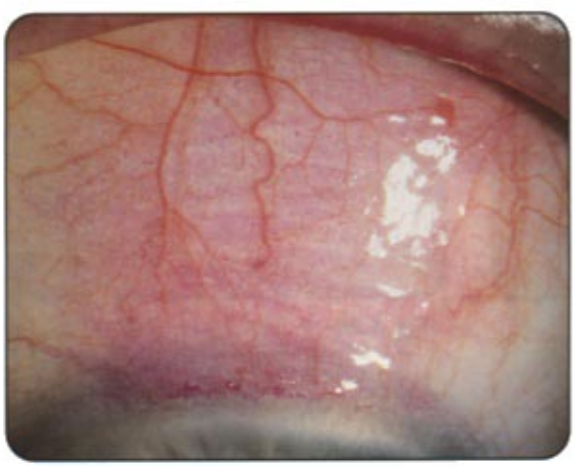


Fig. 3.31

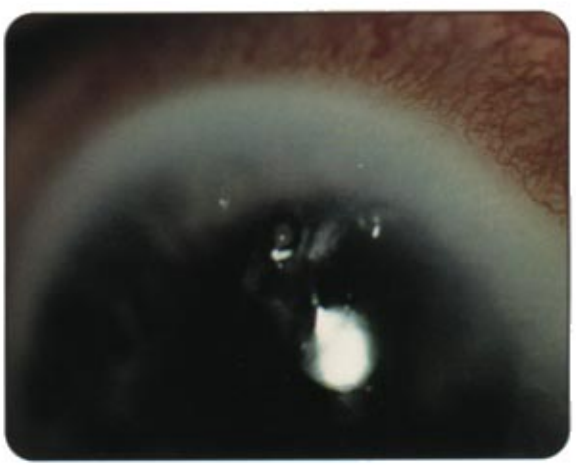


Fig. 3.32

Chronic follicular conjunctivitis

Chronic viral conjunctivitis

Chronic viral conjunctivitis is an uncommon, usually unilateral condition which may be missed if the lid margin is not examined. It is caused by the molluscum contagiosum virus.

Signs

- Single or multiple umbilicated molluscum eyelid lesions associated with ipsilateral follicular conjunctivitis (**Fig. 3.33**).
- Mucoid discharge.
- Corneal micropannus if longstanding (**Fig. 3.34**).

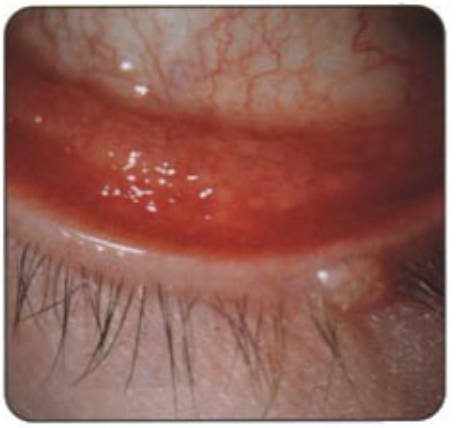


Fig. 3.33

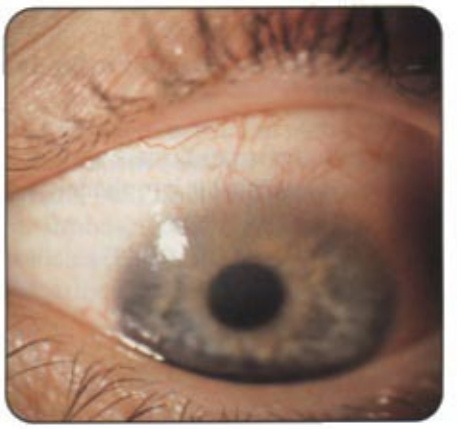


Fig. 3.34

Chronic toxic conjunctivitis

Chronic toxic conjunctivitis may be unilateral or bilateral, depending on the cause. Although common, it may be difficult to diagnose, because the patient may be on multiple topical medications.

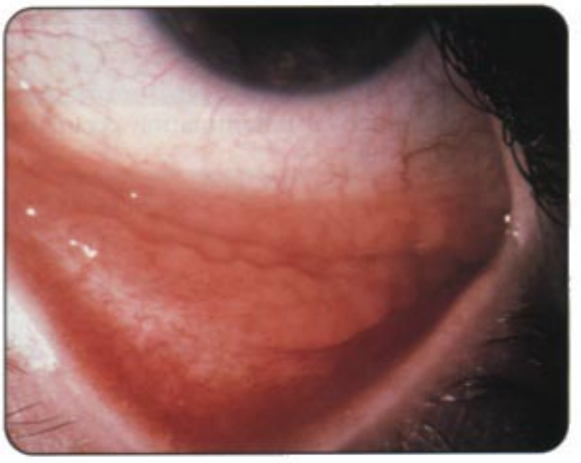


Fig. 3.35

Signs

- Initially papillary reaction, later followed by formation of follicles predominantly involving the inferior fornices (**Fig. 3.35**).
- Inferior punctate epitheliopathy.

Causes

- Aminoglycoside antibiotics.
- Antivirals.
- Glaucoma agents – particularly epinephrine.
- Preservatives.
- Nonproprietary agents (murine).
- Eye makeup.

Parinaud oculoglandular syndrome

Parinaud oculoglandular syndrome is a very rare, usually unilateral condition with many diverse causes which is usually easy to diagnose.

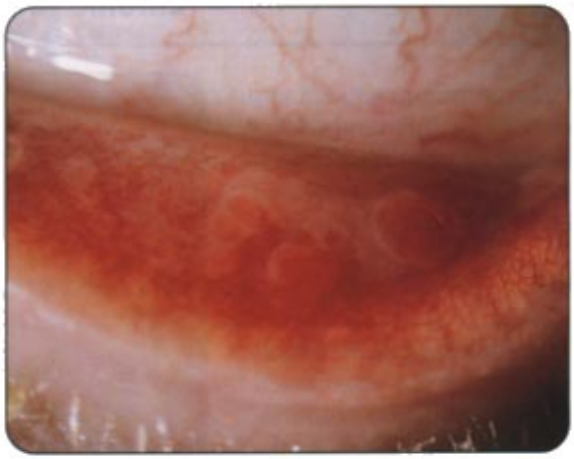


Fig. 3.36

Signs

- Conjunctival granulomas and large follicles (**Fig. 3.36**).
- Severe preauricular lymphadenopathy.

Causes

- Cat-scratch fever.
- Tularemia.
- Sporotrichosis.
- Tuberculosis.
- Syphilis.
- Coccidioidomycosis.
- Chancroid.
- Lymphogranuloma venereum.

NEONATAL CONJUNCTIVITIS

Neonatal conjunctivitis (ophthalmia neonatorum) (**Fig. 3.37**) is defined as conjunctival inflammation occurring during the first month of life.

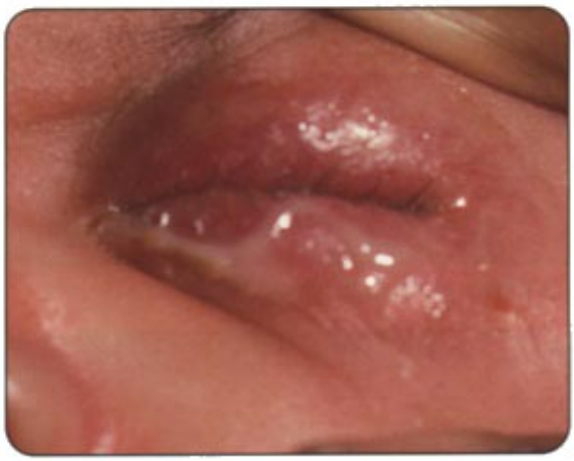


Fig. 3.37

Causes

- Chemical – usually causes relatively mild diffuse injection without discharge.
- Gonococcal – causes copious purulent discharge which may be associated with membrane formations.
- Herpes simplex type 2 – associated with lid vesicles.
- Chlamydial – causes papillary conjunctivitis because the infant cannot produce follicles.
- Simple bacterial.

CICATRIZING CONJUNCTIVITIS

Trachoma

Trachoma is a bilateral conjunctivitis which is common in underdeveloped countries. It is caused by *Chlamydia trachomatis* serotypes A, B, Ba, and C.

Signs (in chronological order)

- Follicles on superior tarsus later associated with papillae.
- Chronic infection of the superior tarsal conjunctiva (**Fig. 3.38**), leading to scarring (Arlt line) (**Fig. 3.39**).
- Herpert pits at the limbus, consisting of cicatricial remains of limbal follicles (**Fig. 3.40**).
- Trichiasis and keratopathy (**Fig. 3.41**).

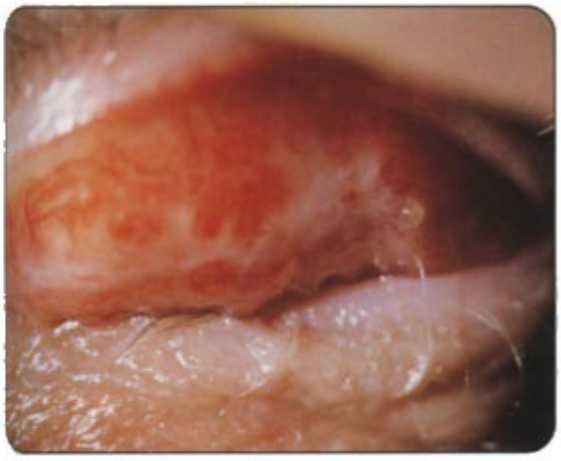


Fig. 3.38

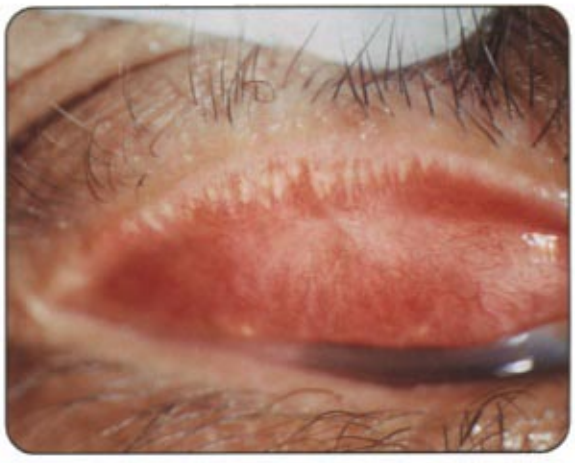


Fig. 3.39

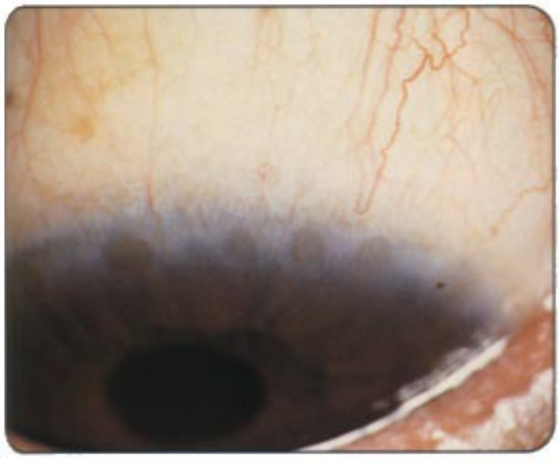


Fig. 3.40

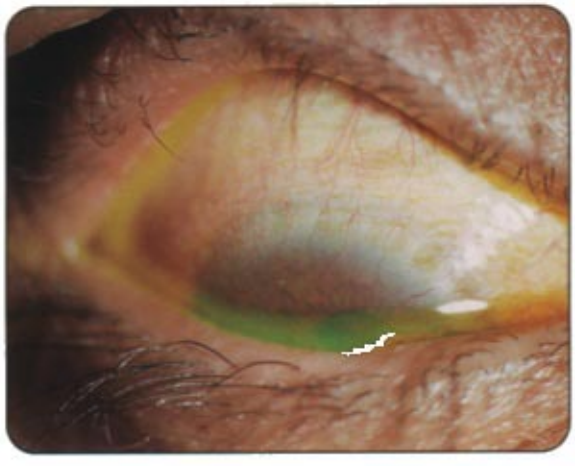


Fig. 3.41

Mucocutaneous disorders

Stevens–Johnson syndrome

Stevens–Johnson syndrome (erythema multiforme major) is an uncommon, mucocutaneous disease which is most frequently triggered by drugs or infections. The associated conjunctivitis is usually self-limiting, and secondary cicatrization may occur but is uncommon.

Signs

- Bilateral generalized conjunctivitis (**Fig. 3.42**) which may be associated with pseudomembranes.
- Oral mucous membrane lesions (**Fig. 3.43**).
- Hemorrhagic crusting of lips (**Fig. 3.44**).
- Skin lesions (**Fig. 3.45**).

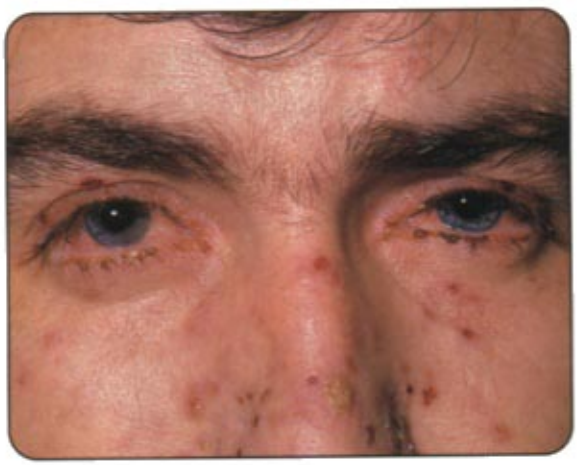


Fig. 3.42

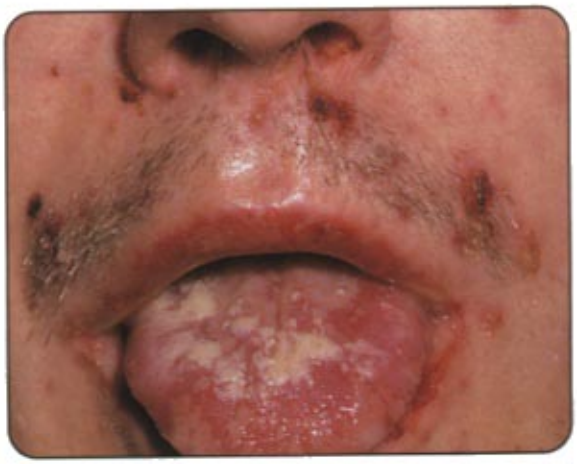


Fig. 3.43

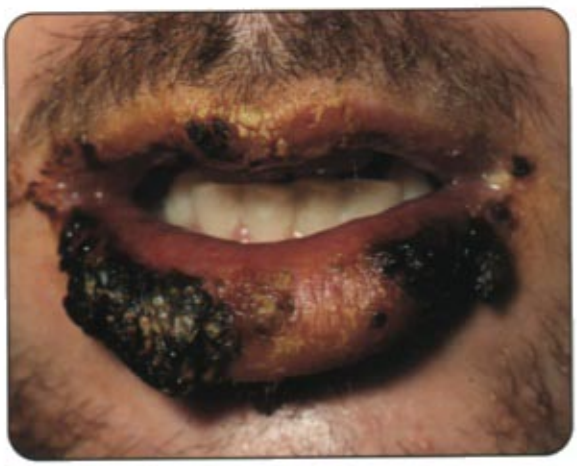


Fig. 3.44

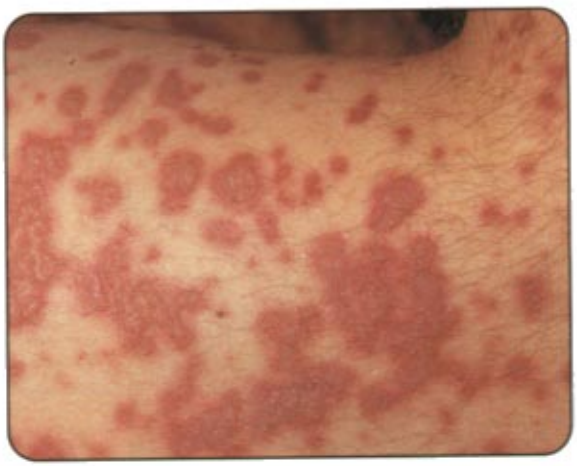


Fig. 3.45

Cicatricial pemphigoid

Cicatricial pemphigoid is an uncommon, mucocutaneous disorder which primarily affects elderly patients. About 90% of patients have lesions of the oral mucosa (**Fig. 3.46**), but skin lesions are less common. Conjunctival involvement occurs in the majority of cases, and in some it may be the only manifestation of the disease.

Signs

- Conjunctival inflammation with fine subepithelial fibrosis.
- Conjunctival shrinkage with shortening of the inferior fornix (**Fig. 3.47**).
- Loss of outline of the plica semilunaris (**Fig. 3.48**).
- Progressive symblepharon formation (**Figs 3.49, 3.50**).
- Corneal keratinization and fusion between the upper and lower lids at the outer canthus (ankyloblepharon) (**Fig. 3.51**).
- Rubbing on the cornea by trichiasis and by new lashes emerging from meibomian gland orifices (metaplastic lashes) (**Fig. 3.52**).
- Corneal ulceration and vascularization which may be complicated by infection (**Fig. 3.53**), associated with persistent epithelial defects, exposure, and drying.

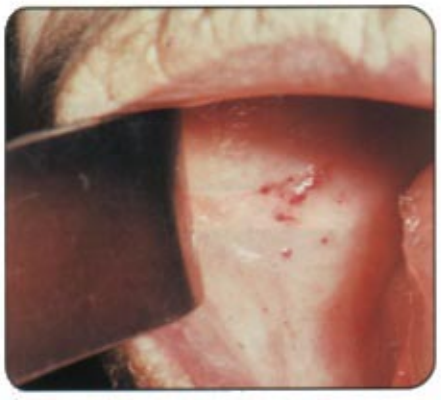


Fig. 3.46

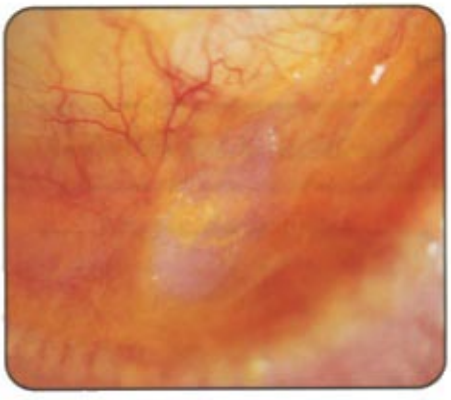


Fig. 3.47

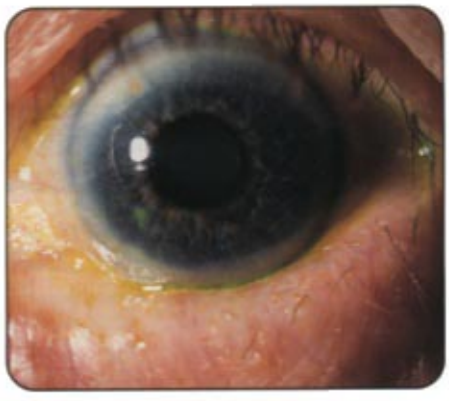


Fig. 3.48

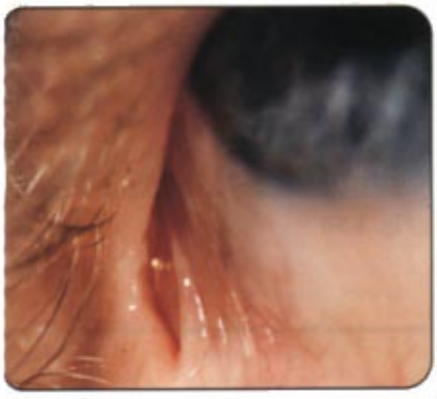


Fig. 3.49

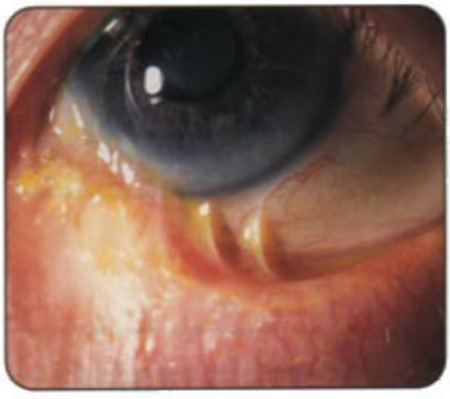


Fig. 3.50

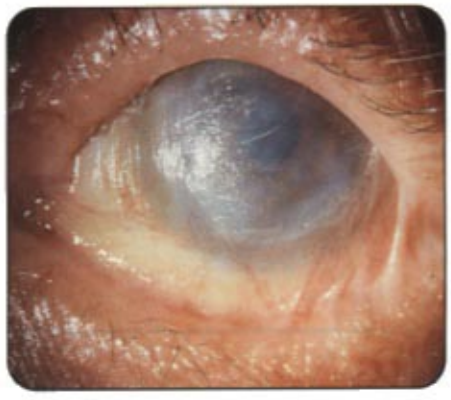


Fig. 3.51

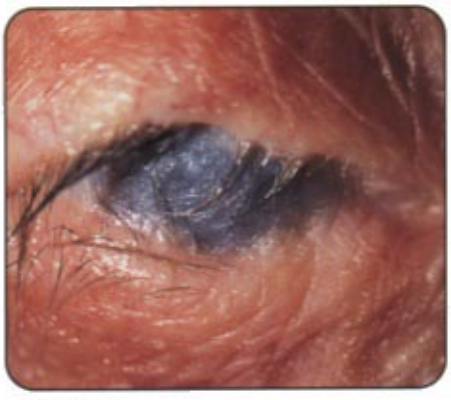


Fig. 3.52

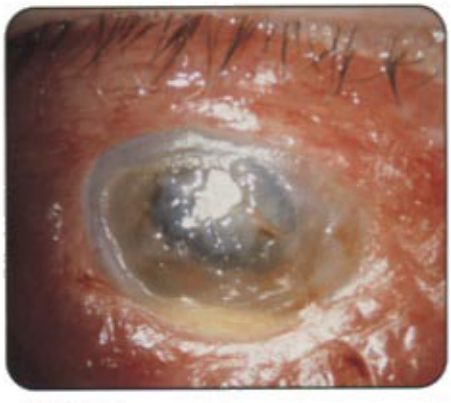


Fig. 3.53

Toxic epidermal necrolysis (Lyell disease)

Toxic epidermal necrolysis is characterized by skin lesions resembling 'scalded skin' (**Fig. 3.54**). Conjunctivitis is similar to, but milder than, that in Stevens–Johnson syndrome.

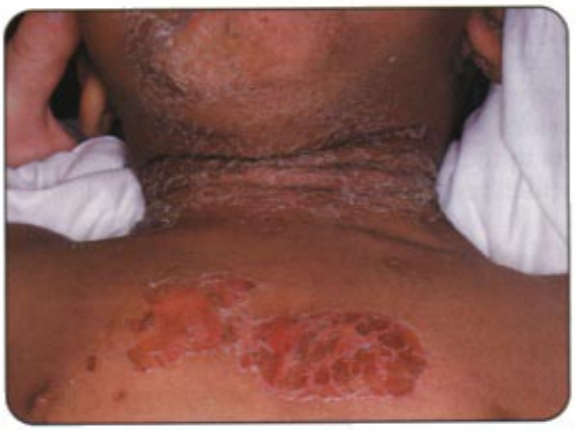


Fig. 3.54

Epidermolysis bullosa

Epidermolysis bullosa is characterized by bullous skin lesions induced by minor trauma (**Fig. 3.55**). Dystrophic nail lesions also occur (**Fig. 3.56**). Conjunctivitis is common and may result in scarring.

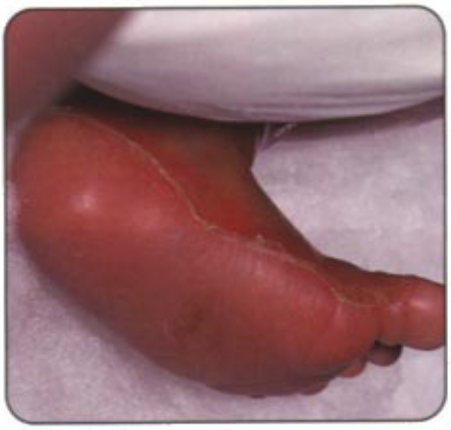


Fig. 3.55

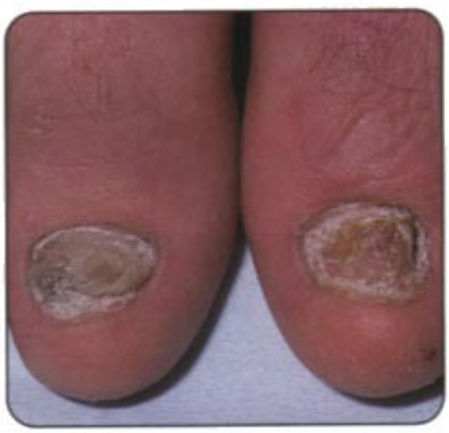


Fig. 3.56

Pemphigus vulgaris

Pemphigus vulgaris is characterized by flaccid bullae that erode, leaving weeping areas (**Fig. 3.57**). Conjunctivitis is uncommon, mild, and self-limiting.

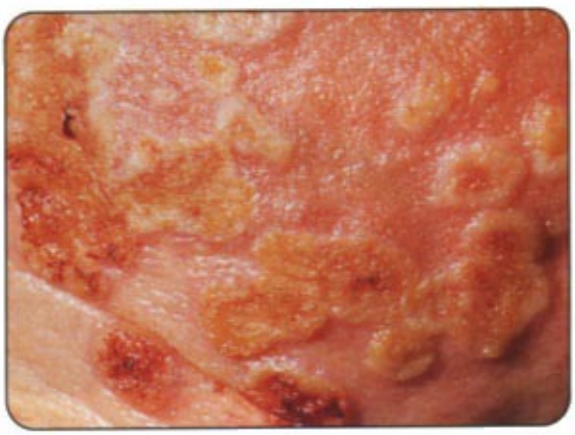


Fig. 3.57

Linear IgA bullous dermatosis

Linear IgA bullous dermatitis is characterized by tense mucocutaneous blisters (**Fig. 3.58**). Conjunctivitis is common and may result in severe scarring.

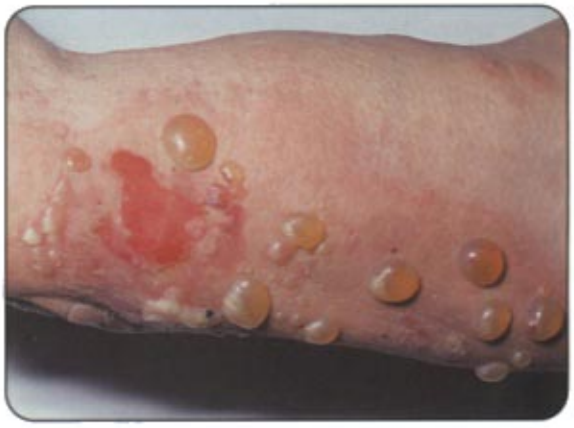


Fig. 3.58

Cutaneous disorders

Atopic dermatitis

Atopic dermatitis is a common condition which is frequently associated with asthma and hay fever. It is characterized by itchy, dry, and excoriated skin lesions, most frequently involving the elbow and knee flexures (**Fig. 3.59**). Severe conjunctivitis may give rise to forniceal shortening and symblepharon formation.

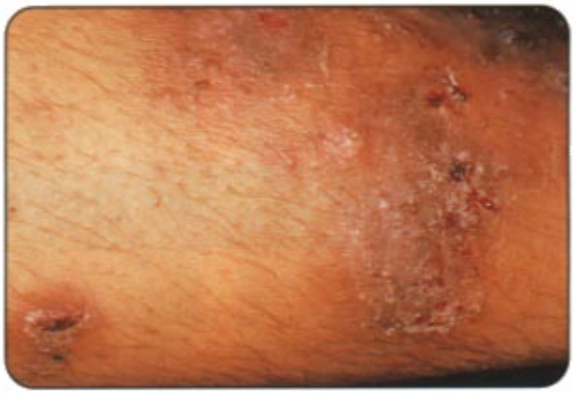


Fig. 3.59

Xeroderma pigmentosum

Xeroderma pigmentosum is a rare, recessively inherited condition characterized by photosensitive skin lesions and multiple skin malignancies (**Fig. 3.60**). Conjunctivitis is common and may result in scarring.

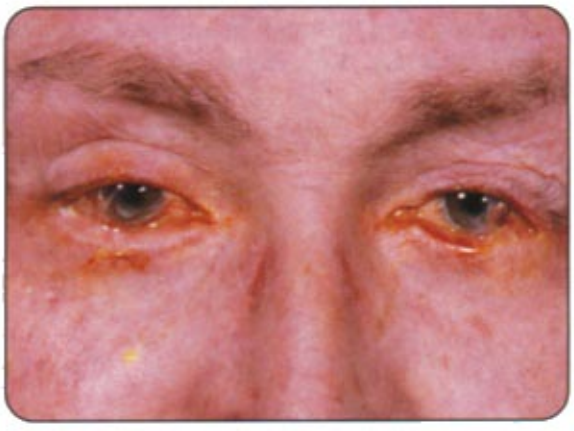


Fig. 3.60

Scleroderma

Scleroderma is a rare disease which is characterized by tight, hard, waxy skin (**Fig. 3.61**) and Raynaud phenomenon. Conjunctivitis may cause mild forniceal shortening in some patients.

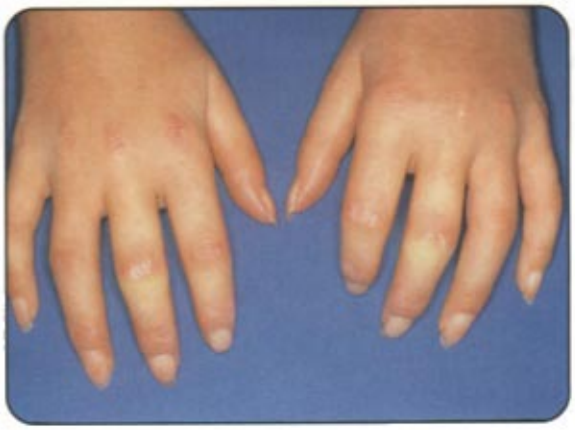


Fig. 3.61

Toxic causes

The following drugs may give rise to a pseudopemphigoid conjunctivitis:

- Topical – epinephrine, pilocarpine, and echothiophate.
- Systemic – practolol and fluorouracil.

Chemical injury, particularly with alkalis, may also result in severe conjunctival scarring.

SECONDARY CHRONIC 'CONJUNCTIVITIS'

The following conditions should be considered in patients with chronic nonspecific ocular irritation, the cause of which is not immediately apparent.

Chronic blepharitis

Chronic blepharitis is a very common, bilateral condition which may cause nonspecific ocular irritation which is worse in the mornings. On the other hand, some patients have severe blepharitis (**Fig. 3.62**) but no symptoms.

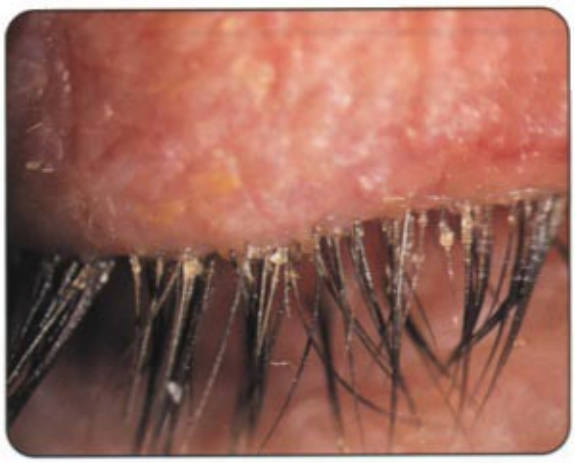


Fig. 3.62

Look for

- Associated atopic and seborrheic dermatitis, and rosacea.

Keratoconjunctivitis sicca

Keratoconjunctivitis sicca is a very common, bilateral condition which may be difficult to diagnose if mild.

Look for

- Reduced or absent tear meniscus.
- Corneal filaments and mucous plaques.
- Interpalpebral staining of conjunctiva and cornea with rose bengal (**Fig. 3.63**).
- Reduced tear film break-up time.

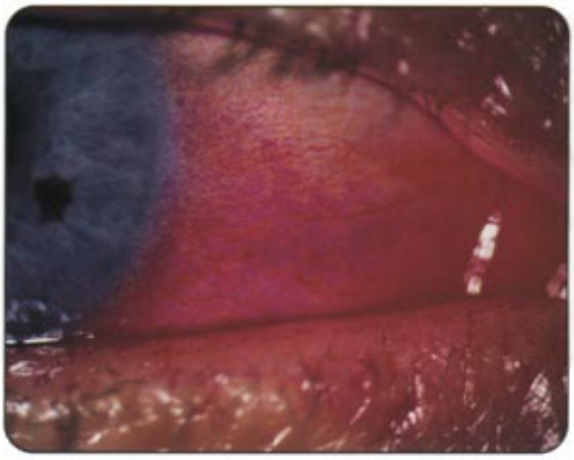


Fig. 3.63

Rosacea

Rosacea is a common skin disorder which may not be easy to diagnose as skin lesions may be mild and ocular symptoms severe.

Look for

- Typical facial telangiectasis involving the nose, cheeks, and forehead (**Fig. 3.64**).
- Posterior blepharitis (**Fig. 3.65**).
- Inferior keratopathy.
- Episcleritis.

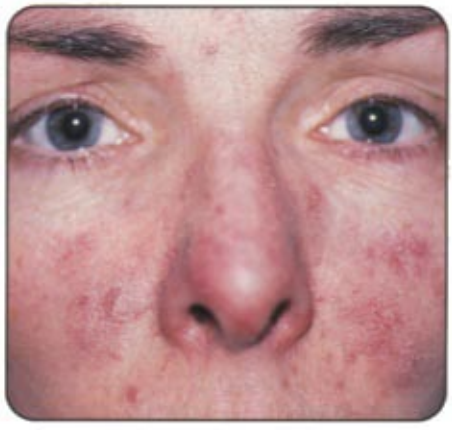


Fig. 3.64

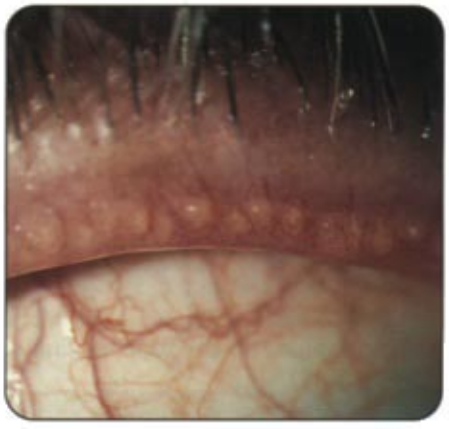


Fig. 3.65

Chronic canaliculitis

Chronic canaliculitis is an uncommon, usually unilateral condition which may be difficult to diagnose.

Look for

- Pouting punctum (**Fig. 3.66**) and concretions within the canaliculus.

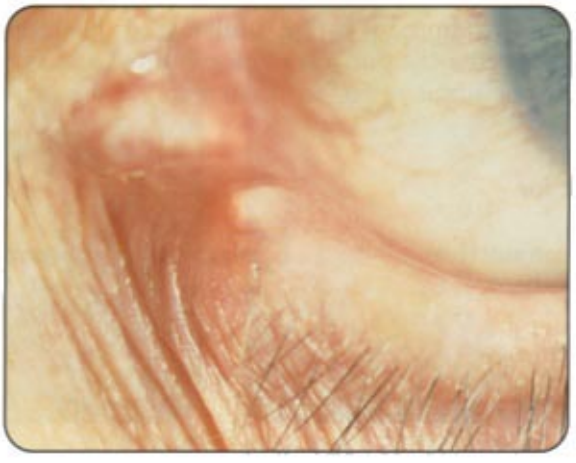


Fig. 3.66

Chronic dacryocystitis

Chronic dacryocystitis is a common, usually unilateral condition which is easy to diagnose because the patient complains of epiphora and a mucoid discharge.

Look for

- Mucocele (**Fig. 3.67**) – pressing on the lacrimal sac causes regurgitation of pus and mucus.

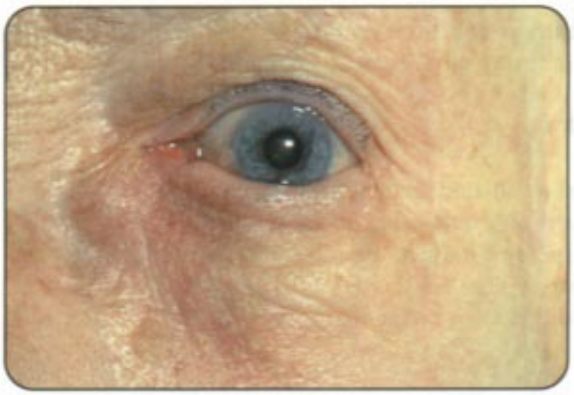


Fig. 3.67

Floppy eyelid syndrome

Floppy eyelid syndrome is a rare, unilateral or bilateral condition which may be difficult to diagnose. It typically affects very obese men with very lax upper eyelids which become everted and are traumatized during sleep.

Signs

- Severe laxity of upper eyelids (**Fig. 3.68**).
- Fine papillae on the superior tarsus (**Fig. 3.69**).

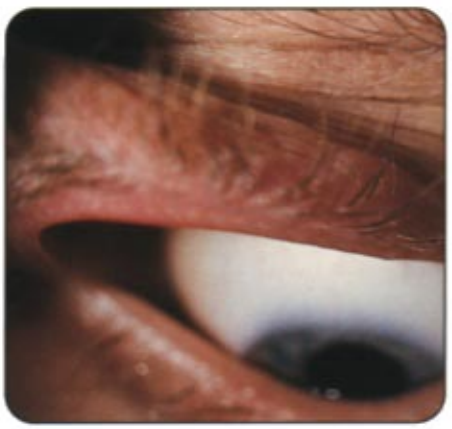


Fig. 3.68

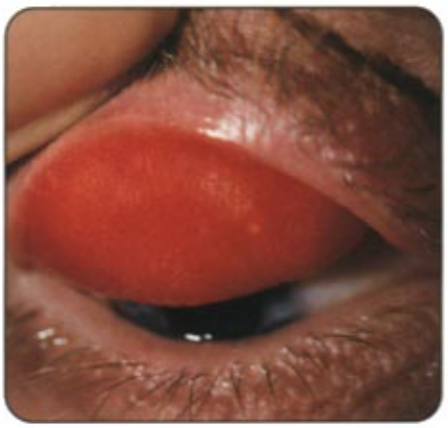


Fig. 3.69

Mucus fishing syndrome

Mucus fishing syndrome is a rare, unilateral or bilateral condition which may be difficult to diagnose because the patient may not admit to self-traumatization of the conjunctiva.

Look for

- Excess conjunctival mucus and isolated areas staining with rose bengal (**Fig. 3.70**).

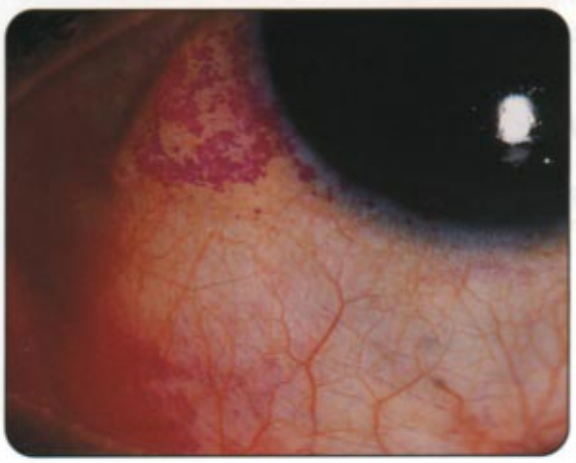


Fig. 3.70

Lid inbrication syndrome

Lid inbrication syndrome is a rare condition caused by overriding of the lower lid by the upper lid (**Fig. 3.71**).

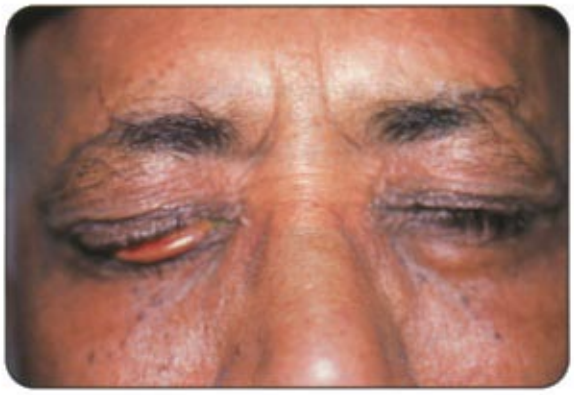


Fig. 3.71

Look for

- Staining of the upper tarsus with rose bengal.

Masquerade syndrome

Masquerade syndrome is very rare and unilateral. It is most frequently caused by infiltration of the conjunctiva by spreading sebaceous gland carcinoma (pagetoid spread) (**Fig. 3.72**).

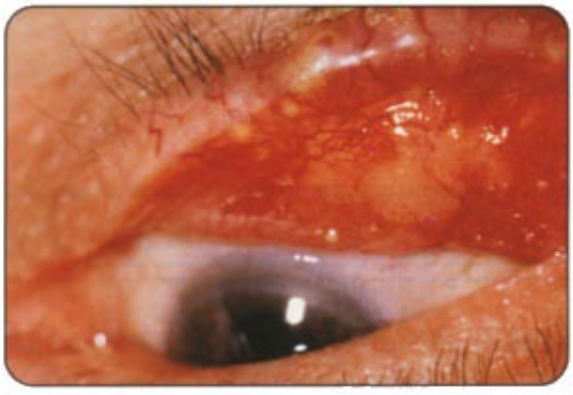


Fig. 3.72

PIGMENTED LESIONS

Flat superficial pigmentation

Common, small, congenital focal lesions

Causes

- Conjunctival freckle (**Fig. 3.73**).
- Melanosis around Axenfeld loop (**Fig. 3.74**) – an intrascleral nerve loop located about 4 mm from the limbus.
- Melanosis around anterior ciliary arteries.

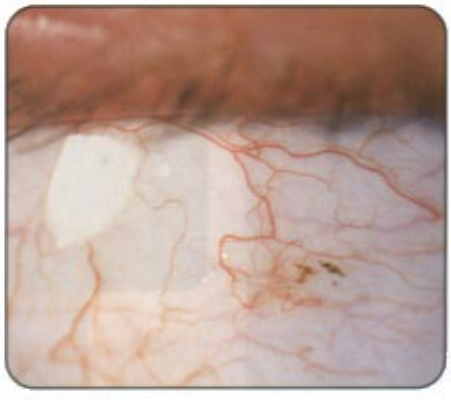


Fig. 3.73

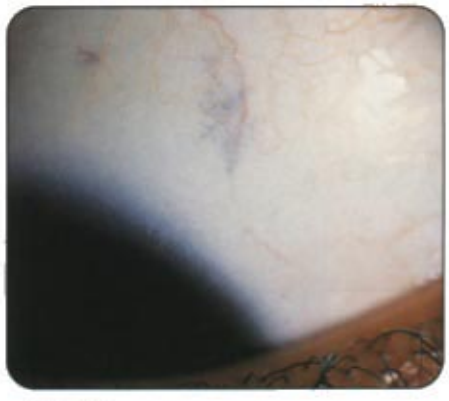


Fig. 3.74

Benign epithelial (racial) melanosis

Signs

- Bilateral, yellow–brown or brown–black patches most prominent at the limbus (**Fig. 3.75**) and in the interpalpebral bulbar conjunctiva (**Fig. 3.76**), which fade toward the fornices. The lesions can be moved easily on the globe.

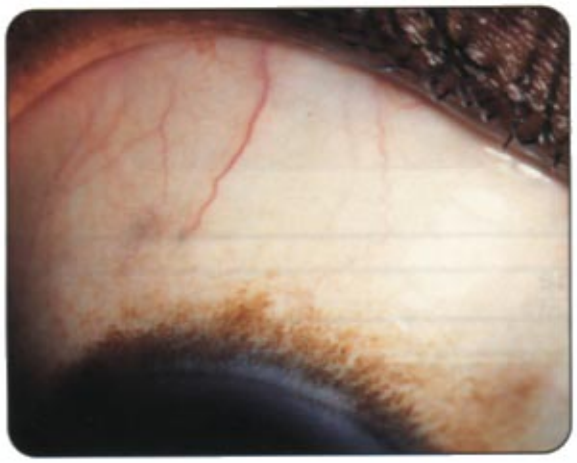


Fig. 3.75

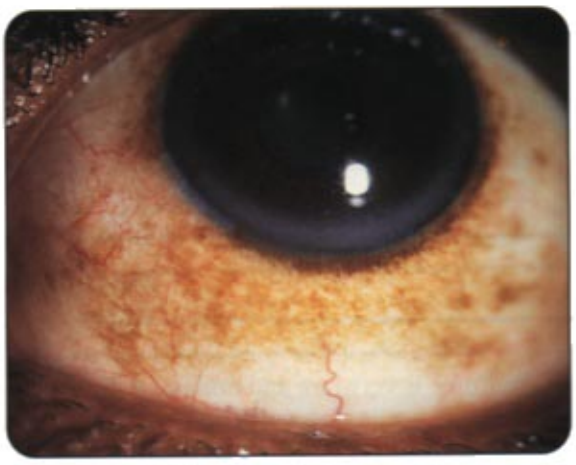


Fig. 3.76

Primary acquired melanosis

Primary acquired melanosis is a rare, unilateral, premalignant condition which is usually seen in whites.

Signs

- Patches with indistinct margins which may involve any part of the conjunctiva (**Fig. 3.77**). Malignant change should be suspected if the patches become nodular (**Fig. 3.78**).

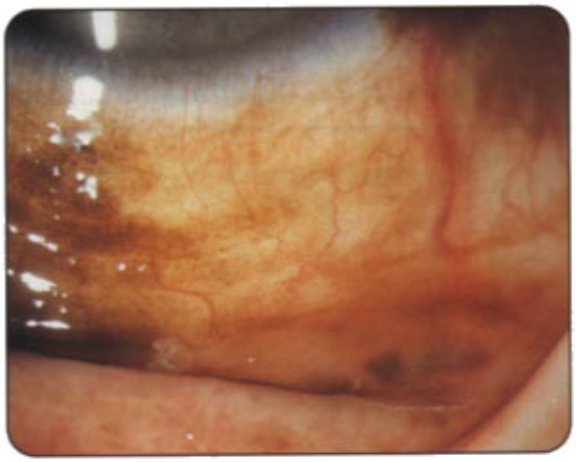


Fig. 3.77

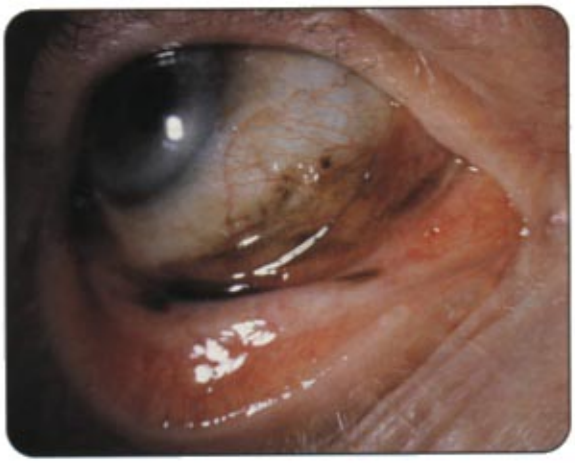


Fig. 3.78

Secondary acquired melanosis

Causes

- Adrenochrome deposits — discrete clumps of melanin on tarsal and forniceal conjunctiva (**Fig. 3.79**) associated with long-term use of topical epinephrine.
- Mascara (**Fig. 3.80**).
- Chronic epithelial irritation (e.g. inflammation, glaucoma blebs).
- Age-related.
- Metabolic disorders (e.g. Addison disease).
- Dark foreign bodies (**Fig. 3.81**).

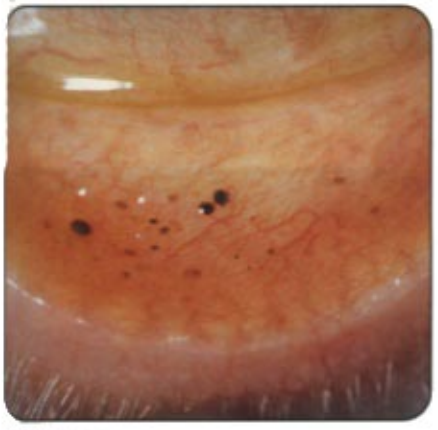


Fig. 3.79

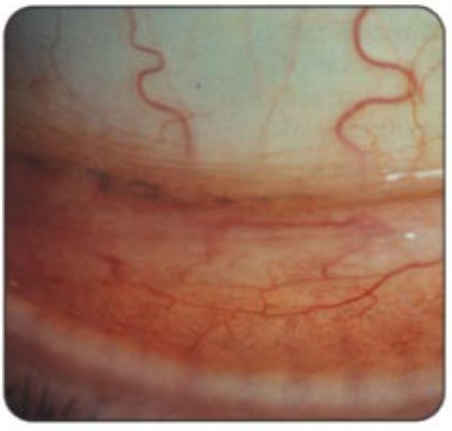


Fig. 3.80

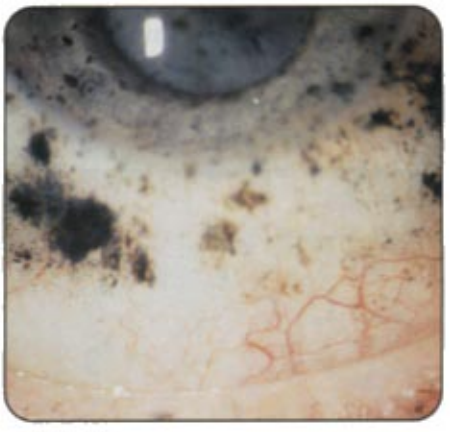


Fig. 3.81

Flat deep pigmentation

Flat deep pigmentation is characteristic of subepithelial melanocytosis – a rare, unilateral, congenital condition which has a slate blue–gray color. Because it affects the episcleral and scleral tissues, it cannot be moved over the globe. The condition may affect the skin and mucous membranes in the distribution of the trigeminal nerve. The three possible patterns are:

- Dermal melanocytosis – involves only the skin.
- Isolated melanocytosis oculi – involves only the globe (**Fig. 3.82**).
- Oculodermal melanocytosis (nevus of Ota) – involves both the skin and the globe (**Fig. 3.83**).

Ocular associations

- Iris hyperpigmentation.
- Iris mammillations.
- Trabecular hyperpigmentation.
- Increased risk of glaucoma.
- Increased risk of uveal melanoma.

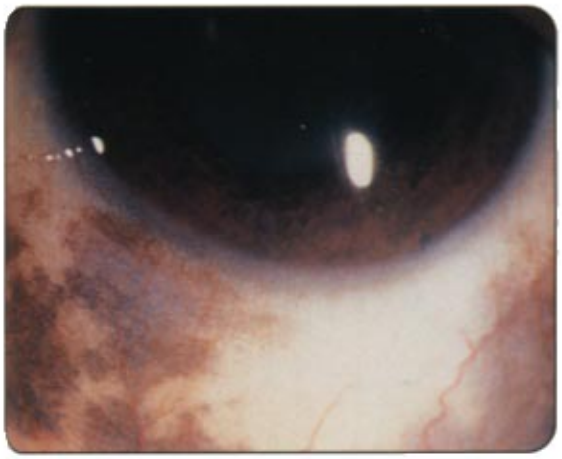


Fig. 3.82

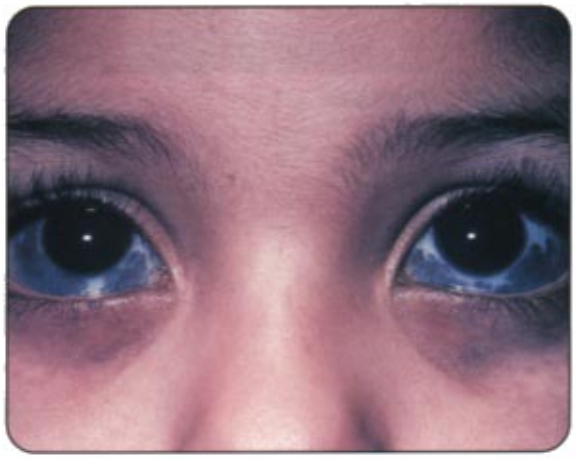


Fig. 3.83

Miscellaneous flat discoloration

Red

Red flat discoloration is due to subconjunctival hemorrhage.

Causes

- Spontaneous hemorrhage is by far the most common type. The hemorrhage is usually unilateral, but when precipitated by coughing or straining it may rarely be bilateral (**Fig. 3.84**).
- Associated with bleeding disorders and the use of anticoagulants, when it may also be associated with periocular cutaneous ecchymosis (**Fig. 3.85**).
- Associated with conjunctivitis or scleritis (**Fig. 3.86**).
- Traumatic hemorrhage may be associated with hyphema (**Fig. 3.87**) and chemosis (**Fig. 3.88**).

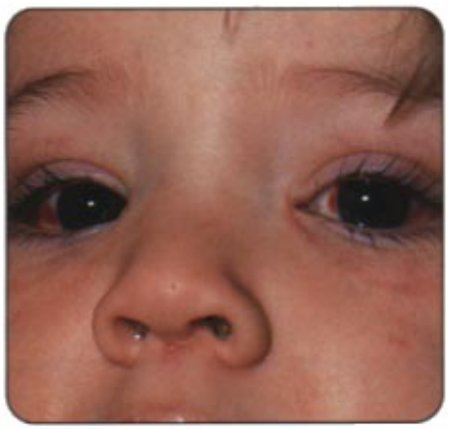


Fig. 3.84

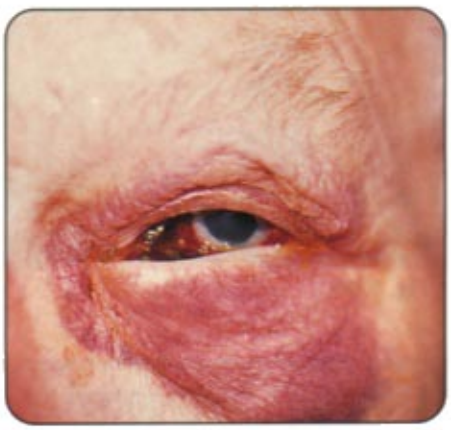


Fig. 3.85

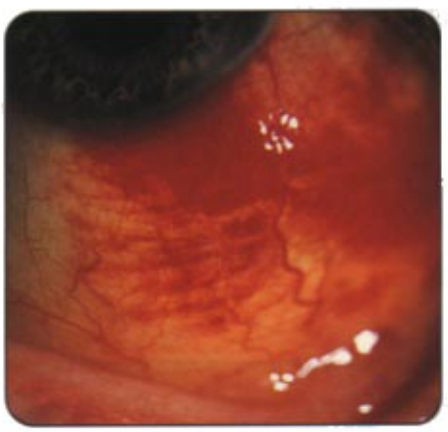


Fig. 3.86

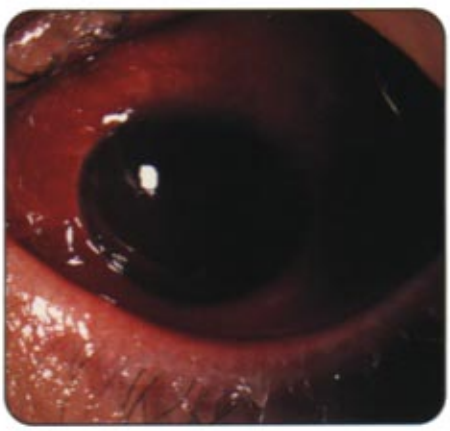


Fig. 3.87

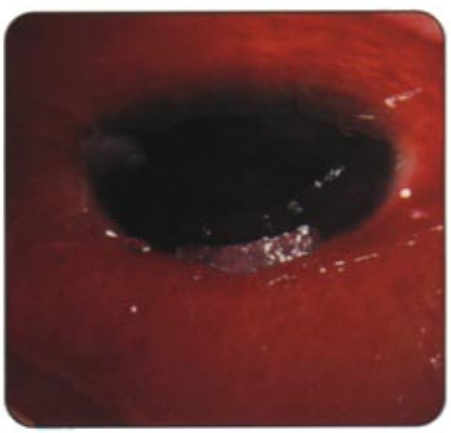


Fig. 3.88

Other types of flat discoloration

Causes

Gray or black

- Alkaptonuria – interpalpebral, bluish-gray or black pigmentation of the conjunctiva, episclera, sclera, and tendons of horizontal recti.
- Argyrosis – diffuse slate-gray pigmentation.
- Mercury – bluish-gray deposits around conjunctival blood vessels.

Rusty brown

- Resolving subconjunctival hemorrhage.
- Hemochromatosis – affects mainly the inferior perilimbal conjunctiva.
- Clofazimine, a drug used to treat leprosy – diffuse reddish discoloration.

Yellow

- Jaundice – bilirubin deposition in conjunctiva and sclera.
- Intravenous fluorescein injection.
- Rarely, chronic administration of chloroquine and hydroxychloroquine.

Pigmented nodules

Nevus

A nevus is an uncommon, benign tumor.

Signs

- Solitary, well-defined, slightly elevated lesion (**Fig. 3.89**) which moves freely over the globe. Nevi have a predilection for the limbus (**Fig. 3.90**), plica, caruncle, and lid margin (**Fig. 3.91**), hence any elevated pigmented lesion of either the palpebral or forniceal conjunctiva is unlikely to be a nevus. Most nevi have a tan or brown color, but about 25% are nonpigmented.

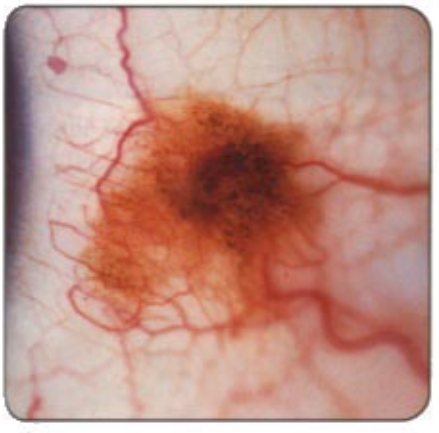


Fig. 3.89

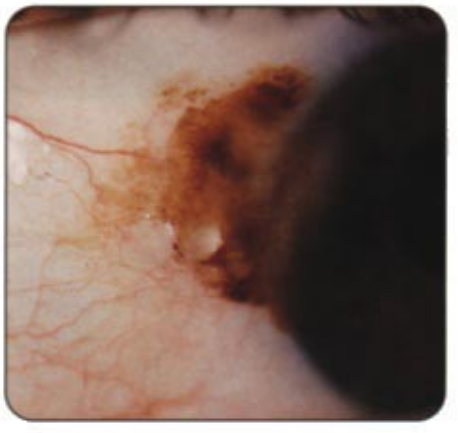


Fig. 3.90

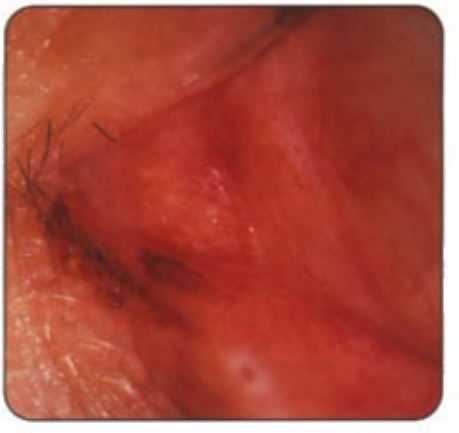


Fig. 3.91

Melanocytoma

A melanocytoma is a very rare, congenital, benign tumor.

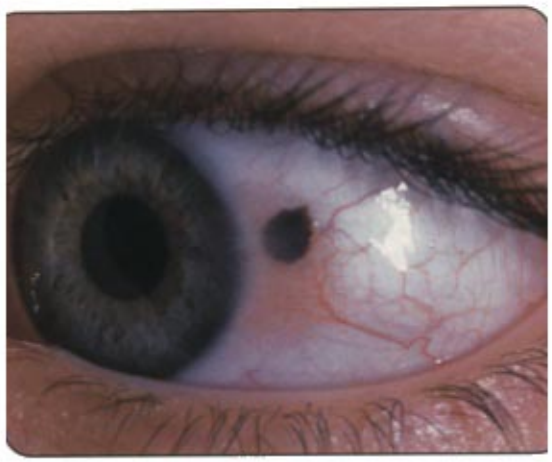


Fig. 3.92

Signs

- Black, slowly growing lesion (**Fig. 3.92**) which does not move freely over the globe.

Primary melanoma

A primary melanoma is a very rare, malignant tumor which may be pigmented or nonpigmented. It may arise *de novo* or from pre-existing primary acquired melanosis.

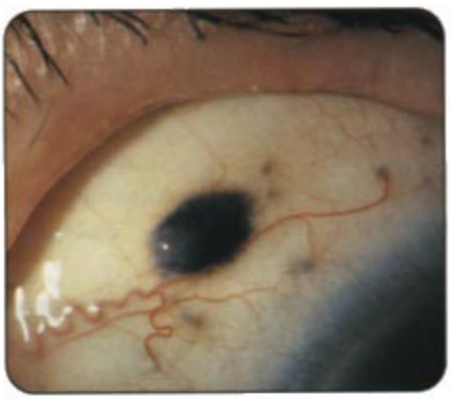


Fig. 3.93

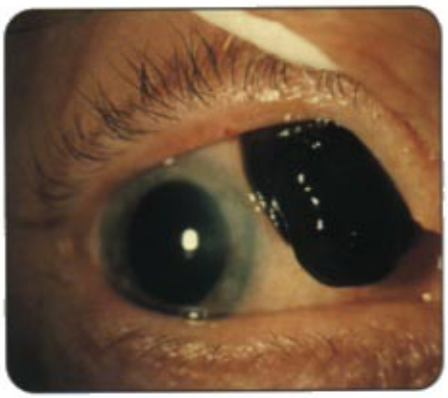


Fig. 3.94

Signs

- Elevated nodule which can affect any part of the conjunctiva (**Figs 3.93, 3.94**) but has a predilection for the limbus and may extend onto the cornea (**Fig. 3.95**). Advanced melanomas may invade the eyelids and orbit (**Fig. 3.96**).

Dx Differential diagnosis

- Pigmented papilloma.
- Pigmented squamous cell carcinoma.
- Extension of ciliary-body melanoma.

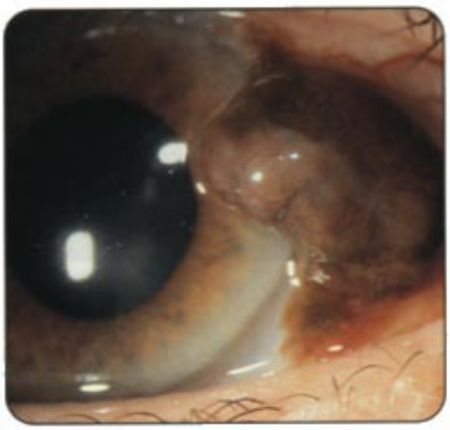


Fig. 3.95

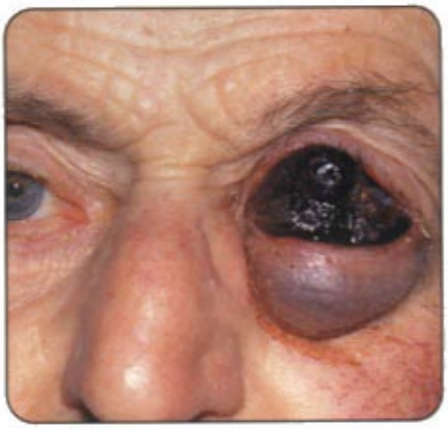


Fig. 3.96

Melanoma developing in primary acquired melanosis

Signs

- One or more nodules gradually growing within an area of primary acquired melanosis (**Fig. 3.97**).

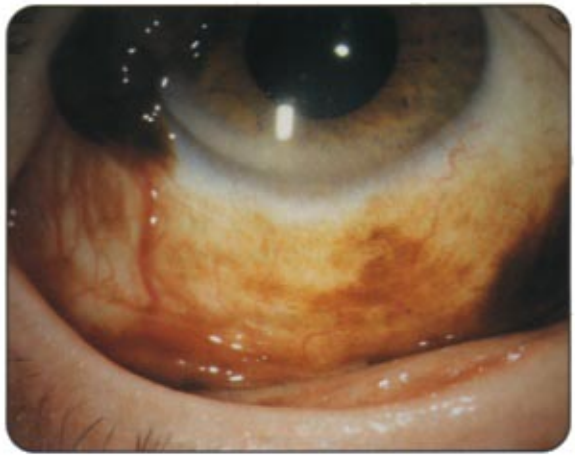


Fig. 3.97

NONPIGMENTED LESIONS

Small nodules

Granulomas

- Chalazion – single nodule on the tarsal conjunctiva (**Fig. 3.98**).
- Pyogenic granuloma – single nodule at the limbus and most commonly occurring after conjunctival surgery (**Fig. 3.99**).
- Sarcoid – multiple nodules involving the tarsal or forniceal conjunctiva (**Fig. 3.100**).
- Rhinosporidiosis – very rare fungal infection which may cause conjunctival granulomas (**Fig. 3.101**).
- Vasculitides (e.g. polyarteritis nodosa, Churg–Strauss syndrome) – very friable lesions.

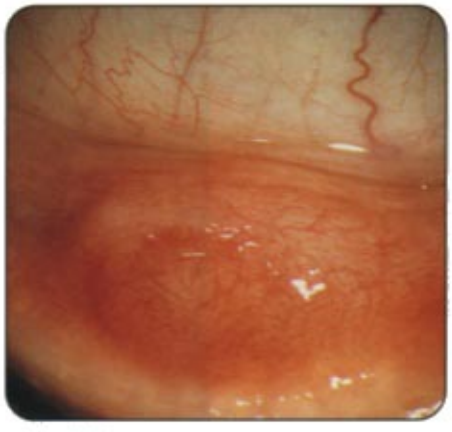


Fig. 3.98

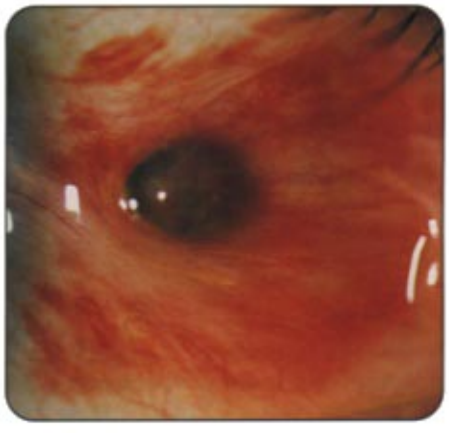


Fig. 3.99

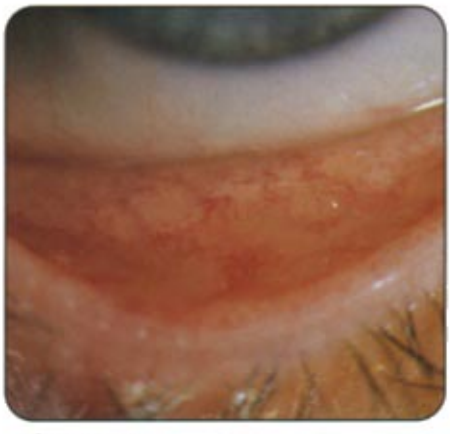


Fig. 3.100

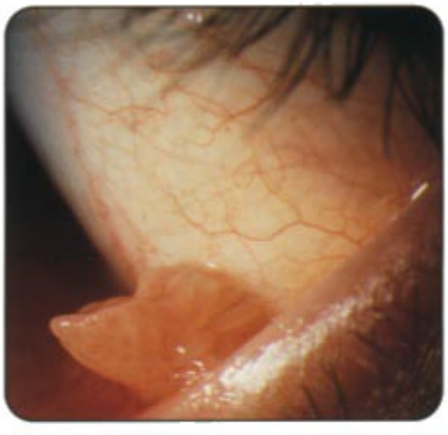


Fig. 3.101

Pinguecula

Pingueculas are very common, usually bilateral lesions on the bulbar conjunctiva, adjacent to the nasal or temporal limbus.

Signs

- Gray–white or yellow nodule near the nasal limbus (**Fig. 3.102**), which may occasionally become inflamed.

Differential diagnosis of pinguecula-like lesions

- Adult-type Gaucher disease – prominent, dark-yellow, usually nasal lesions are found in 25% of cases.
- Multiple endocrine neoplasia IIb – lesions consist of coiled myelinated nerves and may also be seen on the eyelids.
- Primary localized nodular amyloidosis – brownish-red nodules on the conjunctiva and eyelids.
- Xanthelasma – usually seen in type IIa hypercholesterolemia.
- Tophi in gout.

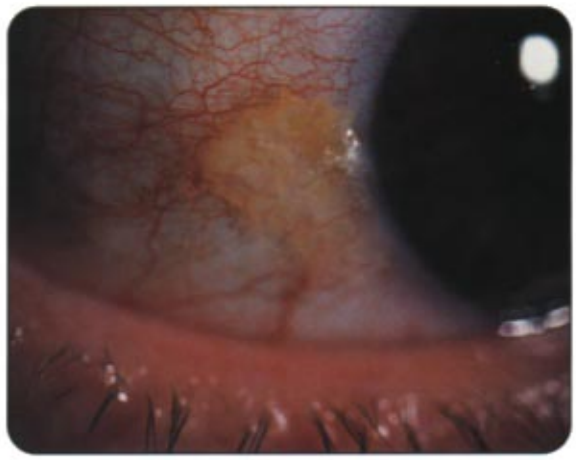


Fig. 3.102

Phlycten

A phlycten is an uncommon, unilateral, oval lesion associated with surrounding hyperemia (**Fig. 3.103**), which may extend onto the limbus.

Causes

- Staphylococcal infection.
- Adenoviral infection.
- Herpes simplex.
- Tuberculosis.
- Candidal infection.
- Lymphogranuloma venereum.

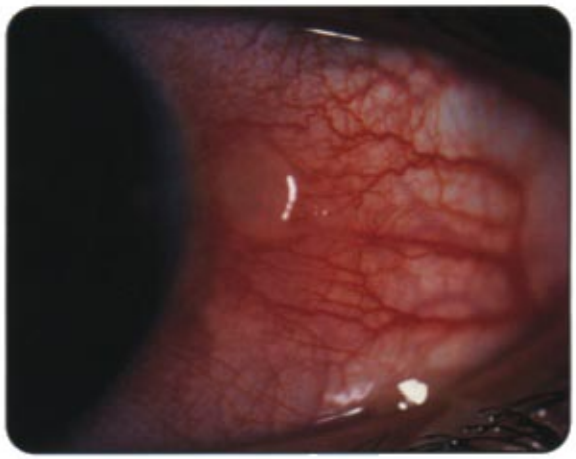


Fig. 3.103

Large nodules

Pseudoepitheliomatous hyperplasia

Pseudoepitheliomatous hyperplasia is an uncommon, rapidly developing condition secondary to irritation.

Signs

- White, elevated, hyperkeratotic nodule near the limbus (**Fig. 3.104**).

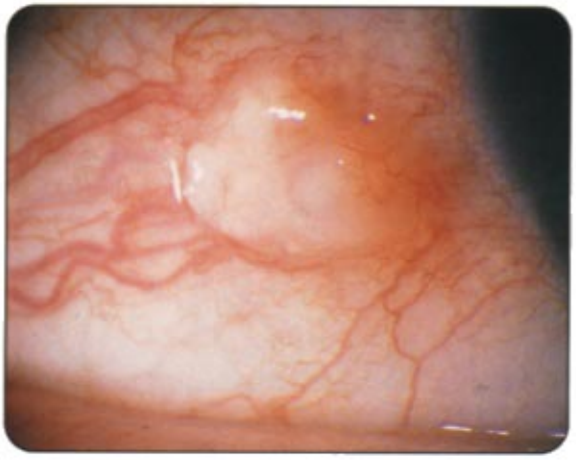


Fig. 3.104

Epibulbar dermoid

Epibulbar dermoid is an uncommon, congenital lesion which may occur in isolation or in association with other ocular or systemic anomalies.

Signs

- Solid, smooth, round white mass located at the limbus (**Figs 3.105, 3.106**). Large lesions may encroach onto the cornea (**Fig. 3.107**).

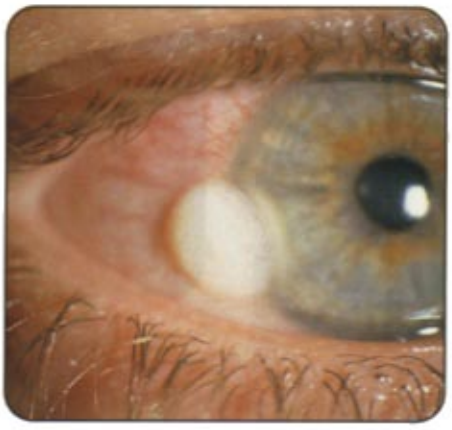


Fig. 3.105

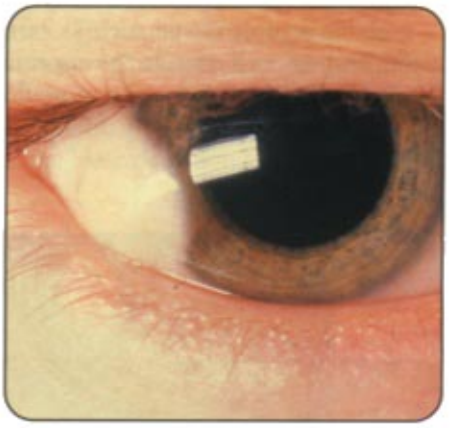


Fig. 3.106

Ocular associations

- Lid coloboma.
- Ocular coloboma.
- Microphthalmos.
- Aniridia.

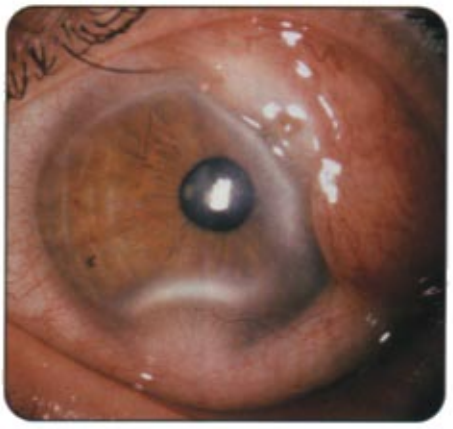


Fig. 3.107

Systemic associations

- Goldenhar syndrome (**Fig. 3.108**).
- Treacher Collins syndrome.
- Franceschetti syndrome.

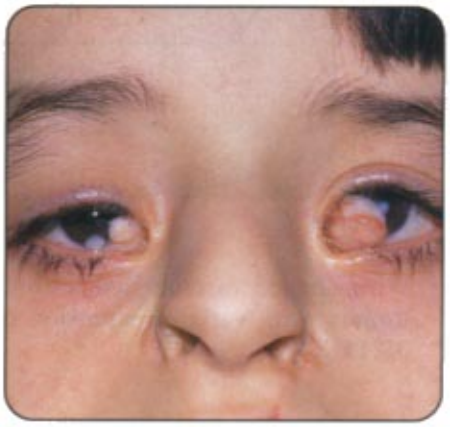


Fig. 3.108

Bitot spot

Bitot spot is an uncommon, usually bilateral lesion associated with vitamin A deficiency.

Signs

- Foamy-looking plaque temporal to the limbus (**Fig. 3.112**).

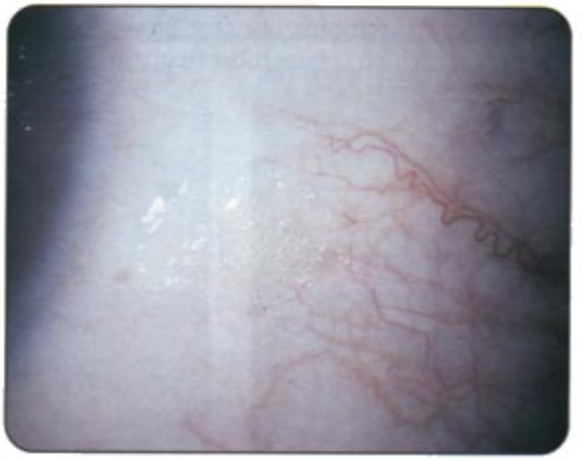


Fig. 3.112

Conjunctival intraepithelial neoplasia

Conjunctival intraepithelial neoplasia is a rare, unilateral, premalignant condition which is seen in older, fair-skinned individuals. Formerly, this condition was referred to as Bowen disease, intraepithelial epithelioma, and conjunctival dyskeratosis.

Signs

The lesions are usually located at the limbus and may involve adjacent cornea. The three clinical types are:

- Fleshy gelatinous lesion with variable keratinization (**Fig. 3.113**).
- White plaque (leukoplakic-type) (**Fig. 3.114**).
- Papillary, which is the least common.

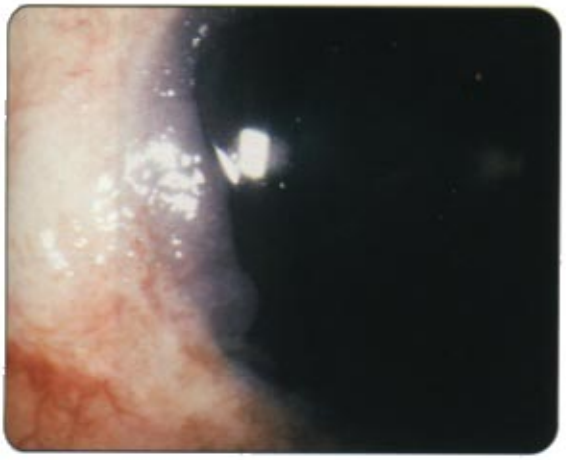


Fig. 3.113

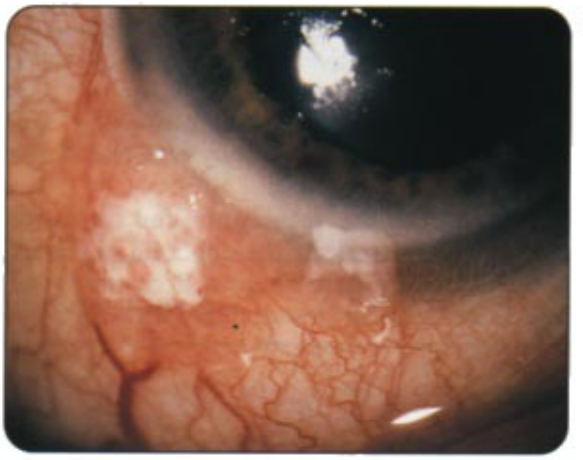


Fig. 3.114

Invasive squamous cell carcinoma

Invasive squamous cell carcinoma is a rare, slow-growing, locally invasive tumor which occurs most frequently at the limbus.

Signs

- Papillary (**Figs 3.115, 3.116**) or gelatinous tumor which is frequently associated with the feeder blood vessels (**Fig. 3.117**). The lesion is usually larger than conjunctival intraepithelial hyperplasia and may involve a greater area of the limbus.

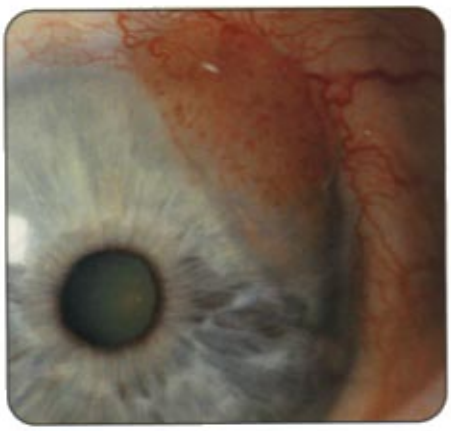


Fig. 3.115

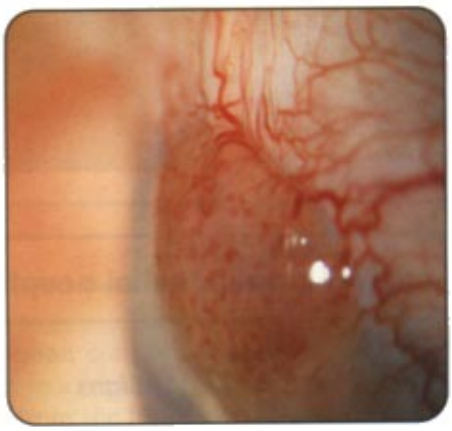


Fig. 3.116

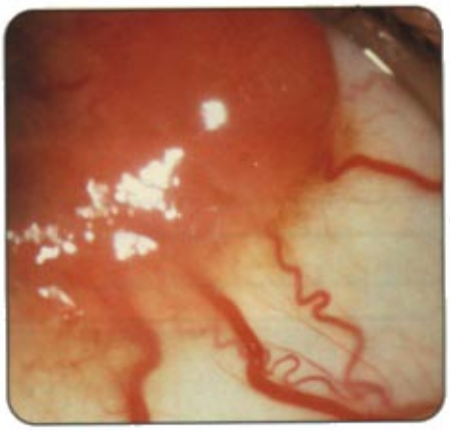


Fig. 3.117

Other carcinomas

- Mucoepidermoid carcinoma is similar to squamous cell carcinoma but more aggressive, and may arise anywhere on the conjunctiva.
- Spindle cell carcinoma is similar to squamous cell carcinoma but more aggressive, and may arise anywhere on the conjunctiva.
- Sebaceous gland carcinoma is a very rare and aggressive tumor which typically involves the upper lid of elderly patients but may rarely arise *de novo* from the tarsal conjunctiva as a plaque-like lesion (**Fig. 3.118**).

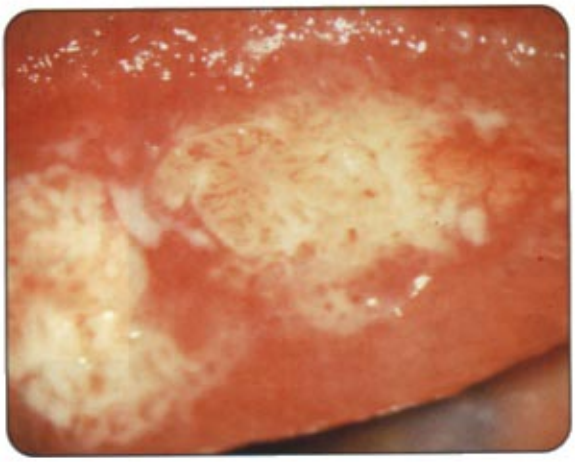


Fig. 3.118

Diffusely elevated lesions

Lipodermoid

Lipodermoid is a common and usually bilateral condition, typically found in adults.

Signs

- Large, yellow, soft, movable, subconjunctival lesions located most commonly at the outer canthus (**Fig. 3.119**). The lesions extend into the superior fornices, and therefore it is impossible to visualize their posterior limits (**Fig. 3.120**).

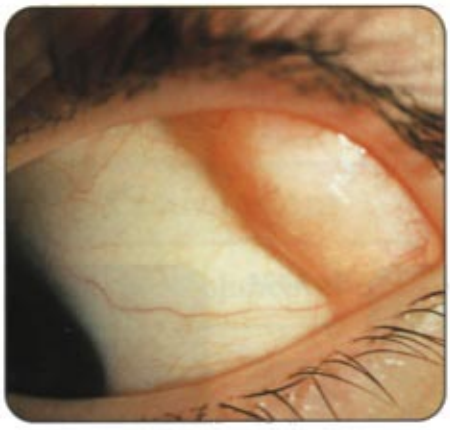


Fig. 3.119

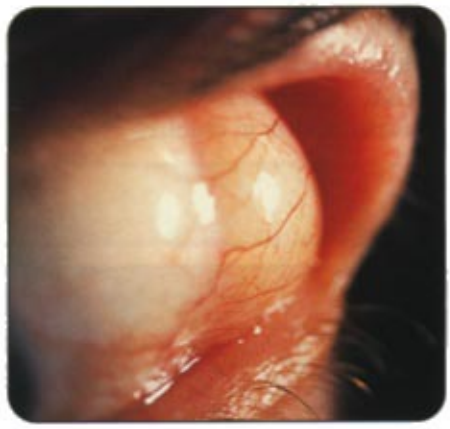


Fig. 3.120

Non-Hodgkin lymphoma

Signs

- Smooth, fleshy, subconjunctival infiltrate (**Fig. 3.121**) which may involve a large area (**Fig. 3.122**). The lesion may be single or multiple and involves both eyes in about 20% of cases. Small affected areas are called 'salmon patches'.

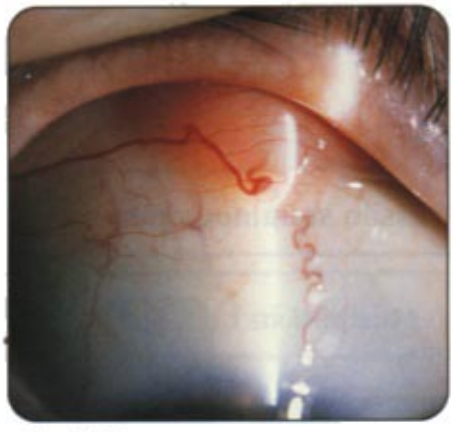


Fig. 3.121

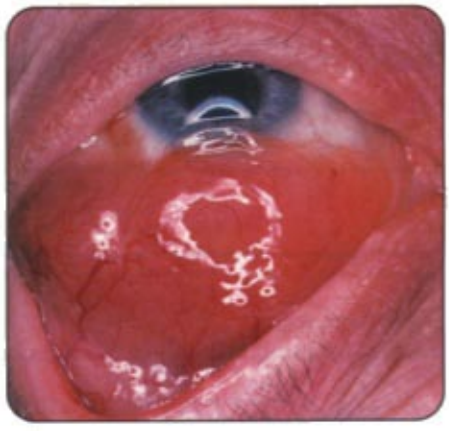


Fig. 3.122

Reactive lymphoid hyperplasia

Signs

- The appearance (**Fig. 3.123**) is similar to that of lymphoma.

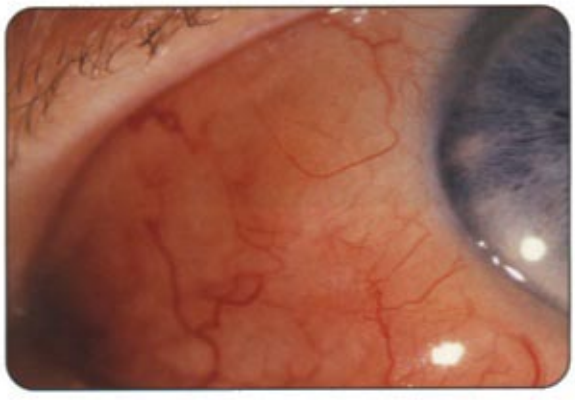


Fig. 3.123

Papillary lesions

Pedunculated squamous papilloma

Pedunculated squamous papilloma is an uncommon, benign tumor caused by the human papillomavirus. It typically occurs in children and young adults.

Signs

- Tumor with 'finger-like' projections, located in the palpebral conjunctiva (**Fig. 3.124**), fornix, or caruncle (**Fig. 3.125**). The lesion may be multifocal and bilateral.

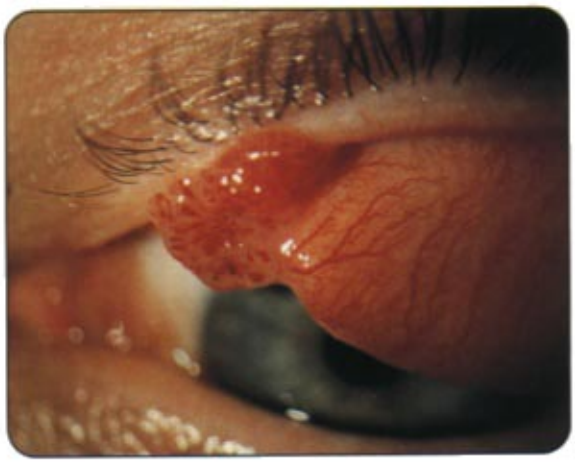


Fig. 3.124

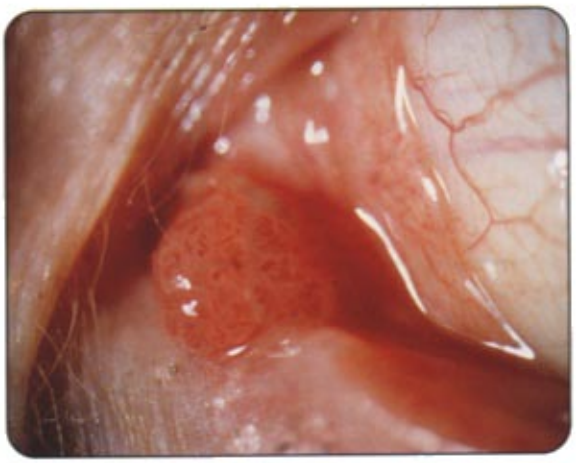


Fig. 3.125

Sessile squamous papilloma

Sessile squamous papilloma is an uncommon, benign, non-infectious tumor which affects older patients.

Signs

- Unilateral, single flat lesion located on the bulbar conjunctiva, near the limbus (**Fig. 3.126**) .

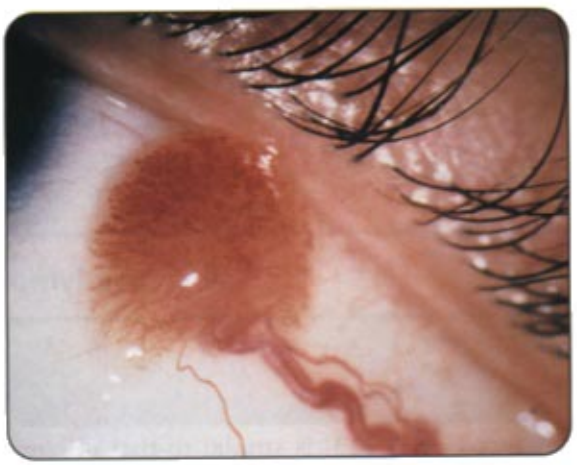


Fig. 3.126

Other papillary lesions

- Inverted conjunctival papilloma is a rare, unilateral tumor containing mucus-secreting cells.
- Conjunctival intraepithelial neoplasia may occasionally assume a papillary configuration.
- Conjunctival carcinomas – squamous cell, mucoepidermoid, and spindle cell carcinomas may assume a papillary configuration.

Cystic lesions

Primary conjunctival cyst

Signs

- A common, translucent cyst on the bulbar conjunctiva (**Fig. 3.127**).

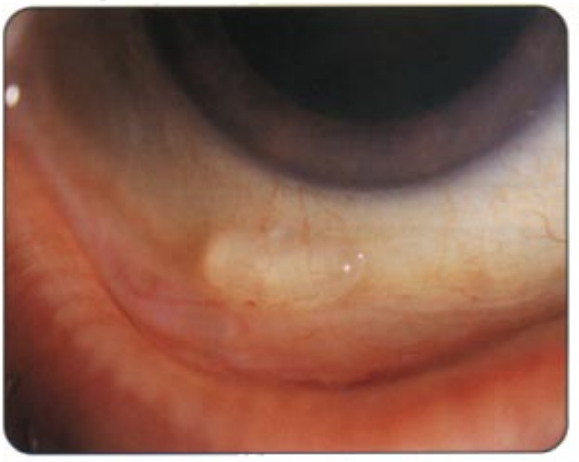


Fig. 3.127

Cyst of gland of Wolfring

Signs

- A rare, large cystic swelling involving the lower or upper tarsus (**Fig. 3.128**).

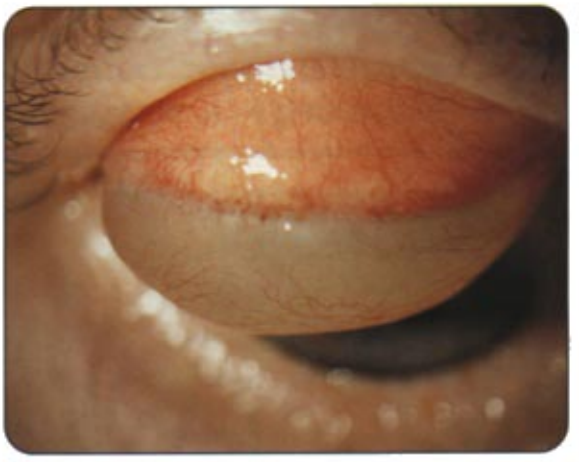


Fig. 3.128

Iatrogenic cysts

These cysts may take the following forms:

- Secondary implantation cysts following surgery (**Fig. 3.129**).
- Drainage bleb following filtration surgery, which may be flat and diffuse (**Fig. 3.130**) or localized (**Fig. 3.131**).
- Tenon cyst associated with a filtration bleb; characterized by an elevated cyst-like cavity with engorged surface vessels (**Fig. 3.132**).

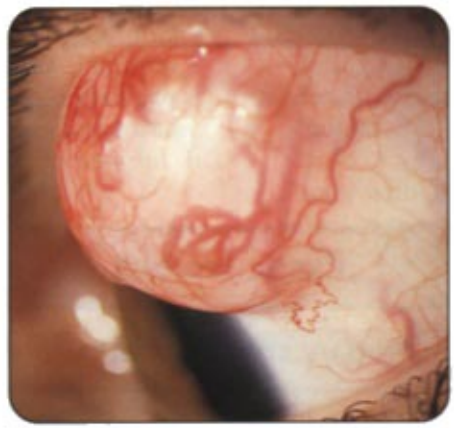


Fig. 3.129

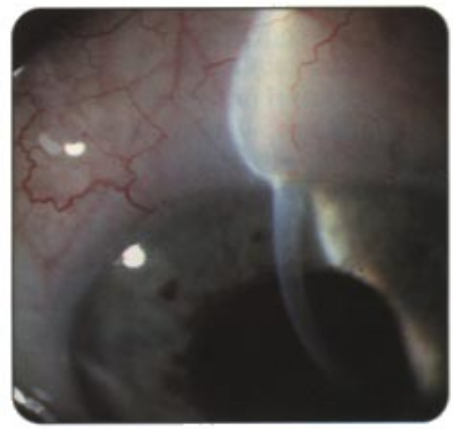


Fig. 3.130

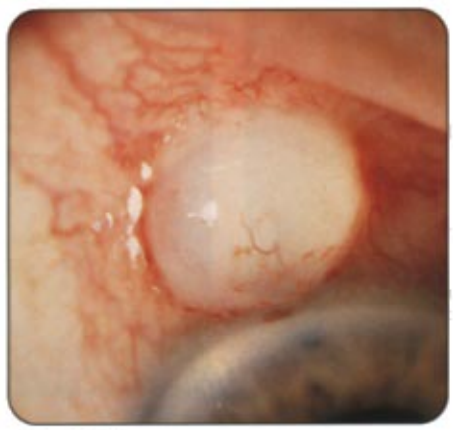


Fig. 3.131

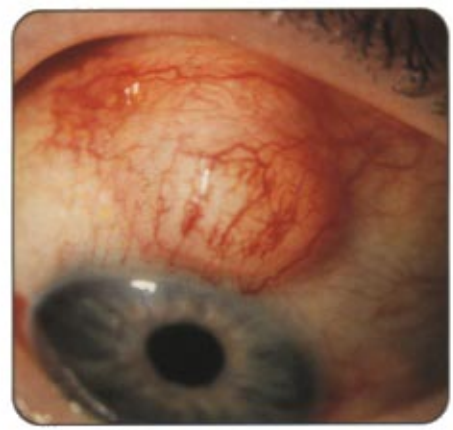


Fig. 3.132

VASCULAR LESIONS

Telangiectasias

Metabolic disorders

The following metabolic disorders may be associated with dilated and tortuous blood vessels on the bulbar conjunctiva (**Fig. 3.133**):

- Diabetes mellitus.
- Fabry disease – frequently associated with aneurysm formation.
- Fucosidosis.
- GM1 gangliosidosis.
- Multiple endocrine neoplasia IIb – associated with prominent paralimbal nerve bundles.

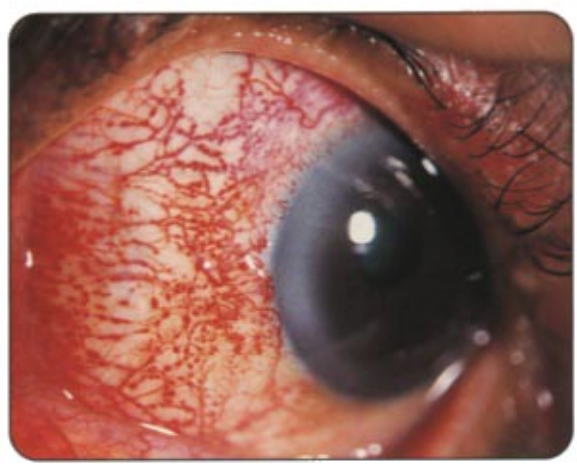


Fig. 3.133

Hematological disorders

The following hematological disorders may be associated with sludging of the blood column (**Fig. 3.134**):

- Dysproteinemias (e.g. multiple myeloma).
- Sickle cell anemia – isolated, corkscrew, or comma-shaped vessels.
- Polycythemia rubra vera.

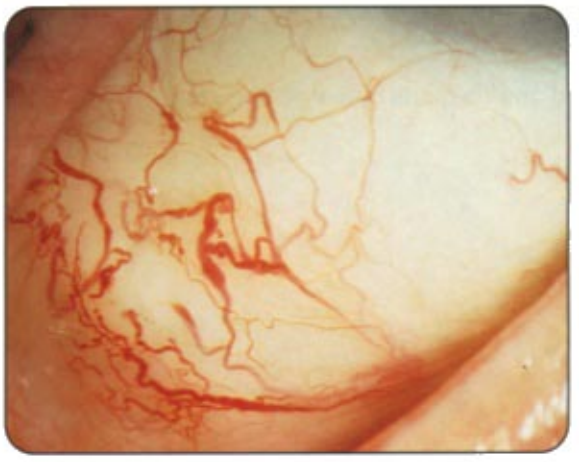


Fig. 3.134

Hemorrhagic lymphangiectasia

Hemorrhagic lymphangiectasia is an uncommon condition which may occur after mild inflammation or trauma. It may also be associated with vascular malformations of the eyelid and parotid gland.

Signs

- Dilated and tortuous bulbar lymphatic vessels (**Fig. 3.135**) which may become filled with blood if they communicate with conjunctival veins.

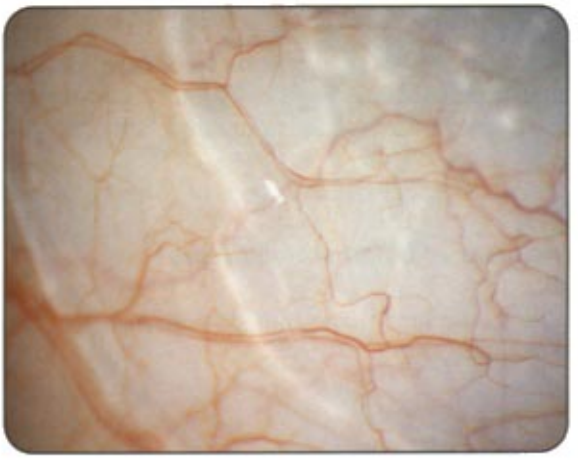


Fig. 3.135

Sturge–Weber syndrome (encephalotrigeminal angiomatosis)

Signs

- Localized telangiectasis (**Fig. 3.136**), probably associated with episcleral hemangiomas.

Look for

- Glaucoma.
- Iris hyperchromia.
- Diffuse choroidal hemangioma.

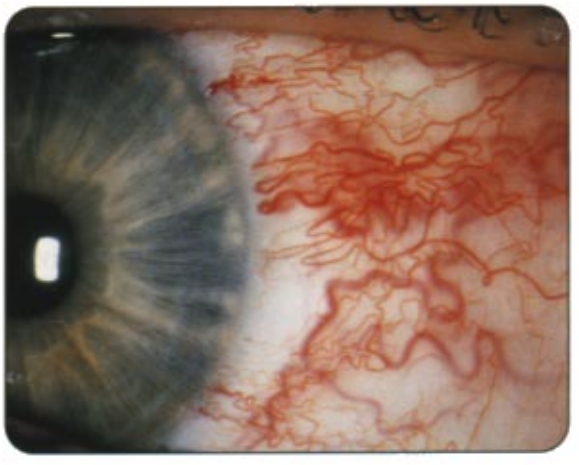


Fig. 3.136

Louis-Bar syndrome (ataxia telangiectasia)

Signs

- Engorged and tortuous bulbar conjunctival vessels (**Fig. 3.137**).

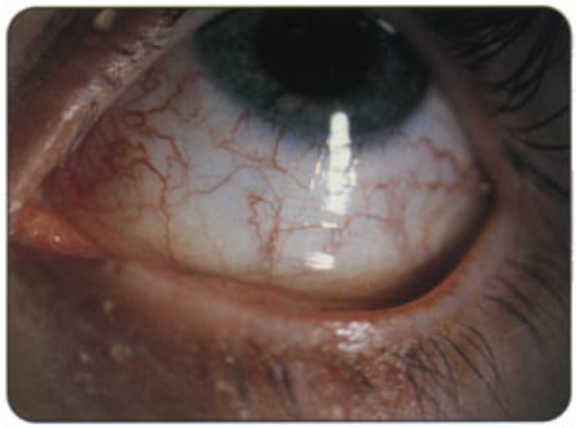


Fig. 3.137

Rendu–Osler–Weber disease (hereditary hemorrhagic telangiectasia)

Signs

- Stellate vascular conjunctival lesions are located on the palpebral (**Fig. 3.138**) but not the bulbar conjunctiva. These may cause recurrent subconjunctival hemorrhages and 'bloody' tears.
- Retinal vascular telangiectasia is seen in some patients.

Fig. 3.138

Other causes of blood-stained tears

- Conjunctival vascular tumors and granulomas.
- Lacrimal sac tumors.
- Hemophilia.

Vascular tumors

Capillary hemangioma

Capillary hemangioma is an uncommon tumor which may be associated with hemangiomas of the lids and orbit.

Signs

- Bright-red lesion which blanches on pressure (**Fig. 3.139**). It may bleed spontaneously or following trivial trauma.

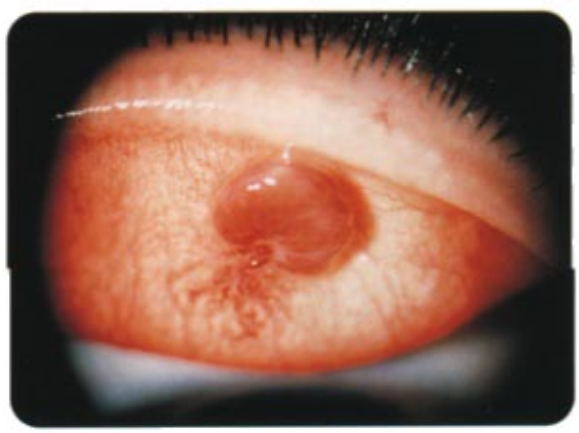


Fig. 3.139

Lymphangioma

Lymphangioma is a rare tumor which may be associated with similar lesions of the orbit, face, sinuses, and oropharynx.

Signs

- Bright-red lesion which is similar to, but may be larger than, a hemangioma (**Fig. 3.140**).

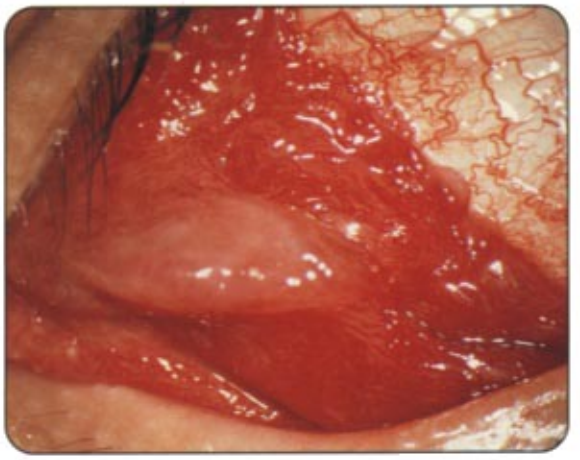


Fig. 3.140

Kaposi sarcoma

Kaposi sarcoma is common in patients with AIDS.

Signs

- A reddish vascular conjunctival lesion which may be diffuse or nodular. A diffuse tumor may resemble a subconjunctival hemorrhage on cursory examination (**Fig. 3.141**).

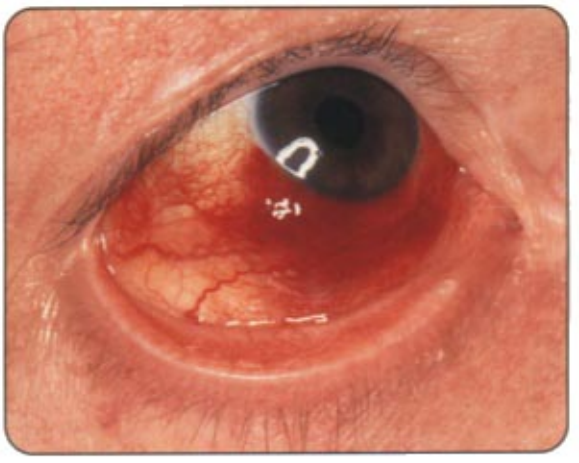


Fig. 3.141

4 The sclera

DISCOLORATION 106

Focal
Diffuse
- Yellow
- Blue

SCLERAL AND EPISCERAL INFLAMMATION 107

Not associated with a nodule
- Simple episcleritis
- Diffuse scleritis

Associated with a nodule
- Nodular episcleritis
- Nodular non-necrotizing scleritis

SCLERAL NECROSIS 109

- Necrotizing scleritis with inflammation
- Without inflammation (scleromalacia perforans)

POSTERIOR SCLERITIS 110

DISCOLORATION

Focal

Causes

- Senile scleral translucency – this is characterized by oval, dark-grayish areas anterior to insertion of the horizontal recti (**Figs 4.1, 4.2**).
- Alkaptonuria – this becomes manifest between the ages of 20 and 40 years and may be associated with pigmentation of the pinnae of the ears. It is characterized by brown–black pigmentation (onchronosis) at the insertions of tendons of the horizontal recti.
- Hemachromatosis – this may cause a rusty-brown discoloration anterior to the insertions of the horizontal recti.
- Longstanding metallic foreign body.

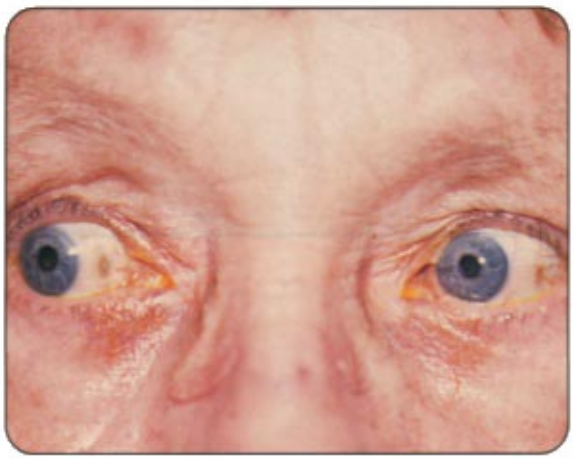


Fig. 4.1

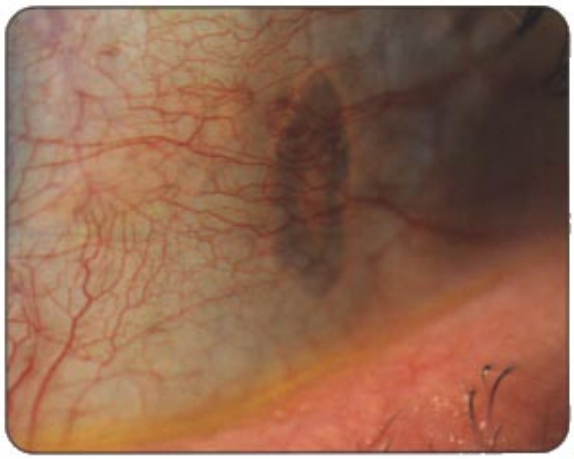


Fig. 4.2

Diffuse

Yellow

Causes

- Jaundice.
- Age-related – caused by subconjunctival fat deposition in the elderly.

Blue

Causes

1. Blue sclera (**Fig. 4.3**) occurs in osteogenesis imperfecta types I and II. Patients suffering from type II are stillborn or die in early infancy.
2. Keratoglobus associated with hyperextensible joints and dental anomalies.
3. Syndromes:
   - Marshall–Smith.
   - Russell–Silver.
   - Roberts.
   - Ehlers–Danlos (usually type I).
   - Marfan.
   - Hallermann–Streiff.
   - Bloch–Sulzberger (incontinentia pigmenti).
   - Turner.

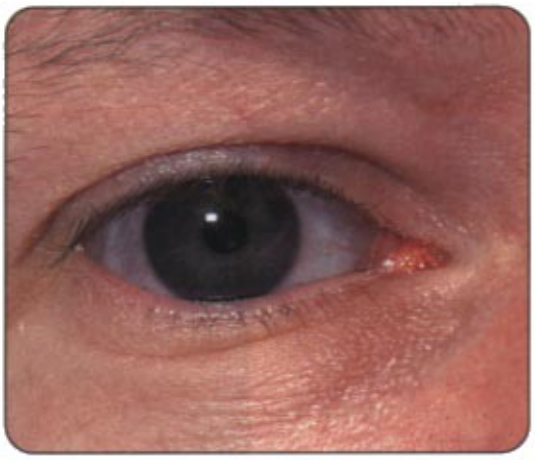


Fig. 4.3

SCLERAL AND EPISCLERAL INFLAMMATION

Not associated with a nodule

Simple episcleritis

Signs

- Sectorial (**Fig. 4.4**) or diffuse (**Fig. 4.5**) hyperemia, primarily involving the middle episcleral plexus, with some secondary involvement of the overlying conjunctival vessels.
- Topical phenylephrine will cause blanching and enhance visualization of the normal deep vascular plexus adjacent to the sclera.

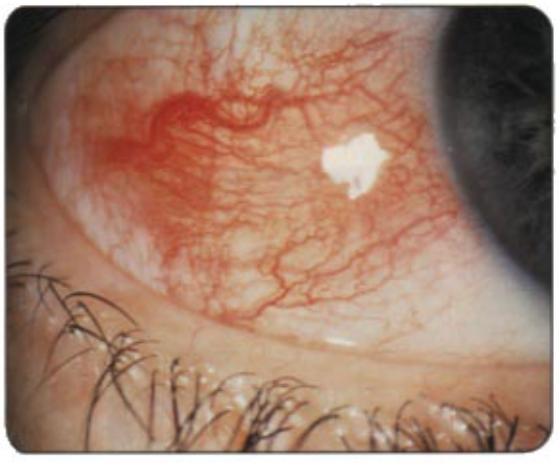


Fig. 4.4

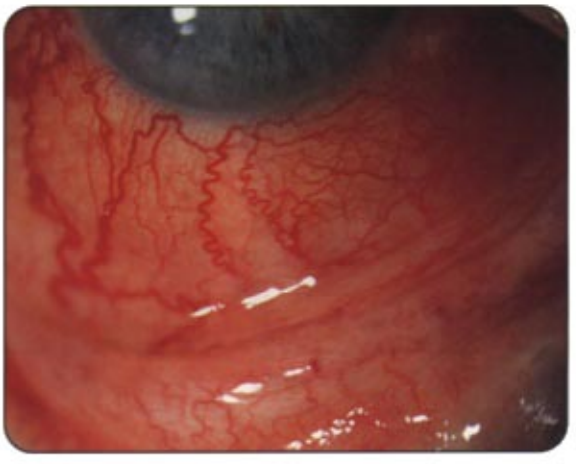


Fig. 4.5

Diffuse scleritis

Signs

- Diffuse hyperemia (**Fig. 4.6**) and distortion of the pattern of the deep vascular plexus, associated with variable episcleral and conjunctival injection.

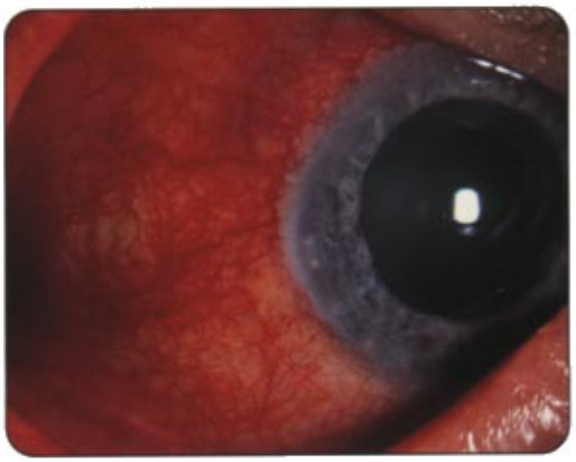


Fig. 4.6

Associated with a nodule

Nodular episcleritis

Signs

- Tender, usually solitary, localized injected nodule which can be moved over the sclera.

Nodular non-necrotizing scleritis

Signs

- Tender, usually solitary, localized injected nodule which cannot be moved over the sclera (**Fig. 4.7**).

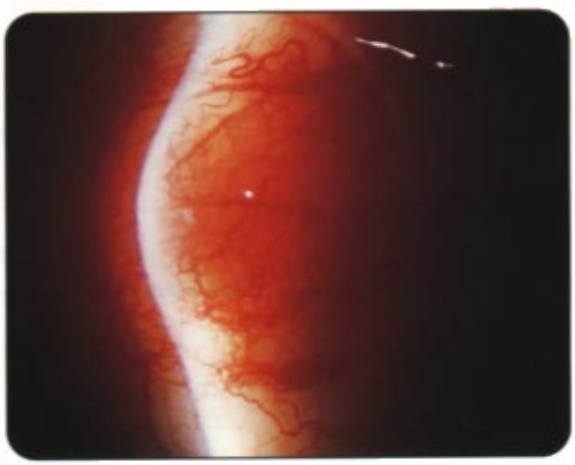


Fig. 4.7

SCLERAL NECROSIS

Necrotizing scleritis with inflammation

Signs (in chronological order)

- Vascular distortion or occlusion of the deep plexus.
- Appearance of painful avascular patches (**Fig. 4.8**).
- Scleral necrosis (**Fig. 4.9**).
- The underlying uvea becomes visible to a variable degree through thinned and necrotic sclera (**Figs 4.10–4.12**).

Complications

- Uveitis.
- Cataract.
- Keratitis (**Fig. 4.13**).
- Glaucoma.
- Vitreous prolapse.

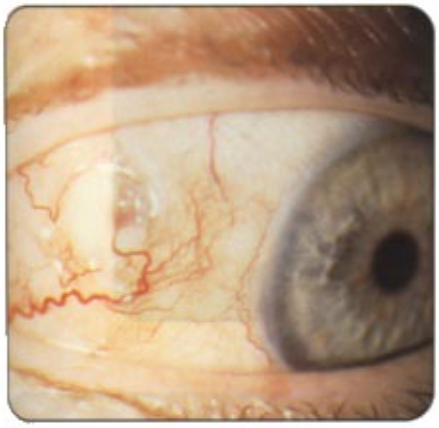


Fig. 4.8

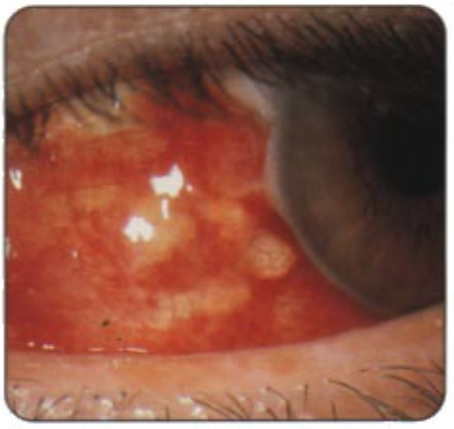


Fig. 4.9

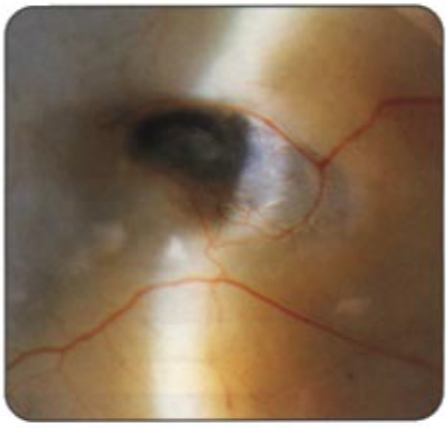


Fig. 4.10

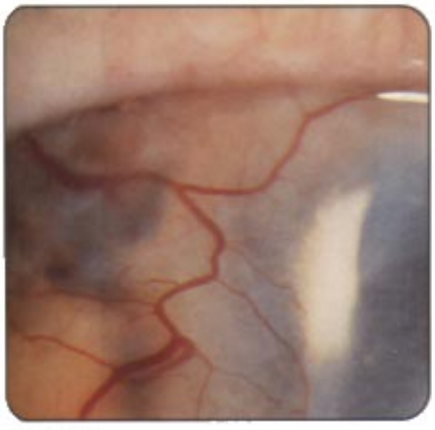


Fig. 4.11

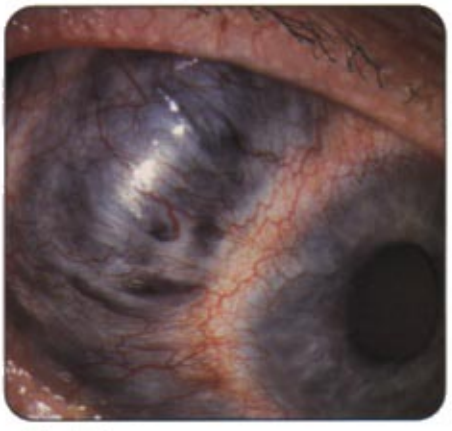


Fig. 4.12

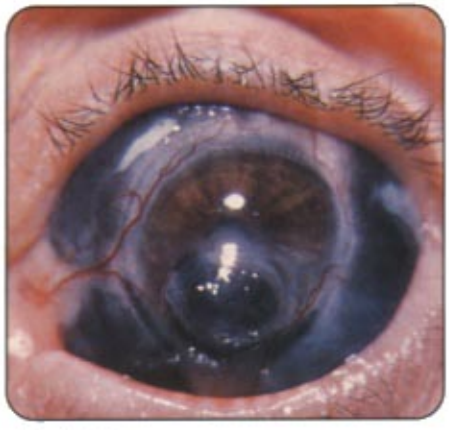


Fig. 4.13

Without inflammation (scleromalacia perforans)

Necrotizing scleritis without inflammation is classically seen in patients with longstanding rheumatoid arthritis.

Signs (in chronological order)

- Asymptomatic appearance of enlarging white patches of scleral necrosis.
- Exposure of large areas of underlying uvea (**Figs 4.14, 4.15**).

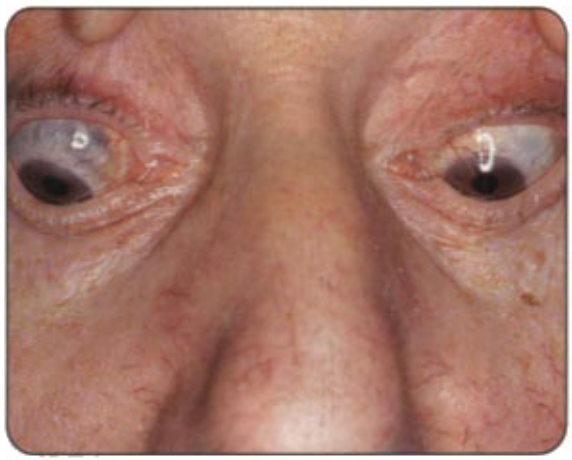


Fig. 4.14

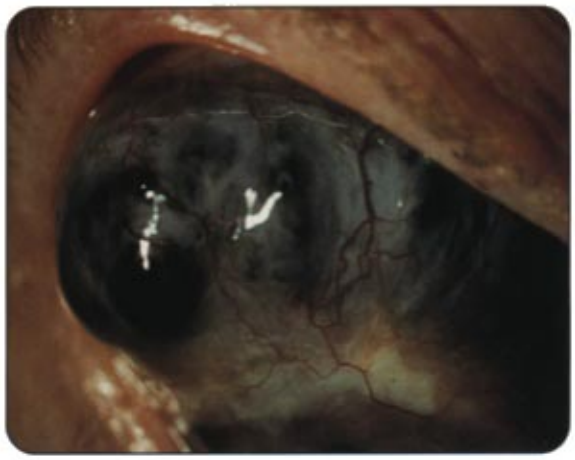


Fig. 4.15

POSTERIOR SCLERITIS

Signs

The signs of posterior scleritis are variable and depend primarily on the site of maximal involvement.

1. Associated anterior scleritis – this is present in about 80% of cases.
2. Raised intraocular pressure – this is caused by angle closure secondary to anterior shift of the iris–lens diaphragm in association with ciliochoroidal detachment.
4. Orbital signs:
   - Inflammation.
   - Mass giving rise to proptosis.
4. Defective ocular motility.
5. Vitritis.
6. Disc edema.
7. Choroid:
   - Ring detachment.
   - Chorioretinal folds – more common in men.
8. Retina:
   - Macular edema.
   - Exudative detachment – more common in young women.
   - Intraretinal deposits and subretinal exudation (**Fig. 4.16**).
   - Mass lesion – more common in middle-aged women.

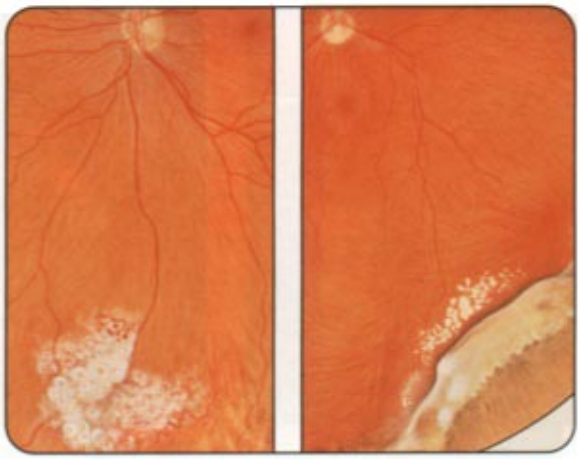


Fig. 4.16

Table 4.1 Causes and associations of scleritis

Idiopathic	**Systemic vasculitides**
	• Wegener granulomatosis (common)
Surgically induced	• polyarteritis nodosa
• diffuse	• giant-cell arteritis
• necrotizing	• Takayasu disease
	• Behçet disease
Rheumatic disorders	
• rheumatoid arthritis (common)	**Granulomatous diseases**
• relapsing polychondritis	• sarcoidosis
• juvenile rheumatoid arthritis	• tuberculosis
• juvenile chronic arthritis	• syphilis
	• Lyme disease
Connective tissue diseases	• Churg–Strauss syndrome
• systemic lupus erythematosus	
• polymyositis	**Skin diseases**
• IgA nephropathy	• herpes zoster ophthalmicus (common)
	• acne rosacea
Enteropathies	• porphyria
• Crohn disease	• pyoderma gangrenosum
• ulcerative colitis	

5 The cornea

ABNORMAL CORNEAL SIZE 115

Small cornea
- Microcornea
- Sclerocornea
- Nanophthalmus
- Microphthalmus

Large cornea
- Megalocornea
- Bulphthalmus
- Pseudo-large cornea

ABNORMAL CORNEAL SHAPE 119

- Keratoconus
- Keratoglobus
- Pellucid marginal degeneration
- Cornea plana
- Keratectasia

EPITHELIAL LESIONS 122

Epithelial microcysts
- Epithelial edema
- Cogan microcystic dystrophy
- Meesmann dystrophy

Punctate epithelial erosions

Epithelial keratitis
- Punctate epithelial keratitis
- Superficial punctate keratitis (Thygeson disease)

Filamentary keratitis
- Keratoconjunctivitis sicca
- Superior limbic keratoconjunctivitis
- Miscellaneous causes of filamentary keratitis

Large epithelial defects
- Corneal abrasion
- Herpes simplex geographic ulcer
- Neurotrophic keratopathy
- Shield ulcer in vernal keratoconjunctivitis
- Bacterial keratitis
- Acanthamoeba keratitis

Linear and branch-shaped epithelial lesions
- Herpes simplex dendritic ulcer
- Herpes zoster pseudodendrites
- Acanthamoeba pseudodendrites
- Miscellaneous causes of pseudodendrites

Epithelial iron deposits

Vortex keratopathy

LESIONS OF BOWMAN LAYER AND ANTERIOR STROMA 132

Focal inflammatory lesions
- Punctate subepithelial keratitis
- Nummular keratitis

PERIPHERAL LESIONS 154

Noninflammatory
- Vogt white limbal girdle
- Corneal arcus
- Posterior embryotoxon
- Pseudogerontoxon in vernal disease (anterior embryotoxon)

Inflammatory peripheral infiltrates
- Marginal keratitis
- Rosacea keratitis
- Chlamydial keratitis
- Phlyctenulosis
- Associated with exposure
- Sclerosing keratitis
- Acute stromal keratitis

Degenerations
- Band keratopathy
- Spheroidal degeneration (Labrador keratopathy)
- Lipid keratopathy
- Salzmann nodular degeneration

Dystrophies
- Reis–Bucklers dystrophy
- Anterior membrane (Grayson–Wilbrandt dystrophy)

STROMAL LESIONS 136

Noninflammatory stromal edema

Inflammatory stromal lesions
- Disciform keratitis
- Herpes simplex necrotic stromal keratitis
- Bacterial keratitis
- Candida keratitis
- Filamentous keratitis
- Acanthamoeba keratitis
- Interstitial keratitis

Stromal dystrophies
- Cornea farinata
- Crocodile shagreen
- Granular dystrophy
- Macular dystrophy
- Lattice dystrophy
- Granular-lattice (Avellino) dystrophy
- Schnyder central crystalline dystrophy

Stromal crystals
- Infectious cystalline keratopahthy
- Systemic causes of corneal crystals

Prominent corneal nerves

Diffuse corneal clouding at birth

LESIONS OF DESCEMET MEMBRANE 148

- Tears
- Folds and wrinkles
- Detachment
- Kayser–Fleischer ring

ENDOTHELIAL LESIONS 150

Keratic precipitates
- Small KP
- Mutton-fat KP
- Old KP
- Linear KP

Pigment deposits
- Nonspecific pigment dusting
- Krukenberg spindle
- Chlorpromazine

Dystrophies
- Fuchs endothelial dystrophy
- Posterior polymorphous dystrophy

Peripheral thinning not associated with systemic disease
- Furrow degeneration
- Dellen
- Terrien marginal degeneration
- Mooren ulcer
- Fuchs superficial marginal keratitis

Peripheral thinning associated with systemic disease
- Rheumatoid arthritis
- Polyarteritis nodosa and Wegener granulomatosis

ABNORMAL CORNEAL SIZE

Small cornea

Microcornea

Microcornea is a very rare, congenital, unilateral or bilateral condition. Inheritance is autosomal dominant or recessive.

Signs

- Adult horizontal corneal diameter is 10 mm or less (**Figs 5.1, 5.2**).
- Shallow anterior chamber.
- Other ocular dimensions are normal.

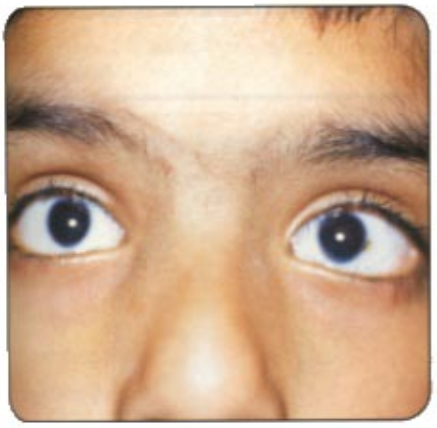


Fig. 5.1

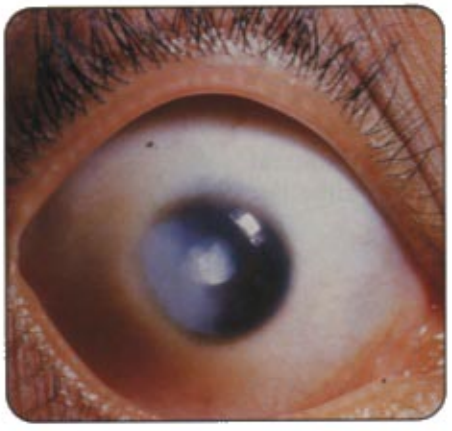


Fig. 5.2

Table 5.1 Associations of microcornea

Ocular
• cornea plana
• corneal leukoma
• iris abnormalities
• microphakia
• congenital cataract
• glaucoma
Systemic syndromes
• Turner
• Ehlers–Danlos
• Weill–Marchesani
• Waardenburg
• Nance–Horan
• Cornelia de Lange

Sclerocornea

Sclerocornea is a rare, congenital, usually bilateral condition.

Signs

- Opacification and vascularization of the peripheral or entire cornea (**Fig. 5.3**). If only the peripheral cornea is involved, the resulting 'scleralization' makes the cornea appear smaller than normal (**Fig. 5.4**).

Ocular associations

- Microphthalmos.
- Blue sclera.
- Cornea plana.
- Iris abnormalities.
- Congenital cataract.
- Glaucoma.

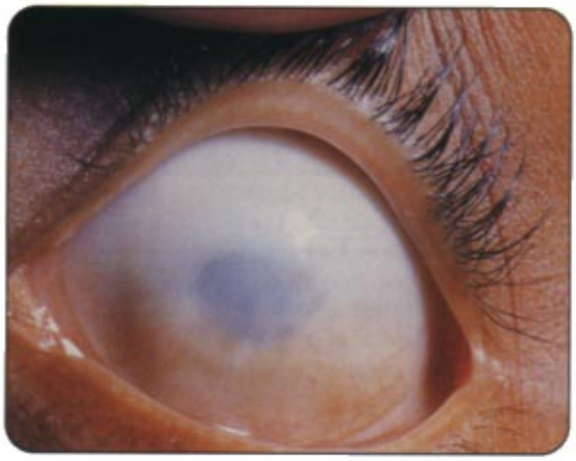


Fig. 5.3

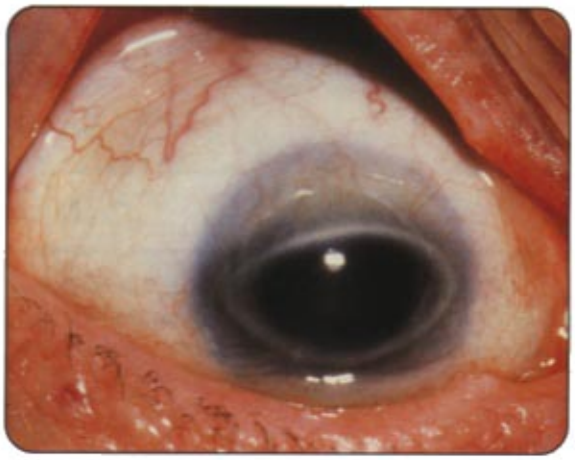


Fig. 5.4

Nanophthalmos

Nanophthalmos is an uncommon, congenital, bilateral condition in which the globes have reduced volume but are otherwise grossly normal.

Fig. 5.5

Signs

- Very high hypermetropia (e.g. +12D) (**Fig. 5.5**).
- Adult corneal diameter is reduced but the lens has a normal volume.
- Short axial length averaging 18 mm.
- Shallow anterior chamber.
- Thick sclera.
- Fundus may show a crowded disc, vascular tortuosity, and macular hypoplasia.

Late problems

- Angle-closure glaucoma.
- Uveal effusion.
- Retinal detachment.
- Poorly tolerated intraocular surgery.

Microphthalmos

Microphthalmos is an uncommon, unilateral or bilateral condition in which the axial length of the eye is reduced. The effects on vision depend on its severity and the presence of associated anomalies. The two types of microphthalmos are noncolobomatous (**Fig. 5.6** and **Table 5.2**) and colobomatous (**Fig. 5.7** and **Table 5.3**).

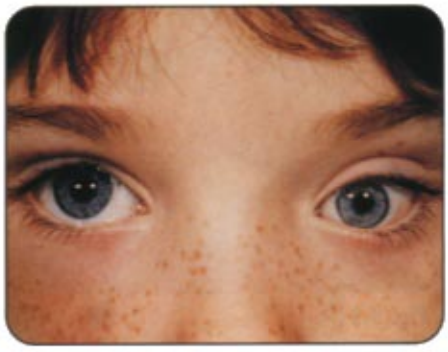


Fig. 5.6

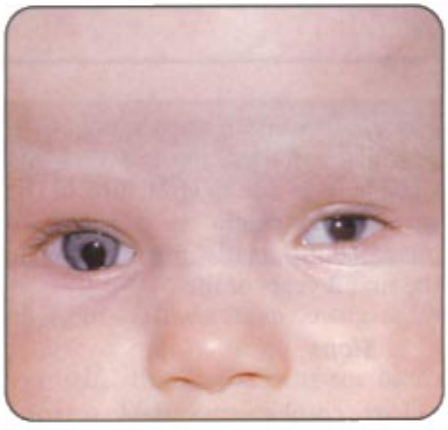


Fig. 5.7

Table 5.2 Classification of noncolobomatous microphthalmos

Isolated
• sporadic
• inherited (dominant, recessive, X-linked recessive)
With anterior persistent hyperplastic primary vitreous
Microphthalmos with cyst
Intrauterine infections
• rubella
• toxoplasmosis
• cytomegalovirus
• varicella

Table 5.3 Classification of colobomatous microphthalmos

Isolated
• sporadic
• inherited (dominant)
With systemic syndromes
• Patau (trisomy 13)
• Edward (trisomy 18)
• cat-eye (partial trisomy 22)
• Pagon (CHARGE)
• Meckel–Gruber
• Lenz microphthalmos
• Temple–Al Gazali
• Delleman
• Golz focal dermal hypoplasia

Large cornea

Megalocornea

Megalocornea is a rare, congenital, bilateral (**Fig. 5.8**) condition which is usually inherited in an X-linked recessive manner.

Signs

- Clear cornea with a horizontal diameter of 13 mm or more (**Fig. 5.9**).
- Very deep anterior chamber (**Fig. 5.10**).
- Normal intraocular pressure.
- High myopia and astigmatism, but good visual acuity.
- Lens subluxation may occur as a result of zonular stretching.

Systemic associations

- Marfan syndrome.
- Apert syndrome.
- Ehlers–Danlos syndrome.
- Down syndrome.
- Osteogenesis imperfecta.
- Progressive facial hemiatrophy.
- Renal carcinoma.
- Mental retardation.

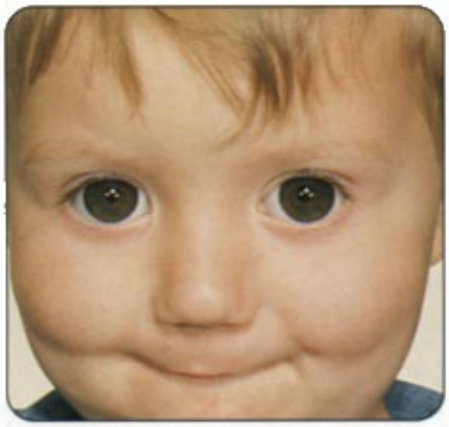


Fig. 5.8

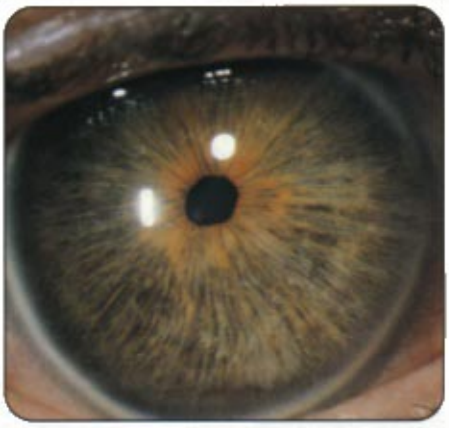


Fig. 5.9

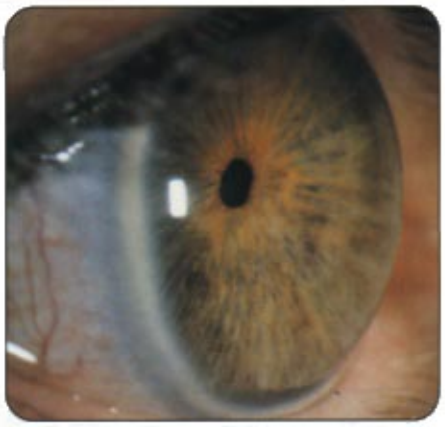


Fig. 5.10

Buphthalmos

Buphthalmos is an uncommon, usually bilateral condition in which the globe is large due to stretching of the cornea and sclera as a result of increased intraocular pressure during the first 3 years of life.

Signs

- Large cornea with variable scarring (**Fig. 5.11**).
- Very deep anterior chamber.
- Angle anomalies.
- Horizontal ruptures in Descemet membrane (Haab striae).
- Myopia.
- Optic disc cupping.

Figure 5.12 shows left buphthalmos in a boy with Sturge–Weber syndrome.

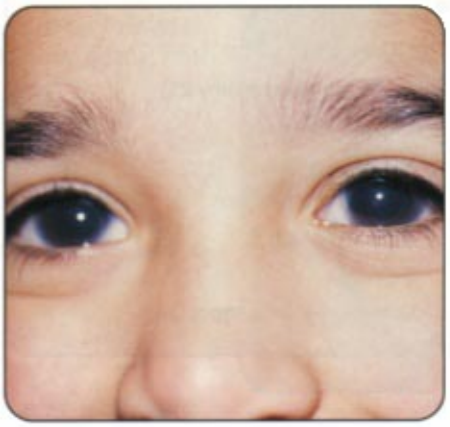


Fig. 5.11

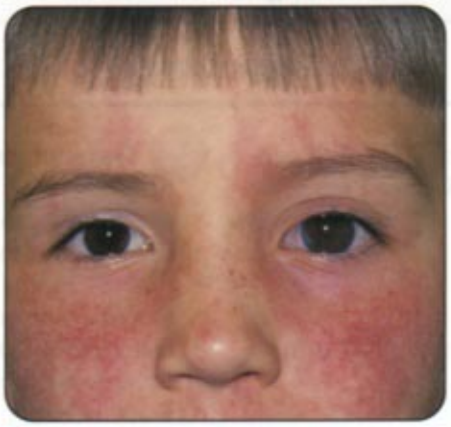


Fig. 5.12

Table 5.4 Associations of infantile glaucoma

Ocular
• Axenfeld–Rieger anomaly
• aniridia
• Peters' anomaly
• congenital ectropion uveae
Systemic
• Lowe syndrome
• Sturge–Weber syndrome
• Down syndrome
• neurofibromatosis type I
• Rieger syndrome
• Rubinstein–Taybi syndrome
• Pierre Robin syndrome
• Patau syndrome (trisomy 13)
• nevus of Ota
• cutis marmorata telangiectasia congenita
• mucopolysaccharidoses

Pseudo-large cornea

Pseudo-large cornea is the false appearance of a large cornea in a patient with microcephaly (small head) (**Fig. 5.13**).

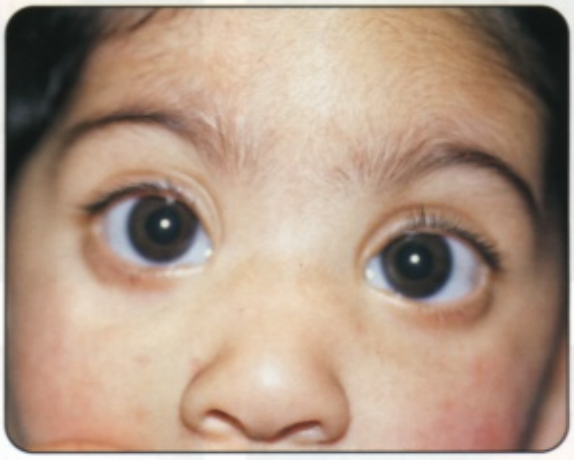


Fig. 5.13

ABNORMAL CORNEAL SHAPE

Keratoconus

Keratoconus is a common, acquired condition which is bilateral in 85% of cases, although the severity of involvement may be asymmetric.

Signs

- Increasing myopia and astigmatism.
- Abnormal 'oil droplet' red reflex (**Fig. 5.14**).
- Scissors reflex on retinoscopy.
- Central or paracentral stromal thinning with inferior apical protrusion (**Fig. 5.15**).
- Munson sign – bulging of the lower lid in downgaze (**Fig. 5.16**).
- Fleischer ring – epithelial iron deposits at the base of the cone.
- Vogt striae – fine deep vertical stromal stress lines which temporarily disappear on digital pressure (**Fig. 5.17**).
- Rizutti sign – conical reflection on the nasal cornea when light is shone from the temporal side (**Fig. 5.18**).
- Prominent corneal nerves.
- Acute hydrops – corneal edema resulting from tears in Descemet membrane.
- Variable corneal scarring, depending on severity (**Figs 5.19–5.21**).

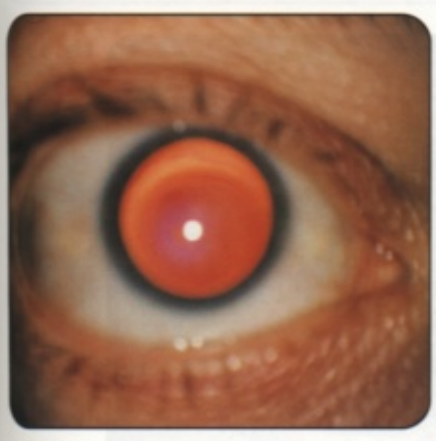


Fig. 5.14

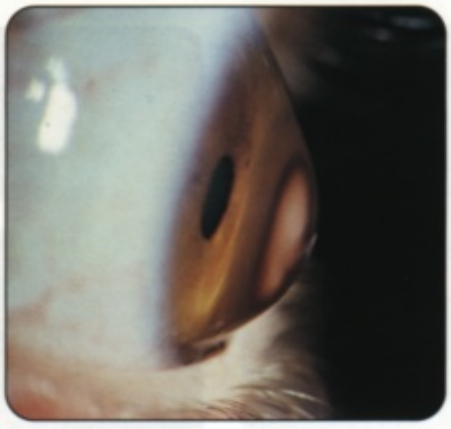


Fig. 5.15

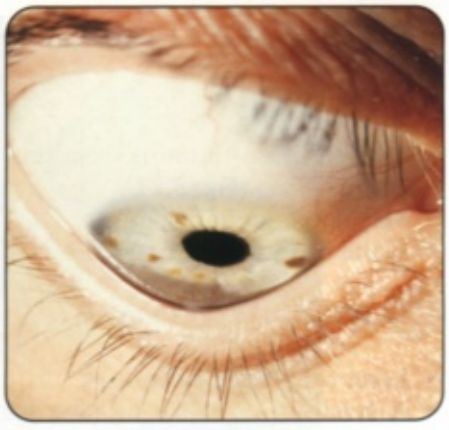


Fig. 5.16

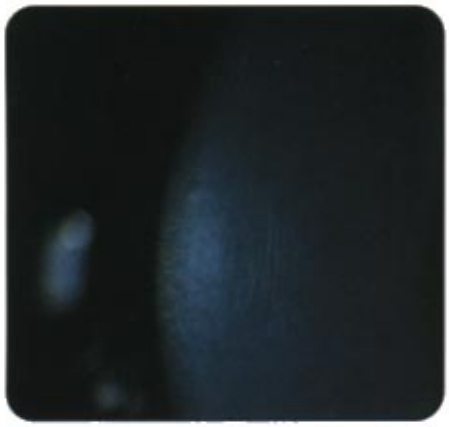


Fig. 5.17

Fig. 5.18

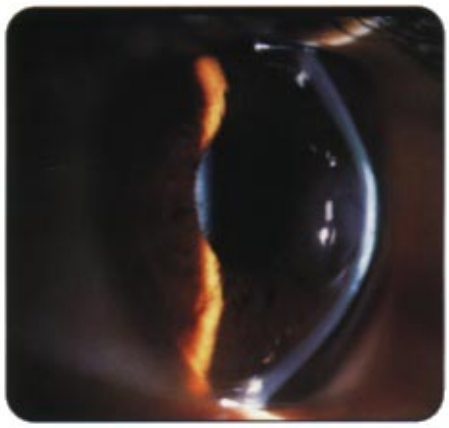


Fig. 5.19

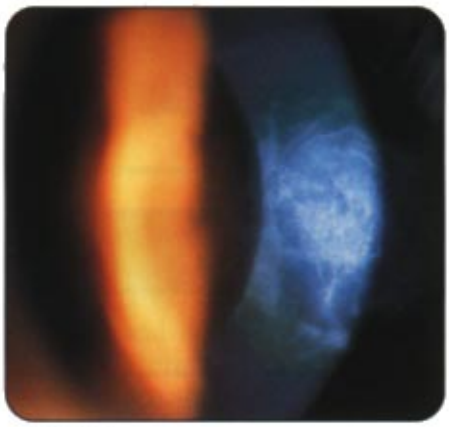


Fig. 5.20

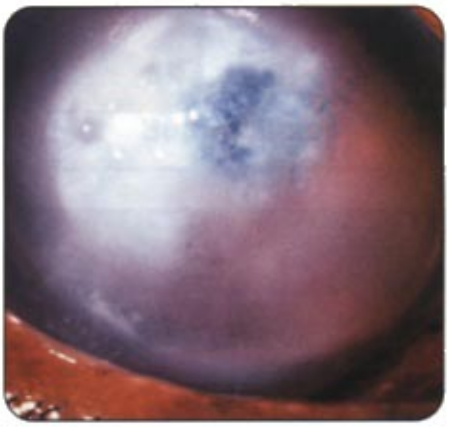


Fig. 5.21

Table 5.5 Associations of keratoconus

Ocular
• vernal disease
• blue sclera
• prominent corneal nerves
• aniridia
• ectopia lentis
• retinitis pigmentosa
• Leber congenital amaurosis
Systemic
• Down syndrome
• Ehlers–Danlos syndrome
• Marfan syndrome
• Apert syndrome
• atopic dermatitis
• osteogenesis imperfecta
• mitral valve prolapse

Keratoglobus

Keratoglobus is a very rare, congenital, bilateral condition.

Signs

- Midperipheral thinning, resulting in protrusion of the entire cornea (**Fig. 5.22**).
- Very deep anterior chamber.
- Acute hydrops may occur in advanced cases.

Systemic associations

- Leber congenital amaurosis.
- A syndrome comprising blue sclera, hyperextensible joints, and dental abnormalities.

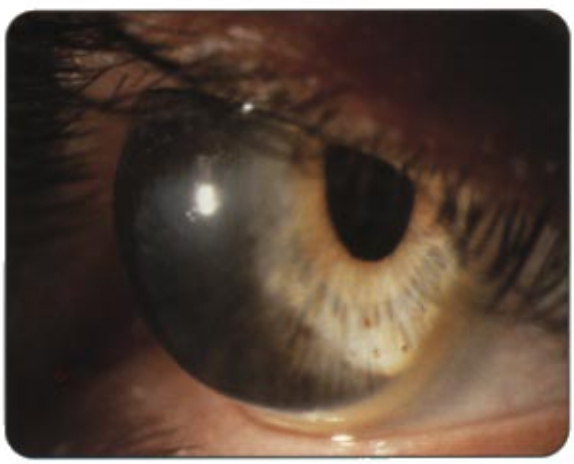


Fig. 5.22

Pellucid marginal degeneration

Pellucid marginal degeneration is an uncommon, acquired, bilateral condition.

Signs

- High astigmatism.
- Inferior, crescent-shaped area of peripheral corneal thinning, 1–2 mm in width, extending from the 4 to 8 o'clock positions, which is separated from the limbus by normal cornea (**Fig. 5.23**).
- Protrusion located above the area of thinning.

Fig. 5.23

Cornea plana

Cornea plana is a rare, congenital, bilateral condition which never occurs in isolation.

Signs

- Hypermetropia.
- Severe decrease in corneal curvature (**Fig. 5.24**).
- Shallow anterior chamber.
- Glaucoma.

Ocular associations

- Microcornea and sclerocornea.
- Microphthalmos.
- Peters anomaly.

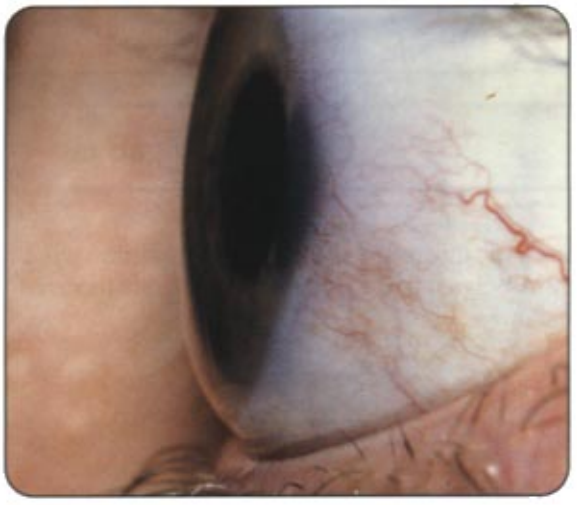


Fig. 5.24

Keratectasia

Keratectasia is a very rare, congenital, usually unilateral condition that is probably due to intrauterine keratitis and perforation.

Signs

- Severe corneal opacification and protruberance beyond the eyelids (**Fig. 5.25**).

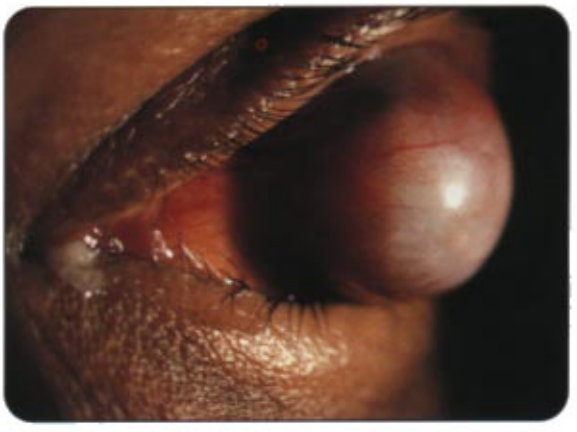


Fig. 5.25

EPITHELIAL LESIONS

Epithelial microcysts

Epithelial edema

Signs

- Loss of normal corneal luster (**Fig. 5.26**) which may be associated with vesicles and bullae (**Fig. 5.27**). It is an important sign of endothelial cell dysfunction and of epithelial hypoxia associated with inappropriate contact lens use.

Fig. 5.26

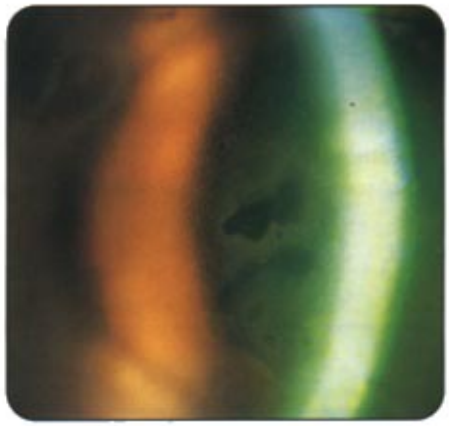


Fig. 5.27

Cogan microcystic dystrophy

Cogan microcystic dystrophy is a common condition which is frequently associated with bilateral spontaneous recurrent corneal erosion.

Signs

- Variety of microcystic (**Fig. 5.28**), dots (**Fig. 5.29**), and fingerprint-like or map-like (**Fig. 5.30**) epithelial lesions, which may occur singly or in various combinations.

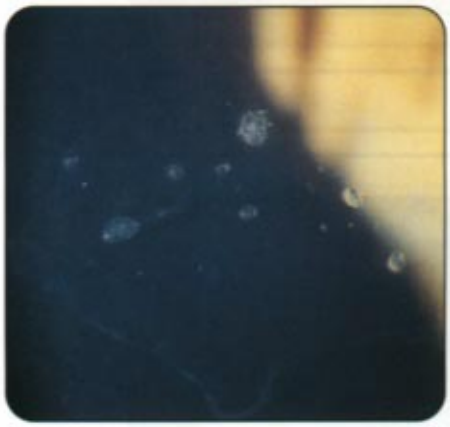


Fig. 5.28

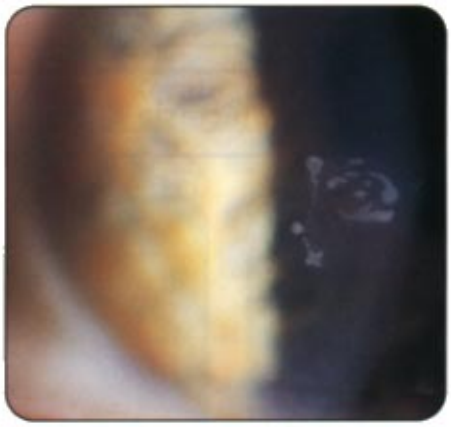


Fig. 5.29

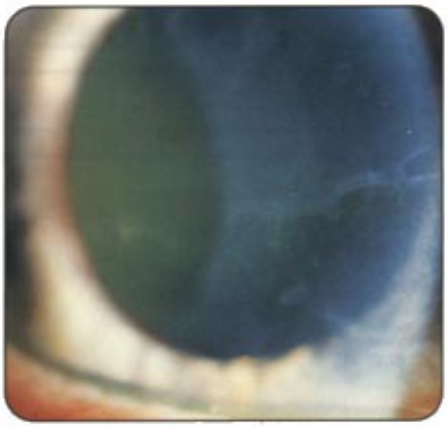


Fig. 5.30

Meesmann dystrophy

Meesmann dystrophy is a very rare, innocuous, dominantly inherited condition.

Signs

- Tiny epithelial cysts extending to the limbus and most numerous in the interpalpebral region. The lesions appear gray in direct illumination (**Fig. 5.31**) but clear in retroillumination.

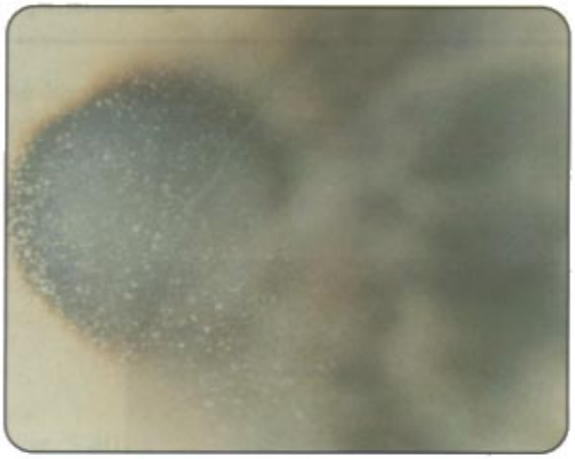


Fig. 5.31

Punctate epithelial erosions

Punctate epithelial erosions (PEE) are a very common nonspecific finding seen in a wide varity of corneal disorders.

Signs

- Tiny, slightly depressed, gray–white spots which represent areas of epithelial discontinuity (**Figs 5.32, 5.33**). They stain well with fluorescein (**Fig. 5.34**) but not with rose bengal.

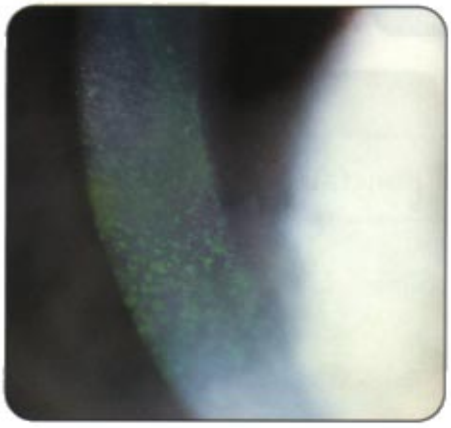


Fig. 5.32

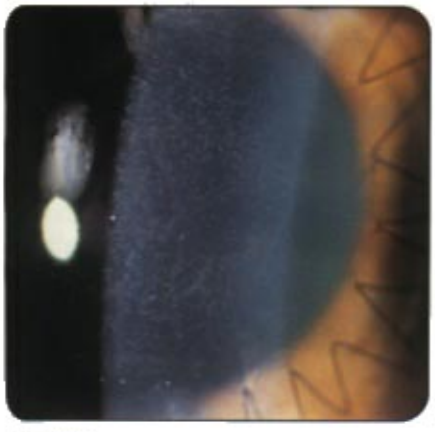


Fig. 5.33

Fig. 5.34

Table 5.6 Causes of punctate epithelial erosions

Superior
• subtarsal foreign body
• atopic keratoconjunctivitis
• superior limbic keratoconjunctivitis
• poorly fitting contact lens
• floppy eyelids
Interpalpebral
• dry eyes
• diminished corneal sensation
• exposure to ultraviolet light
Inferior
• lower lid margin lesions
• lagophthalmos
• rosacea
• toxicity from drops
• self-induced

Epithelial keratitis

Punctate epithelial keratitis

Punctate epithelial keratitis is a common, unilateral or bilateral keratitis caused by adenoviruses or herpes zoster.

Signs

- Granular epithelial opacities (**Figs 5.35, 5.36**) which stain well with rose bengal but poorly with fluorescein.

Fig. 5.35

Fig. 5.36

Superficial punctate keratitis (Thygeson disease)

Superficial punctate keratitis is an uncommon, usually bilateral, idiopathic condition.

Signs

- Stellate, round, or oval conglomerates of distinct grayish-white intraepithelial dots (**Fig. 5.37**), some of which may be associated with a mild subepithelial haze. The conjunctiva is uninvolved.

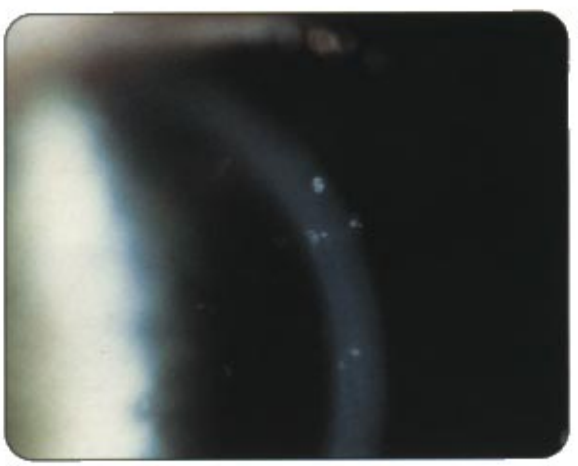


Fig. 5.37

Filamentary keratitis

Keratoconjunctivitis sicca

Keratoconjunctiva sicca is a very common, bilateral condition which may occur in isolation or in association with a systemic disorder.

Signs

- Small, mucous filaments attached to the corneal surface which stain well with rose bengal (**Fig. 5.38**) and with fluorescein (**Fig. 5.39**). Beneath their attachments to the epithelium, gray subepithelial opacities are seen.
- Mucous plaques.
- Decreased marginal tear strip.
- Reduced tear film break-up time.
- Staining of the interpalpebral conjunctiva with rose bengal (**Fig. 5.40**).

Table 5.7 Systemic associations of dry eyes

Autoimmune disorders
• rheumatoid arthritis
• juvenile rheumatoid arthritis
• systemic lupus erythematosus
• scleroderma
• polymyositis and dermatomyositis
• psoriatic arthritis
• primary biliary cirrhosis
• Hashimoto thyroiditis
Infiltrative disorders
• sarcoidosis
• lymphoma
• hemochromatosis
• amyloidosis

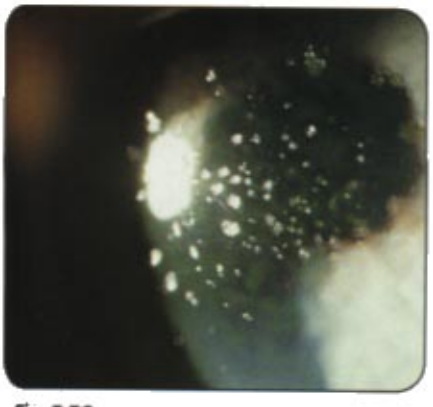


Fig. 5.38

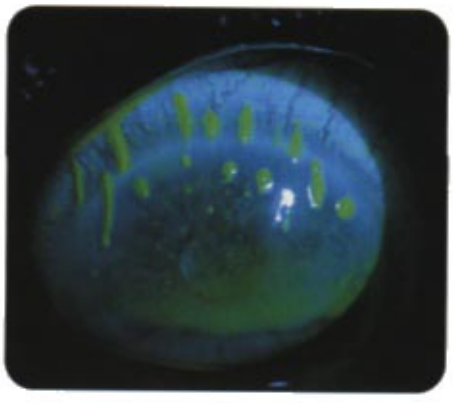


Fig. 5.39

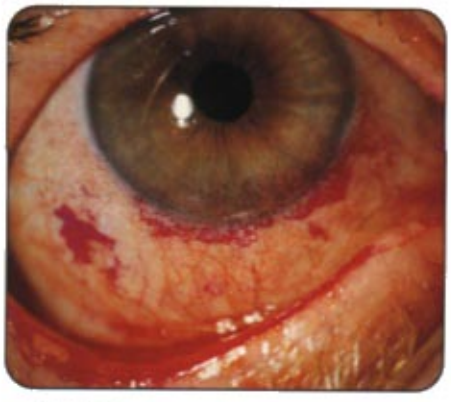


Fig. 5.40

Superior limbic keratoconjunctivitis

Superior limbic keratoconjunctivitis is an uncommon, usually bilateral condition which is frequently associated with thyroid dysfunction.

Signs

- Superior bulbar thickening and injection, and superior cornea filaments (Fig. 5.41).
- Superior tarsal papillary conjunctivitis.

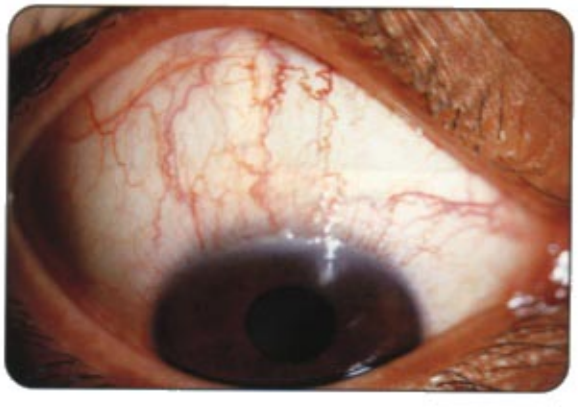


Fig. 5.41

Miscellaneous causes of filamentary keratitis

Table 5.8 Miscellaneous causes of filamentary keratitis

Ocular
• recurrent erosions
• eye patching
• exposure keratopathy
• diminished corneal sensation
• herpes zoster
Systemic
• essential blepharospasm
• midbrain strokes

Large epithelial defects

Corneal abrasion

Signs

- The epithelial defect can be seen with the naked eye (**Fig. 5.42**) but shows up more clearly with fluorescein (**Fig. 5.43**).

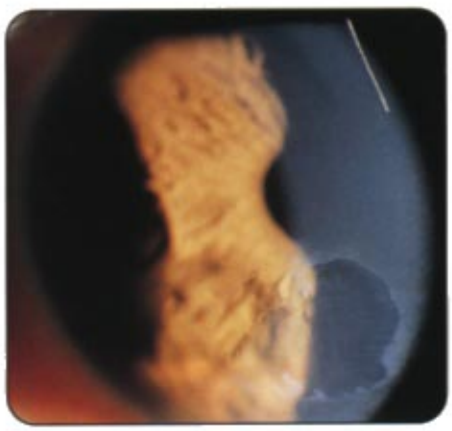


Fig. 5.42

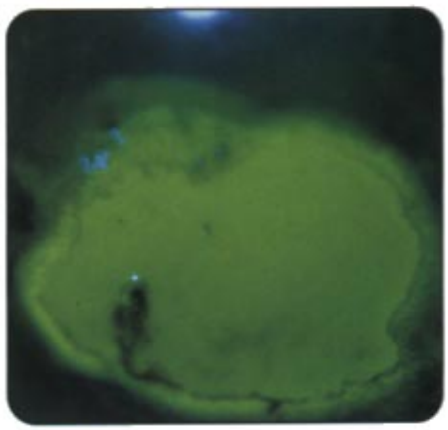


Fig. 5.43

Herpes simplex geographic ulcer

A geographic ulcer is an uncommon condition caused by the continued enlargement of a dendritic ulcer.

Signs

- Large, ameboid-like, fluorescein-staining epithelial defects (**Figs 5.44, 5.45**), the margins of which stain with rose bengal.

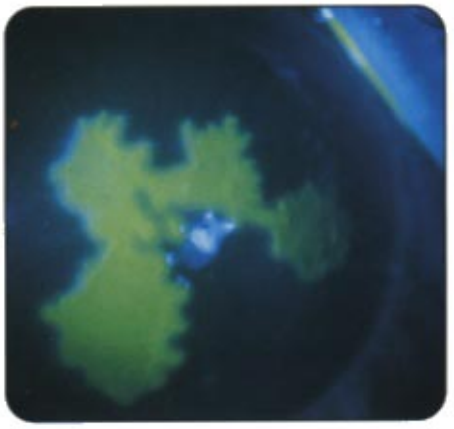


Fig. 5.44

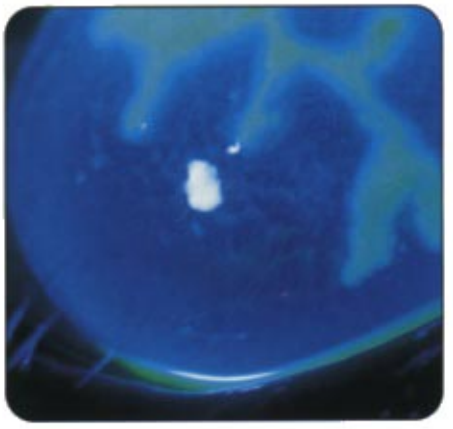


Fig. 5.45

Neurotrophic keratopathy

Neurotrophic keratopathy occurs in eyes with diminished corneal sensation. It is usually acquired but can rarely be congenital (Riley–Day syndrome).

Signs (in chronological order)

- Interpalpebral punctate epithelial erosions.
- Epithelial defects which are slow to heal (**Fig. 5.46**).
- Secondary infection (**Fig. 5.47**).
- Corneal perforation (**Fig. 5.48**) may occur in very advanced cases.

Acquired causes

- Previous herpetic disease.
- Section of the trigeminal nerve.
- Diabetes.
- Leprosy.

Fig. 5.46

Fig. 5.47

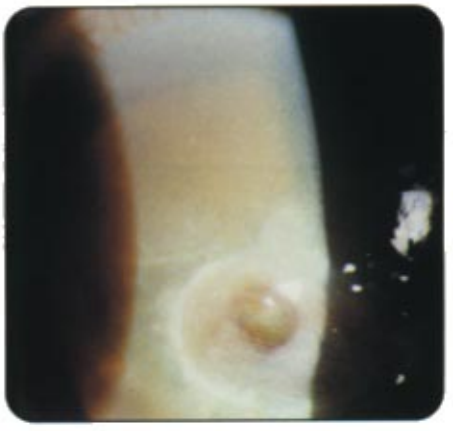


Fig. 5.48

Shield ulcer in vernal keratoconjunctivitis

A shield ulcer occasionally occurs in patients with severe vernal disease.

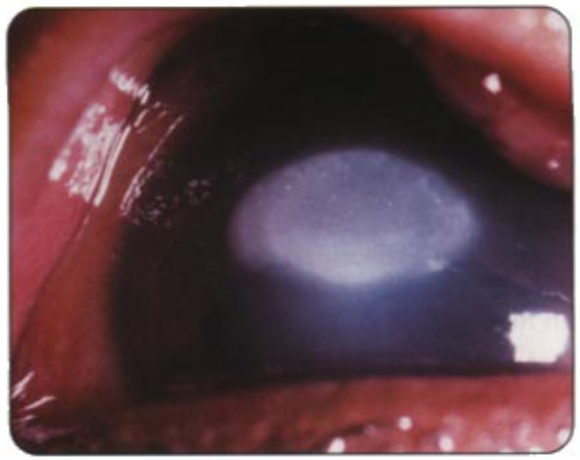


Fig. 5.49

Signs

- Superior, oval or pentagonal defect which may be associated with grayish opacification of the bed and slightly elevated margins (**Fig. 5.49**).

Look for associated

- Superior tarsal papillae.
- Limbal papillae (limbitis).
- Small discrete white spots at the limbus (Trantas dots).
- Pseudogerontoxon.

Bacterial keratitis

Bacterial keratitis typically develops in eyes with pre-existing ocular surface disease and in patients wearing contact lenses.

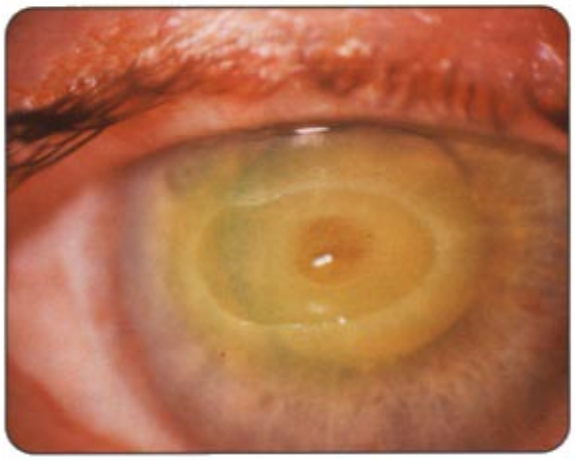


Fig. 5.50

Signs

- Epithelial breakdown which is frequently associated with stromal suppuration (**Fig. 5.50**).

Acanthamoeba keratitis

Acanthamoeba keratitis is a rare, painful, unilateral infection associated with the use of soft contact lenses.

Signs

- A central or paracentral, nonsuppurative stromal ring is frequently associated with variable epithelial breakdown (**Figs 5.51, 5.52**).

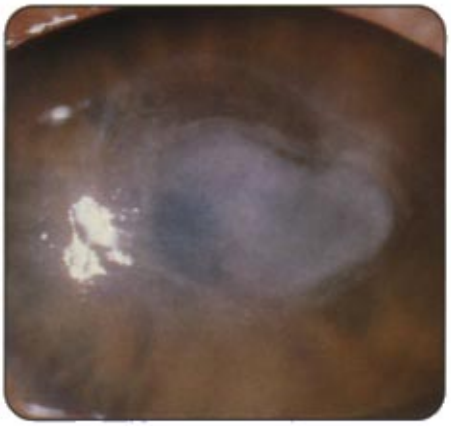


Fig. 5.51

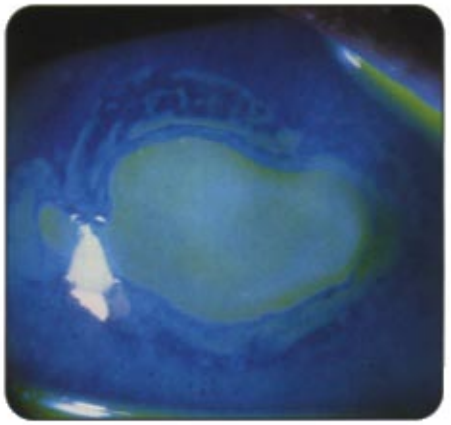


Fig. 5.52

Linear and branch-shaped epithelial lesions

Herpes simplex dendritic ulcer

A herpes simplex dendritic ulcer is a very common, unilateral condition.

Signs

- Single or – less commonly – multiple, branching, ulcerating epithelial lesions with raised edges and terminal bulb-like projections (**Fig. 5.53**).
- Associated anterior stromal haze is common.
- Corneal sensation may be diminished.
- The ulcer bed stains with fluorescein (**Figs 5.54, 5.55**).
- The swollen margins of the lesion stain with rose bengal (**Fig. 5.56**).

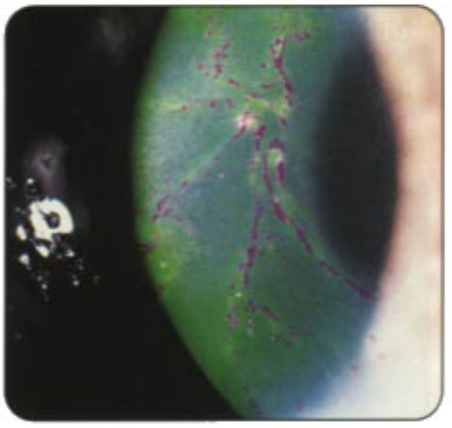


Fig. 5.53

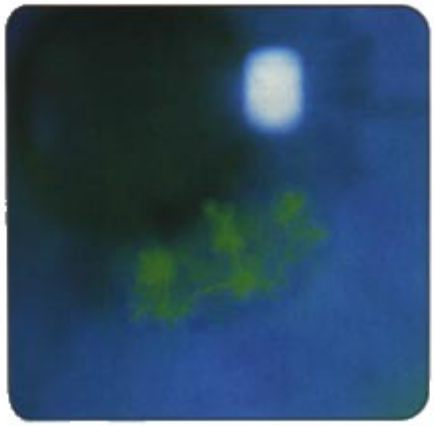


Fig. 5.54

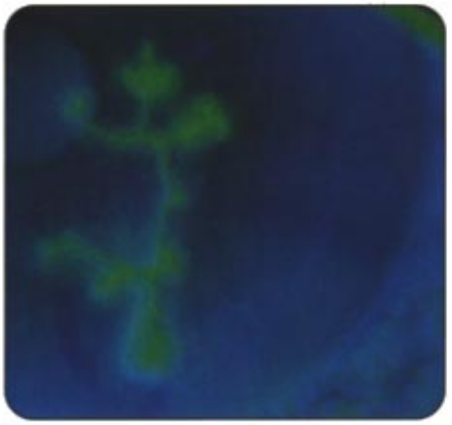


Fig. 5.55

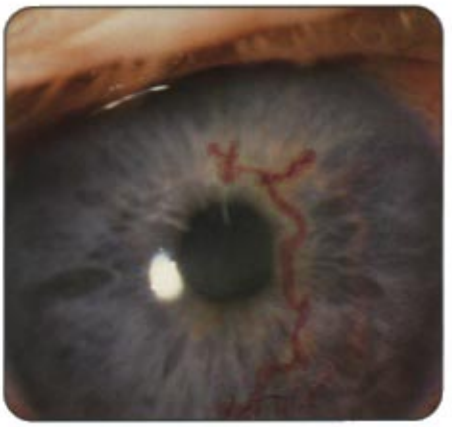


Fig. 5.56

Herpes zoster pseudodendrite

Herpes zoster pseudodendrite is a common, frequently multiple unilateral condition which occurs in association with herpes zoster ophthalmicus.

Signs

- Elevated epithelial lesions with tapered ends which lack terminal bulbs (**Fig. 5.57**). They are usually more peripheral than dendritic ulcers caused by herpes simplex.
- The entire lesion stains with rose bengal (**Fig. 5.58**).

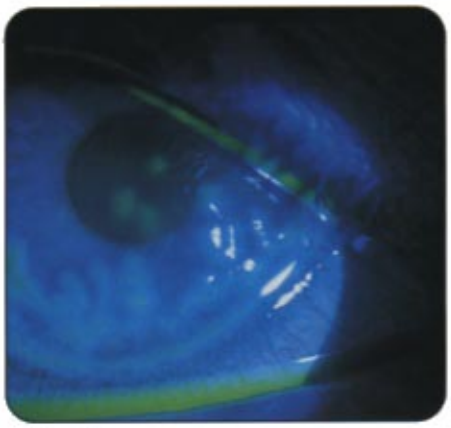


Fig. 5.57

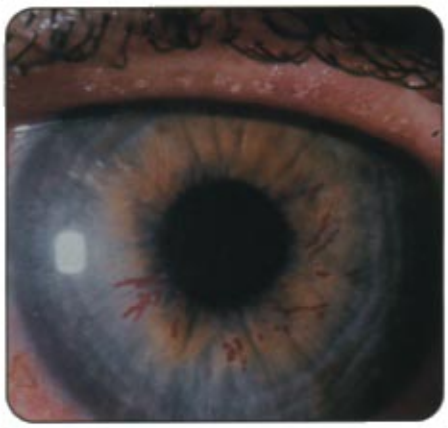


Fig. 5.58

Acanthamoeba pseudodendrite

Signs

- Multiple pseudodendrites may be seen seen in relatively early cases (**Figs 5.59, 5.60**).

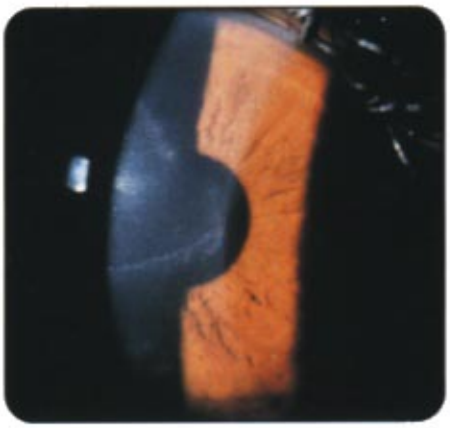


Fig. 5.59

Fig. 5.60

Miscellaneous causes of pseudodendrites

- Epithelial rejection line in a corneal graft (**Fig. 5.61**).
- Healing epithelial defects.
- Chronic epithelial toxicity.
- Use of soft contact lenses.
- Tyrosinemia II.

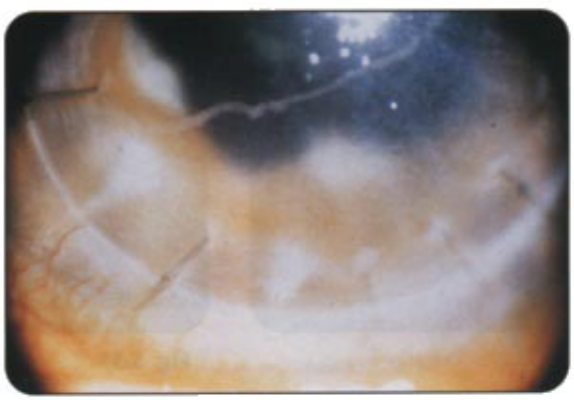


Fig. 5.61

Epithelial iron deposits

- Rust ring consists of residual rust following the removal of a metallic foreign body (**Fig. 5.62**).
- Hudson Stähli line occurs at the junction of the upper two-thirds and the lower third of an otherwise normal cornea (**Fig. 5.63**).
- Ferry line occurs in front of a filtering bleb.
- Stocker line occurs in front of a pterygium.
- Fleischer ring occurs at the base of the cone in keratoconus.
- Dalgleisch line is found in the inferior cornea in patients with hereditary spherocytosis.
- Other iron lines may be found adjacent to corneal elevations in Salzmann degeneration, corneal grafts, and radial keratotomy incisions.

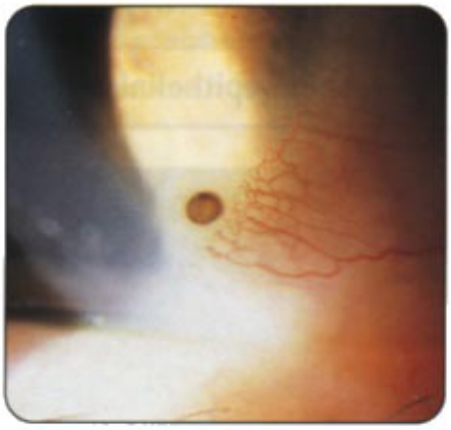


Fig. 5.62

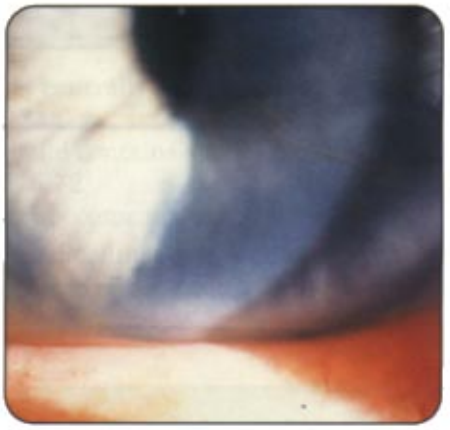


Fig. 5.63

Vortex keratopathy

Vortex keratopathy is a bilateral condition which occurs in patients with Fabry disease but is more commonly caused by a variety of drugs.

Signs

- Grayish or golden epithelial deposits arranged in a vortex fashion from a point below the pupil and swirling outward but sparing the limbus (**Figs 5.64, 5.65**).

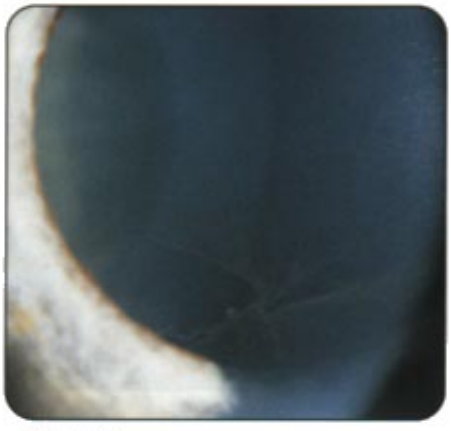


Fig. 5.64

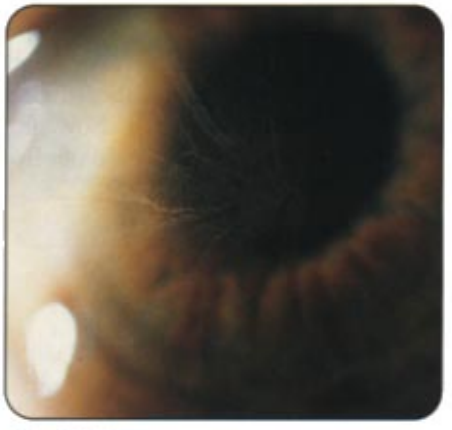


Fig. 5.65

Drugs causing vortex keratopathy

- Chloroquine.
- Hydroxychloroquine.
- Amiodarone.
- Indomethacin.
- Tamoxifen.
- Chlorpromazine.
- Mepacrine.
- Atovaquone.

LESIONS OF BOWMAN LAYER AND ANTERIOR STROMA

Focal inflammatory lesions

Punctate subepithelial keratitis

Signs

- Small, multiple, frequently bilateral subepithelial opacities (**Figs 5.66, 5.67**).

Causes

- Adenovirus infection.
- Epstein–Barr virus infection.
- Reiter disease.
- Epithelial corneal graft rejection in donor cornea.

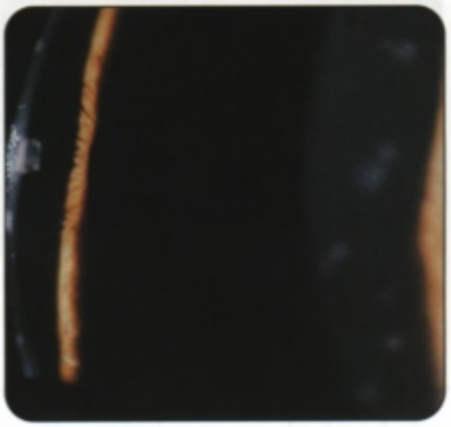


Fig. 5.66

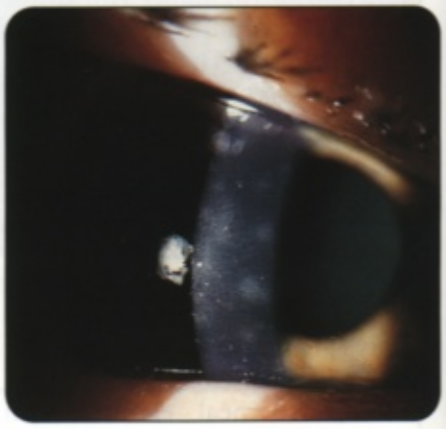


Fig. 5.67

Nummular keratitis

Signs

- Large, multiple, unilateral or bilateral, round, granular subepithelial deposits just beneath Bowman layer, surrounded by a halo of stromal haze (**Figs 5.68, 5.69**).

Causes

- Herpes zoster ophthalmicus.
- Epstein–Barr virus infection.
- Lyme disease.
- Onchocerciasis.
- Brucellosis.
- Dimmler nummular keratitis.

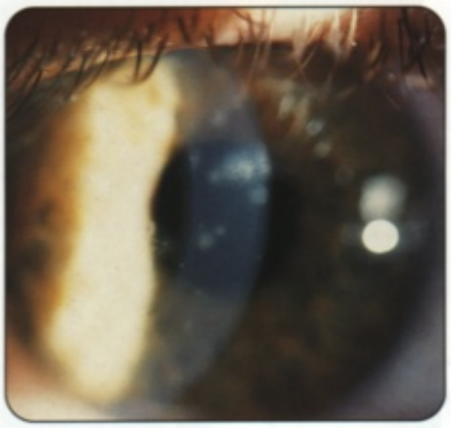


Fig. 5.68

Fig. 5.69

Degenerations

Band keratopathy

Band keratopathy is a common condition which, depending on the cause, may be bilateral or unilateral.

Signs (in chronological order)

- Peripheral interpalpebral calcification with clear cornea separating the sharp peripheral margins of the band from the limbus (**Fig. 5.70**).
- The lesion spreads centrally and has a ground-glass appearance (**Fig. 5.71**).
- Once formed, the band contains small holes and, occasionally, clefts (**Fig. 5.72**).
- Advance lesions may become nodular and elevated (**Fig. 5.73**).

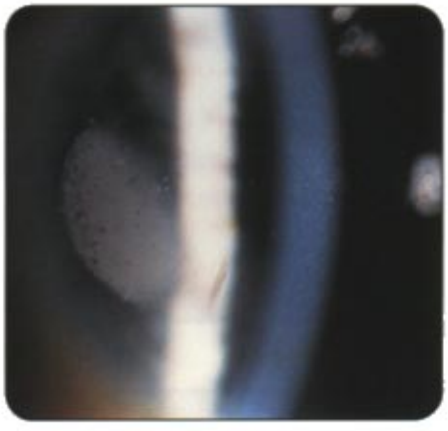


Fig. 5.70

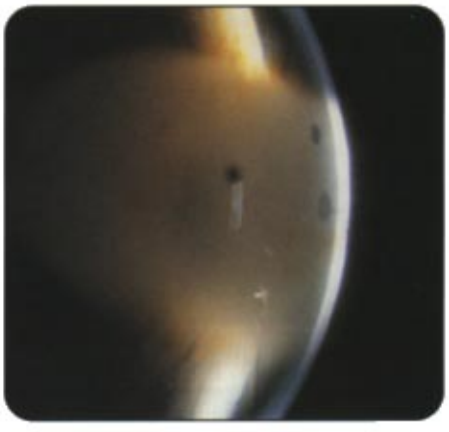


Fig. 5.71

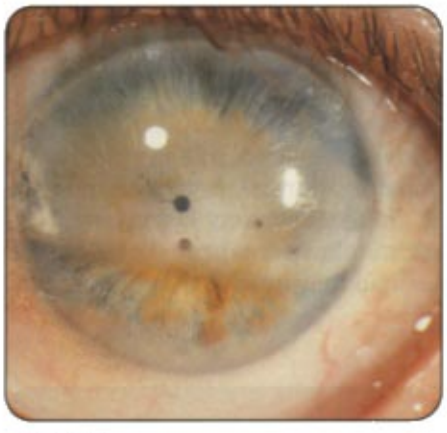


Fig. 5.72

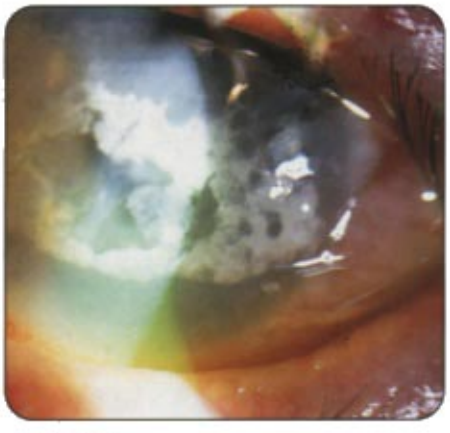


Fig. 5.73

Table 5.9 Causes of band keratopathy

Ocular
• chronic iridocyclitis – particularly in children
• phthisis bulbi
• silicone oil in the anterior chamber
Metabolic
• increased serum calcium and phosphorus
• gout and and other causes of hyperuricemia
• chronic renal failure
Hereditary
• hereditary ichthyosis
• familial
Age-related

Spheroidal degeneration (Labrador keratopathy)

Spheroidal degeneration is a rare, bilateral condition which typically affects outdoor workers although in some cases it occurs secondary to other corneal disorders, particularly Fuchs' endothelial dystrophy and lattice dystrophy.

Signs (in chronological order)

- Peripheral, interpalpebral, small amber-colored granules in the superficial corneal stroma and conjunctiva.
- The lesions spread centrally and coalesce, becoming more opaque (**Fig. 5.74**).
- Advanced lesions become nodular and elevated (**Figs 5.75, 5.76**).

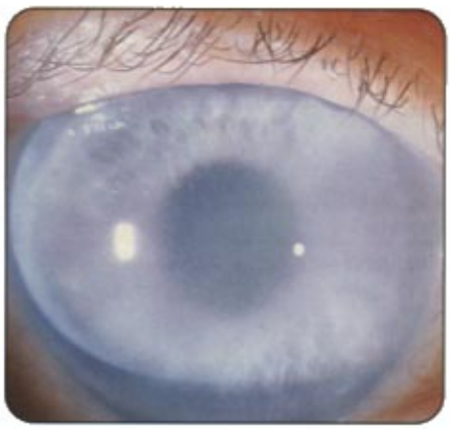


Fig. 5.74

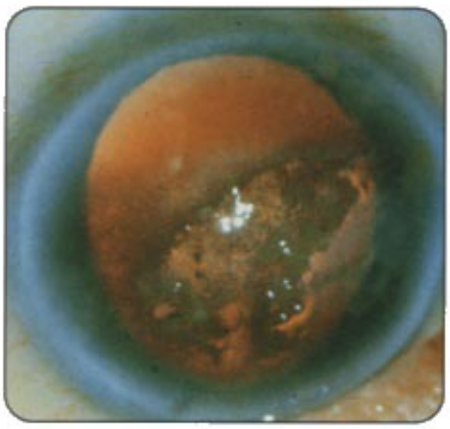


Fig. 5.75

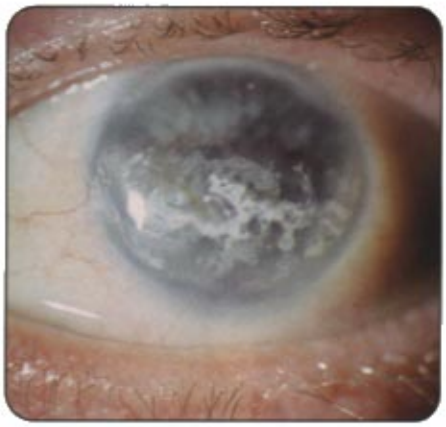


Fig. 5.76

Lipid keratopathy

Lipid keratopathy is a common, unilateral condition most frequently associated with previous herpes simplex or herpes zoster keratitis. Rarely, it develops for no apparent cause.

Signs

- Unilateral, focal, white or yellowish deposit with feathery edges (**Figs 5.77, 5.78**). Secondary lesions are associated with vascularization, whereas primary lesions are avascular. Rarely, the lipid may involve the entire cornea (**Fig. 5.79**).

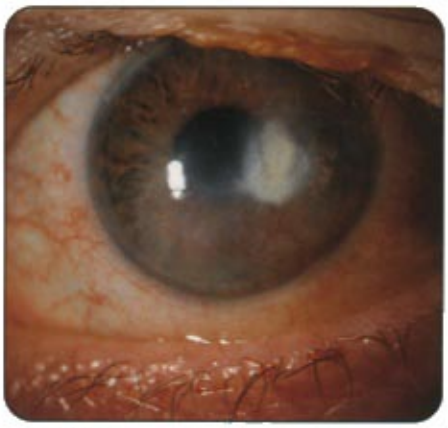


Fig. 5.77

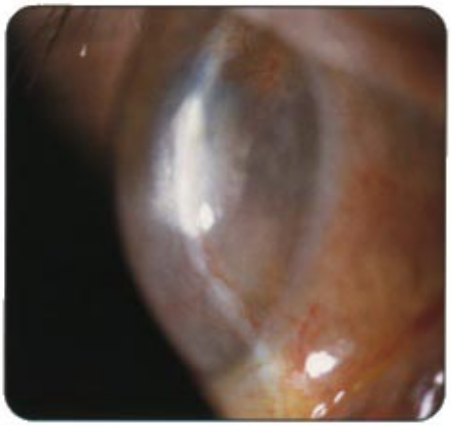


Fig. 5.78

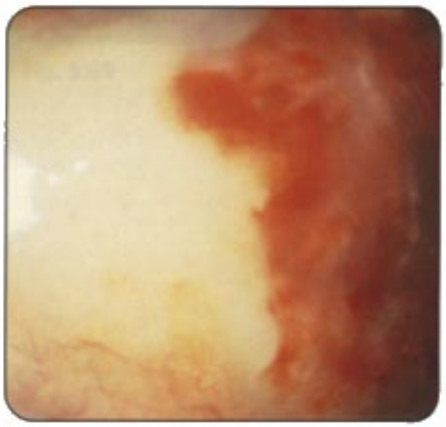


Fig. 5.79

Salzmann nodular degeneration

Salzmann nodular degeneration is an uncommon, usually unilateral condition which is always secondary to a chronic keratopathy.

Signs

- Discrete, elevated, gray, superficial stromal nodules which may be surrounded by iron-pigment deposits (**Figs 5.80–5.82**).

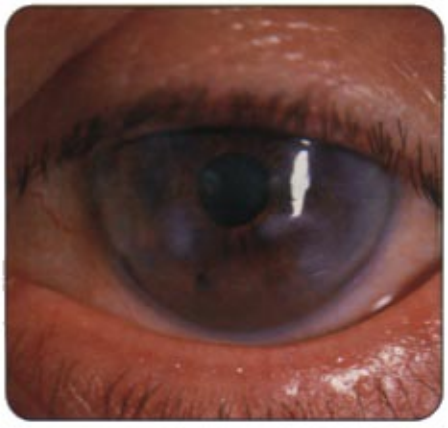


Fig. 5.80

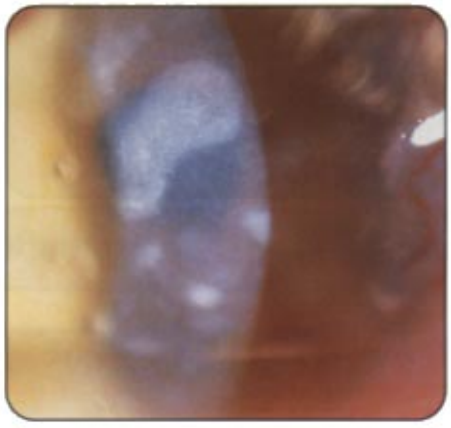


Fig. 5.81

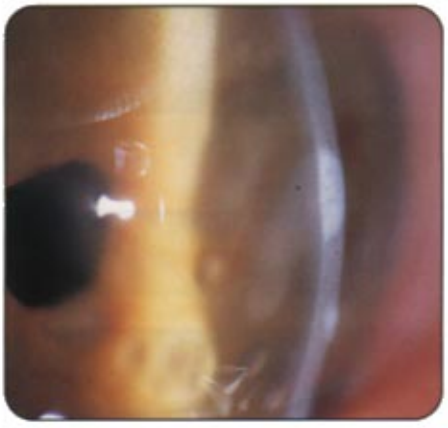


Fig. 5.82

Dystrophies

Reis–Bucklers dystrophy

Reis–Bucklers dystrophy is a common, dominantly inherited condition which may be associated with severe recurrent corneal erosions. Reduced visual acuity occurs in the second to third decades of life.

Signs

- 'Honeycomb' appearance due to ring-shaped subepithelial opacities which involve the entire cornea but are most dense centrally (**Figs 5.83, 5.84**).

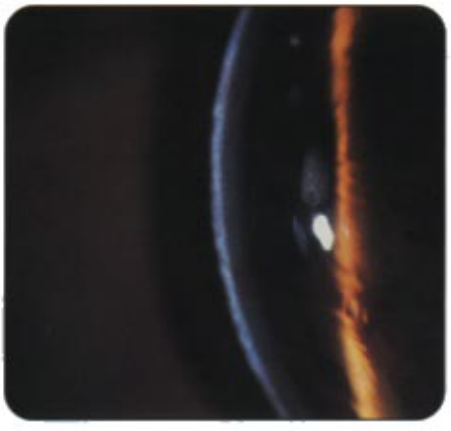


Fig. 5.83

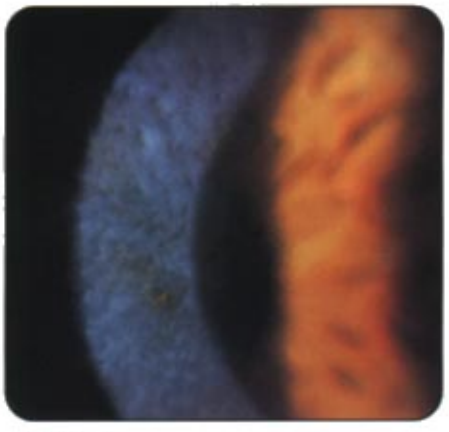


Fig. 5.84

Anterior membrane (Grayson–Wilbrandt dystrophy)

Anterior membrane dystrophy is an uncommon, dominantly inherited condition which is clinically similar to Reis–Bucklers except that:

- Peripheral cornea is uninvolved (**Fig. 5.85**).
- Cornea between the lesions is clear (**Fig. 5.86**).
- Visual acuity is less severely affected.
- Recurrent erosions are less frequent.

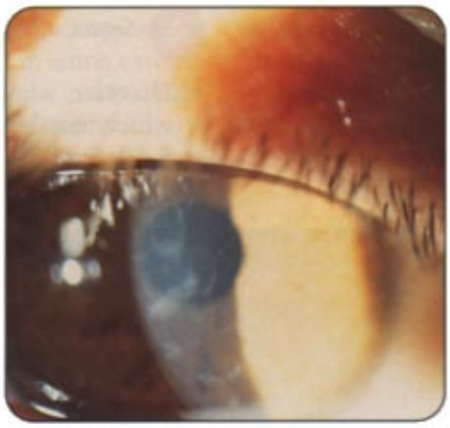


Fig. 5.85

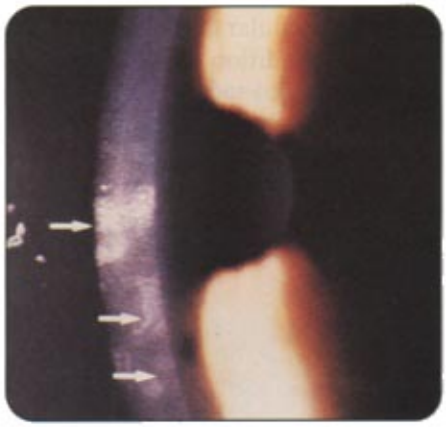


Fig. 5.86

STROMAL LESIONS

Noninflammatory stromal edema

Causes

- Iatrogenic endothelial damage – for example, cataract surgery, particularly when associated with anterior chamber lens implantation (pseudophakic bullous keratopathy) (**Figs 5.87, 5.88**).
- Hydrops of keratoconus (**Figs 5.89, 5.90**).
- Hydrops of keratoglobus (**Fig. 5.91**).
- Fuchs endothelial dystrophy.
- Acute angle-closure glaucoma.
- Corneal graft rejection is shown as endothelial (Khoudadoust) rejection lines and anterior uveitis.
- Blunt anterior segment trauma.
- Congenital hereditary endothelial dystrophy (CHED) gives rise to bilateral corneal decompensation at any time during the first decade of life. When present in infancy, CHED should be differentiated from glaucoma and birth trauma, which can also give rise to corneal edema.

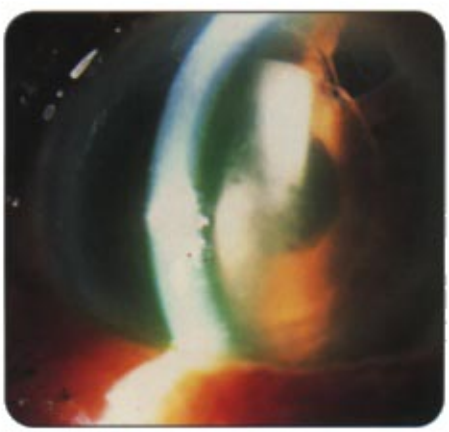


Fig. 5.87

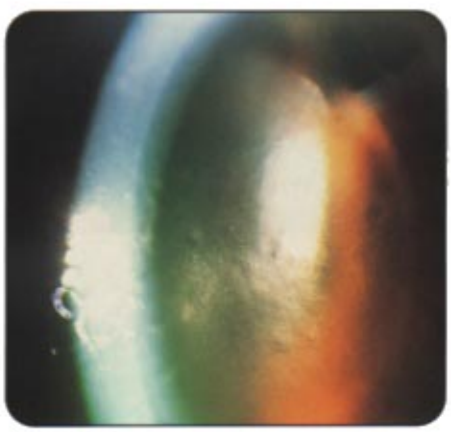


Fig. 5.88

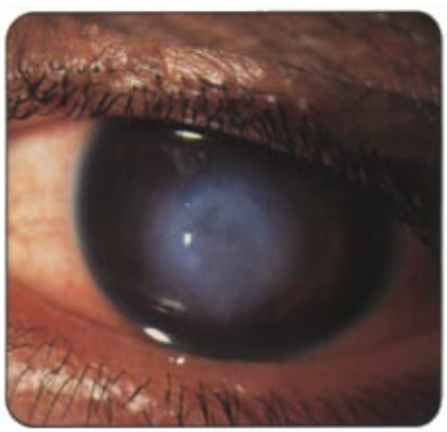


Fig. 5.89

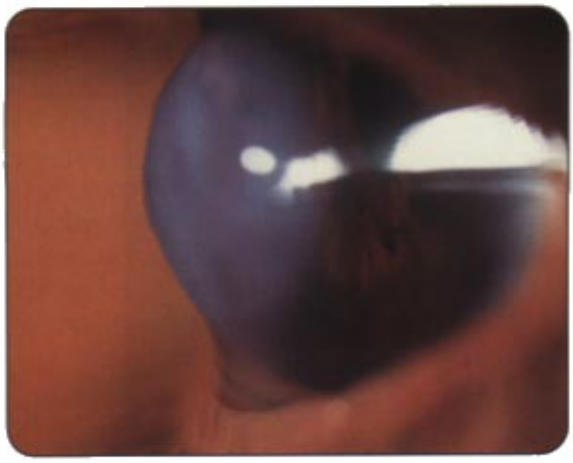


Fig. 5.90

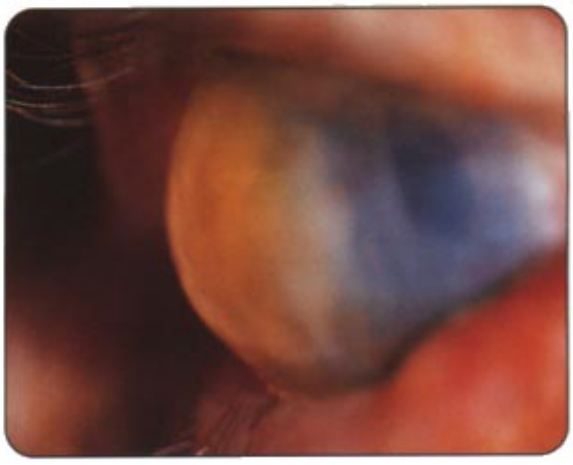


Fig. 5.91

Inflammatory stromal lesions

Disciform keratitis

Disciform keratitis is a common, unilateral condition probably caused by an immunologic reaction to herpes simplex or, less frequently, varicella-zoster antigen.

Signs

- Round or oval area of stromal and epithelial edema (**Fig. 5.92**).
- Small keratic precipitates localized to the involved endothelium (**Fig. 5.93**).
- Folds in Descemet membrane (**Fig. 5.94**).
- Surrounding ring of stromal opacity (Wessely ring) (**Fig. 5.95**) in longstanding cases.

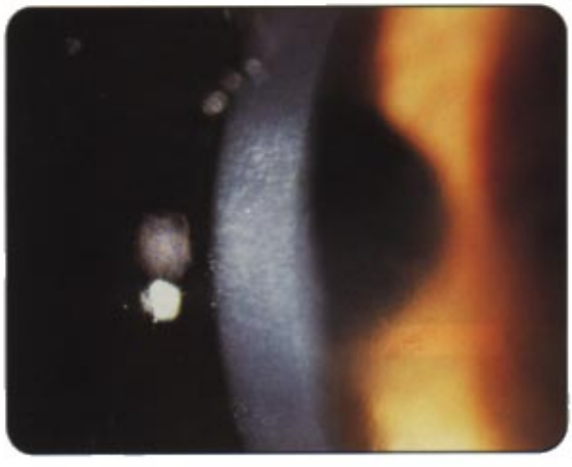


Fig. 5.92

Fig. 5.93

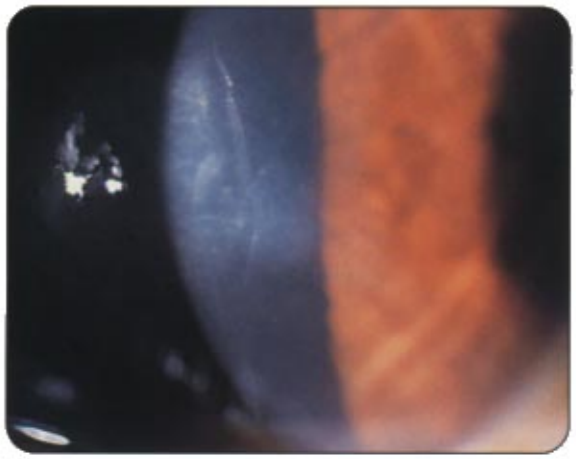


Fig. 5.94

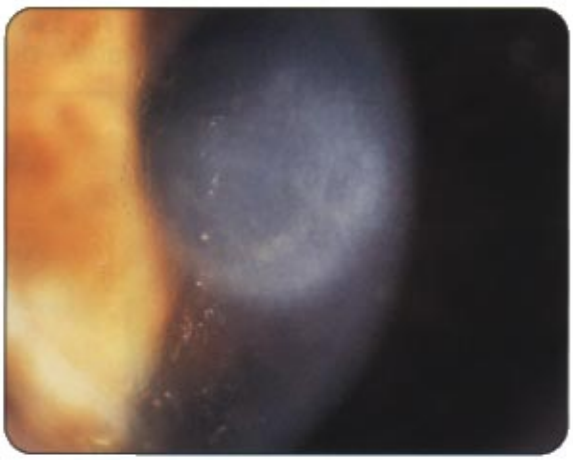


Fig. 5.95

Herpes simplex necrotic stromal keratitis

Herpes simplex necrotic stromal keratitis is an uncommon, unilateral condition.

Signs

- Cheesy, necrotic stromal infiltration (**Fig. 5.96**) which may be associated with epithelial breakdown (**Fig. 5.97**).
- In advanced cases there may be vascularization, scarring (**Fig. 5.98**), and occasionally, perforation (**Fig. 5.99**).

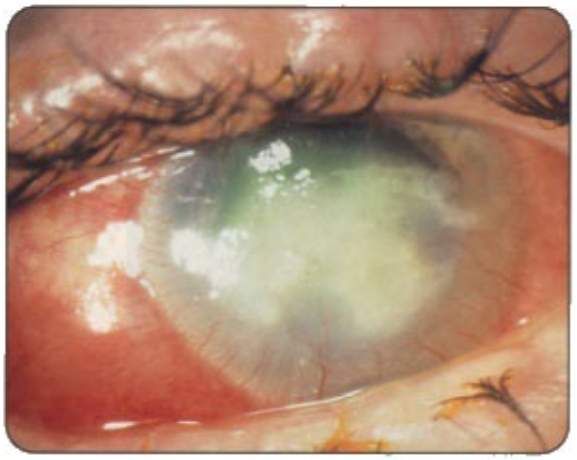


Fig. 5.96

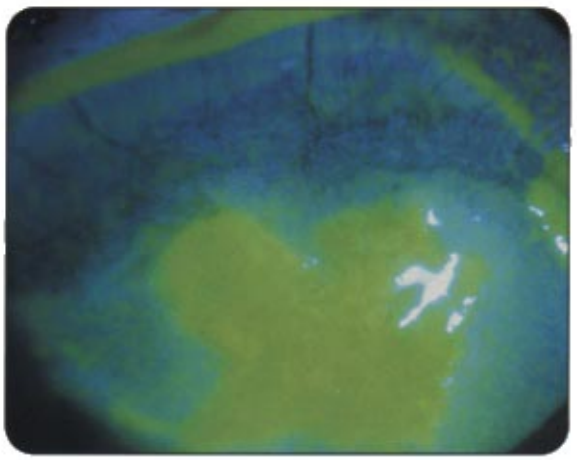


Fig. 5.97

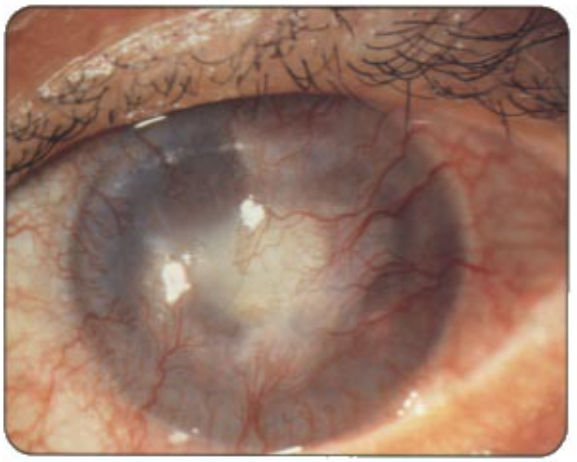


Fig. 5.98

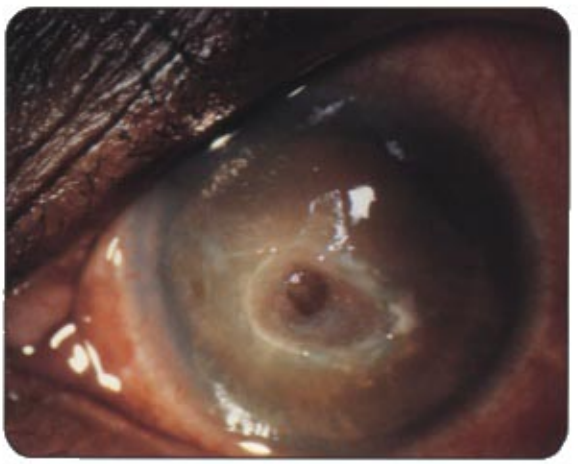


Fig. 5.99

Bacterial keratitis

Bacterial keratitis is a common, unilateral condition which develops in persons with pre-existing ocular surface disease or in those who wear contact lenses. The most frequent causative organisms are *Staphylococcus aureus*, *Streptococcus pneumoniae*, and *Pseudomonas aeruginosa*. The signs vary according to the severity of the infection and, to a lesser extent, on the causative organism.

Signs

- An early, nonspecific finding is a white stromal infiltrate associated with ciliary conjunctival injection (**Fig. 5.100**), an overlying epithelial defect, and secondary anterior uveitis.
- Staphylococcal keratitis is characterized by a well-defined, white–gray or creamy stromal infiltrate (**Fig. 5.101**) which may enlarge to form a dense stromal abscess (**Fig. 5.102**).
- Pneumococcal keratitis tends to spread superficially with a serpiginous leading edge. Severe anterior uveitis and hypopyon formation are common (**Fig. 5.103**).
- Pseudomonal keratitis is characterized by a rapidly extending, suppurative infiltrate associated with hypopyon (**Fig. 5.104**) and a mucopurulent discharge (**Fig. 5.105**).
- Corneal perforation (**Fig. 5.106**) may occur in advanced cases.

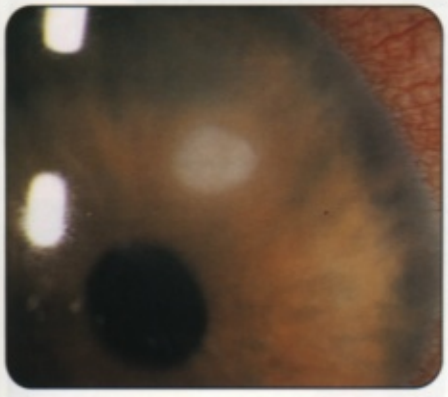


Fig. 5.100

Fig. 5.101

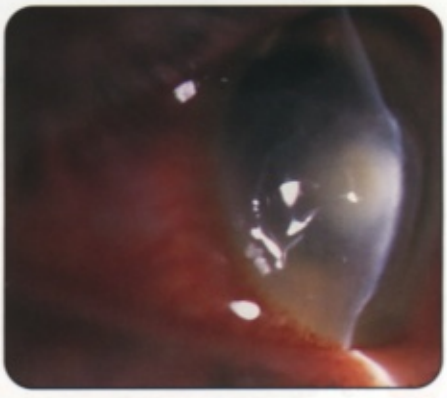


Fig. 5.102

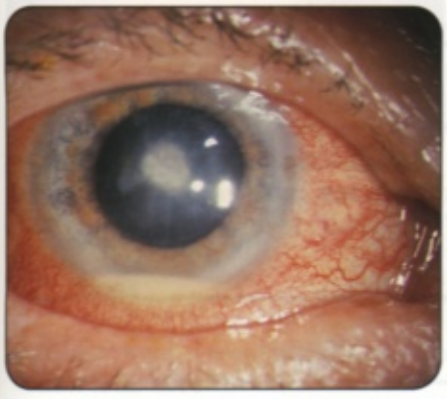


Fig. 5.103

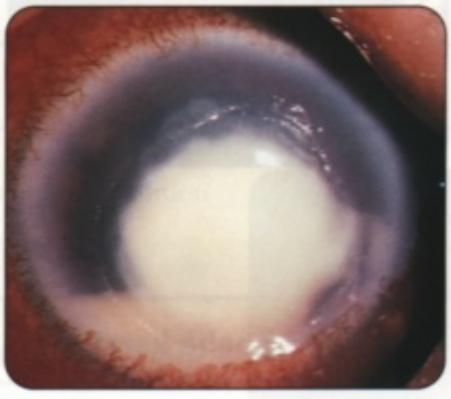


Fig. 5.104

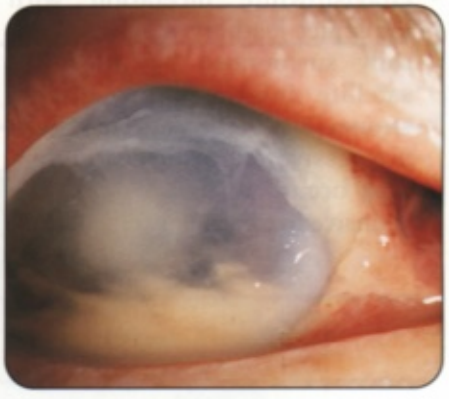


Fig. 5.105

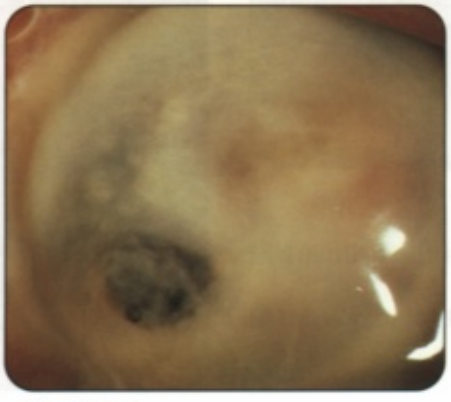


Fig. 5.106

Candida keratitis

Candida keratitis is a rare, unilateral, insidious fungal infection which usually occurs in those with pre-existing chronic corneal disease or in severely debilitated patients.

Signs

- Gray–white infiltrate (**Fig. 5.107**) similar to a bacterial ulcer.

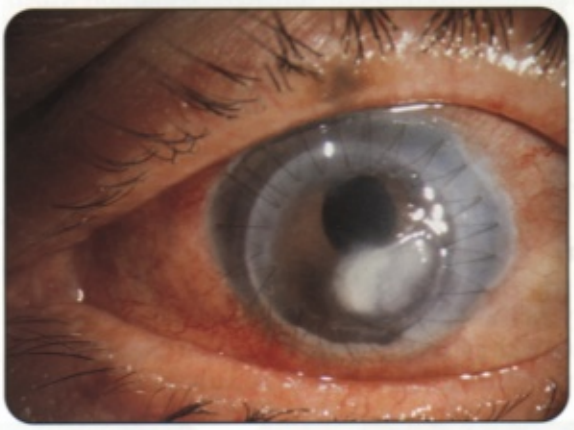


Fig. 5.107

Filamentous keratitis

Filamentous keratitis is a rare, unilateral, insidious fungal infection which is typically preceded by ocular trauma with wood.

Signs

- Grayish-white ulceration with indistinct feathery margins, typically surrounded by finger-like satellite infiltrates in adjacent stroma (**Fig. 5.108**). Severe slowly-progressive corneal destruction may occur in some cases unresponsive to treatment (**Fig. 5.109**).

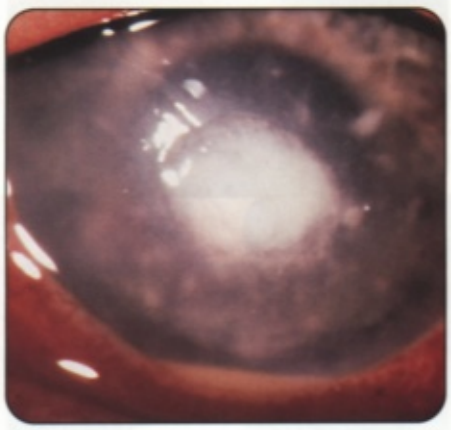


Fig. 5.108

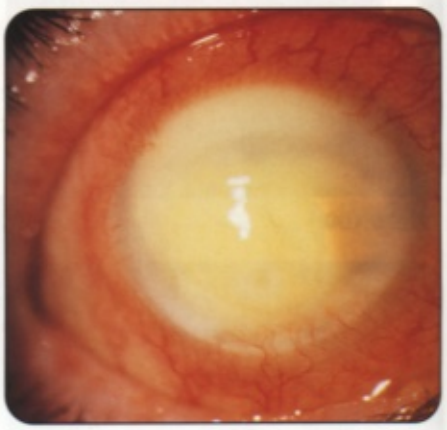


Fig. 5.109

Acanthamoeba keratitis

Acanthamoeba keratitis is a very rare, unilateral condition which typically affects wearers of soft contact lenses.

Signs

- Dendritiform epithelial lesions (*see* **Figs 5.59, 5.60**).
- Nonspecific stromal infiltrates (**Fig. 5.110**).
- Radial keratoneuritis (**Fig. 5.111**).
- A ring infiltrate (**Figs 5.112, 5.113**) is the hallmark of advanced infection.
- Corneal necrosis and thinning (**Fig. 5.114**) may occur in advanced cases.

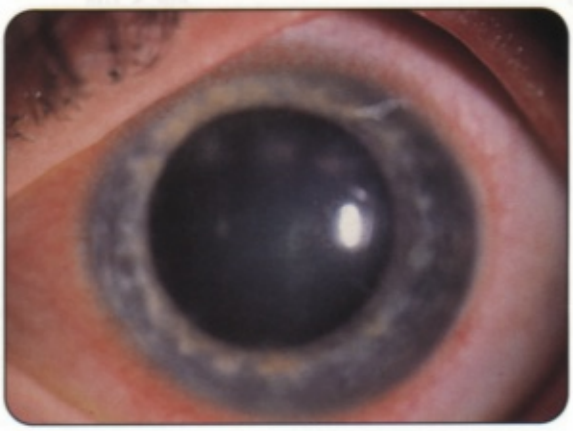


Fig. 5.110

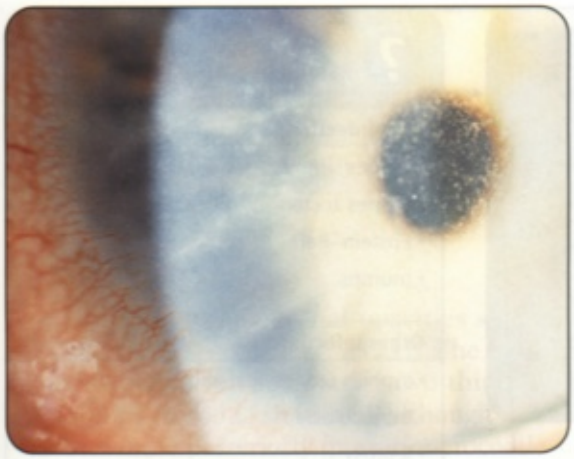


Fig. 5.111

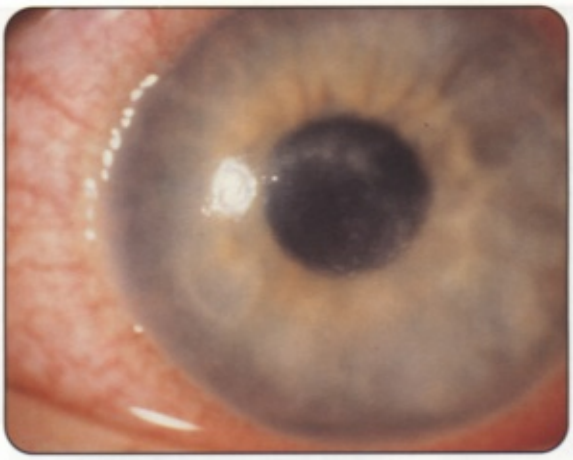


Fig. 5.112

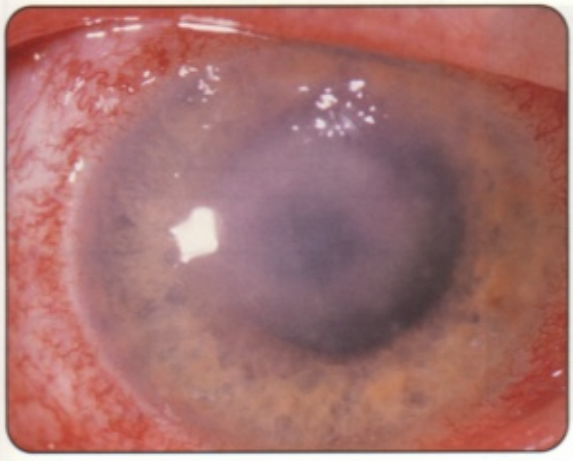


Fig. 5.113

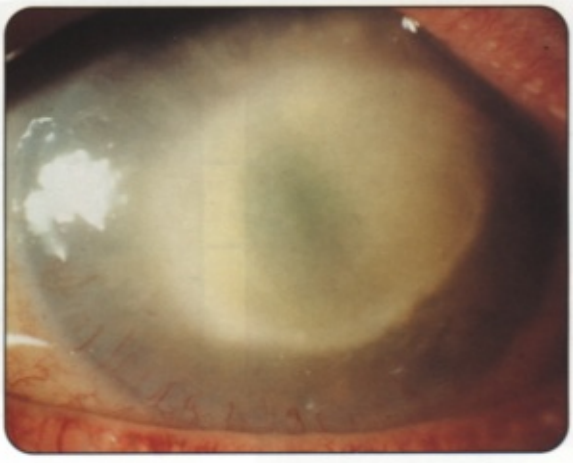


Fig. 5.114

Interstitial keratitis

Interstitial keratitis is an uncommon, bilateral condition which has diverse causes (**Table 5.10**).

Signs

- Midstromal, vascularized and nonsuppurative infiltration (**Fig. 5.115**).
- Inactive signs include variable stromal scarring (**Fig. 5.116**) associated with nonperfused (ghost) vessels (**Fig. 5.117**).
- Rarely, reperfusion may give rise to a corneal intrastromal hemorrhage (**Figs 5.118, 5.119**).

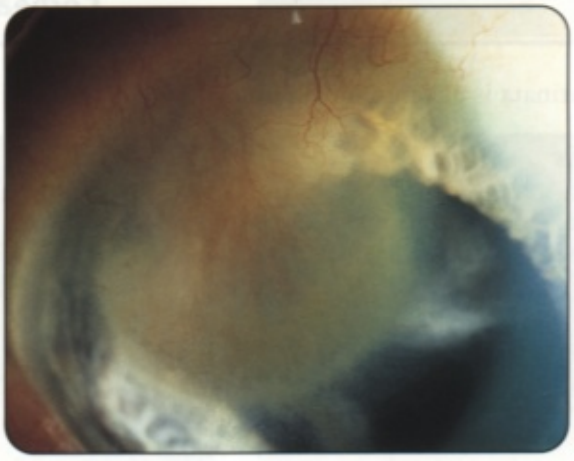


Fig. 5.115

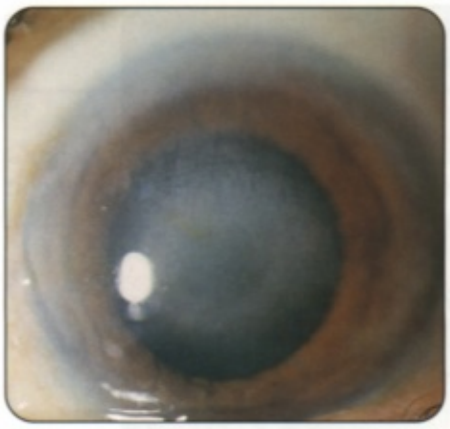


Fig. 5.116

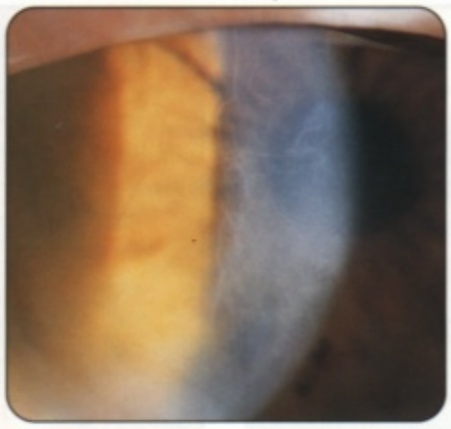


Fig. 5.117

Fig. 5.118

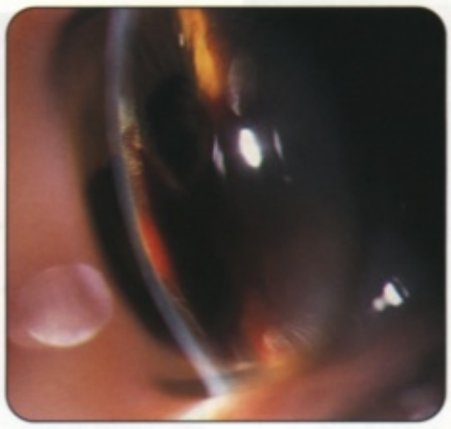


Fig. 5.119

Table 5.10 Causes of interstitial keratitis

Viral infections
• herpes simplex
• herpes zoster
• Epstein–Barr
• mumps
Other infections
• syphilis (congenital and acquired)
• tuberculosis
• leprosy
• onchocerciasis
• lymphogranuloma venereum
• Lyme disease
Cogan syndrome

Stromal dystrophies

Cornea farinata

Cornea farinata is a common and innocuous condition.

Signs

- Minute, flour-like deposits in the deep stroma most prominent centrally (**Fig. 5.120**).

Fig. 5.120

Crocodile shagreen

Crocodile shagreen is an uncommon and innocuous condition.

Signs

- Grayish-white, polygonal opacities separated by relatively clear spaces (**Fig. 5.121**). The lesions most frequently involve the anterior two-thirds of the stroma (anterior crocodile shagreen), although they may also be found more posteriorly (posterior crocodile shagreen).

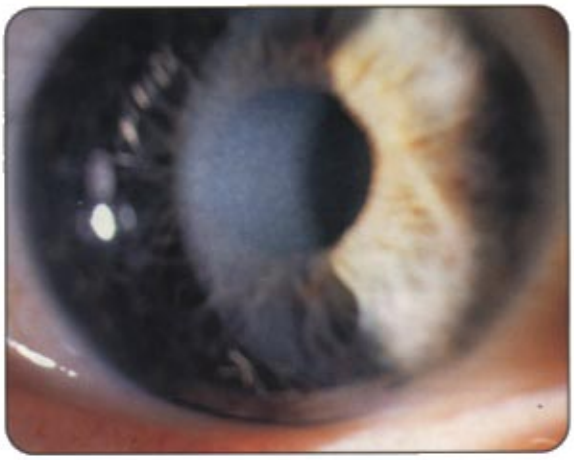


Fig. 5.121

Granular dystrophy

Granular dystrophy is an uncommon, dominantly inherited condition which becomes manifest during the first decade of life.

Signs

- Small, discrete, crumb-like, white granules within the central anterior stroma, with clear cornea in between (**Fig. 5.122**).
- With time, the number of lesions extend deeper within the stroma, and become larger and more numerous (**Figs 5.123–5.126**).
- Coalescence of lesions (**Figs 5.127, 5.128**) may cause visual impairment.

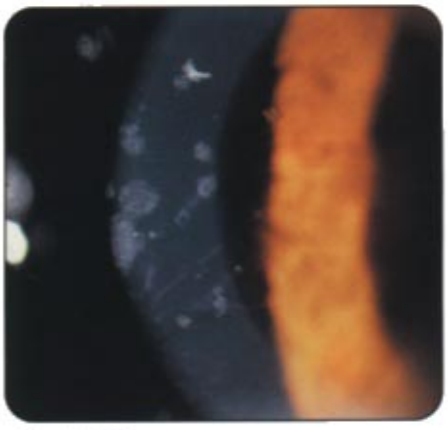


Fig. 5.122

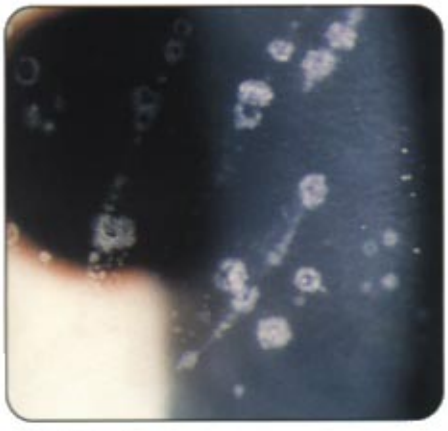


Fig. 5.123

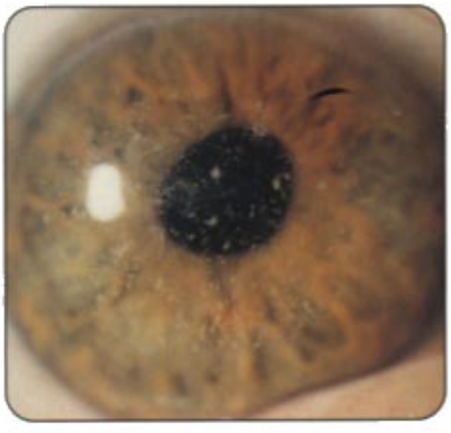


Fig. 5.124

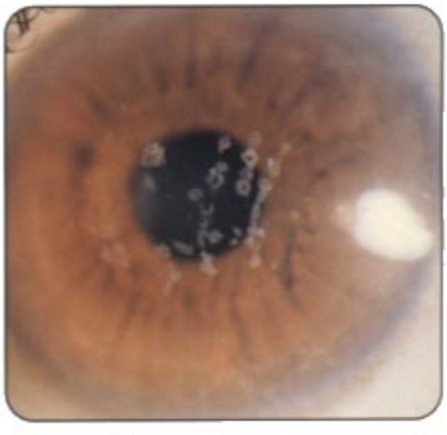


Fig. 5.125

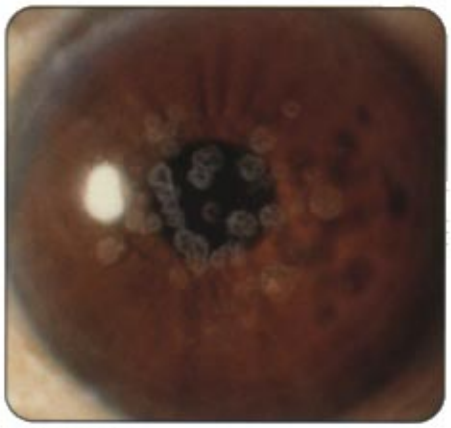


Fig. 5.126

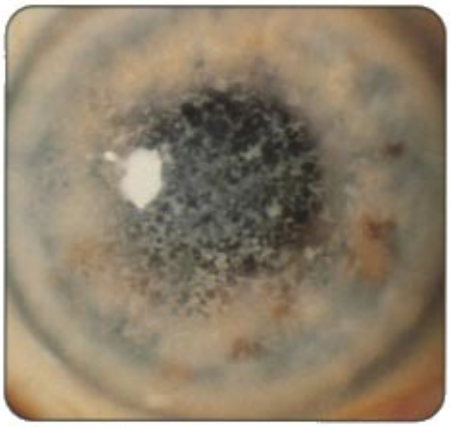


Fig. 5.127

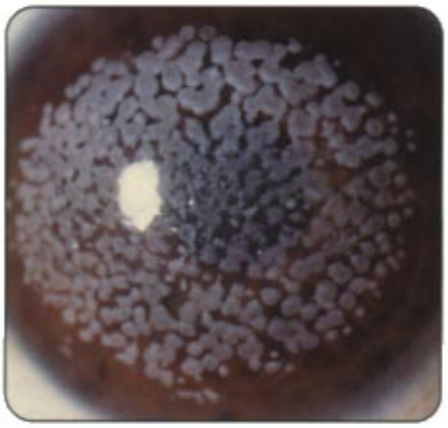


Fig. 5.128

Macular dystrophy

Macular dystrophy is a rare, recessively inherited condition which can be subdivided into three types:

- Type I – presents in childhood with corneal erosive attacks and is the most common.
- Type II – presents in the second decade with mild erosive attacks.
- Type III – presents in infancy with severe erosive attacks.

Signs

- Central, focal, gray–white, poorly delineated opacities with diffuse cloudiness of the intervening stroma (**Figs 5.129–5.131**).
- The lesions eventually involve the entire stromal thickness and extend to the limbus (**Figs 5.132, 5.133**).

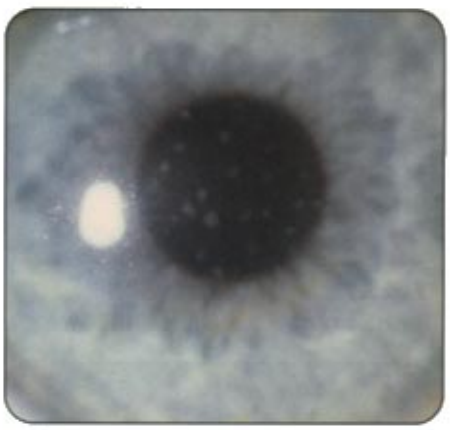


Fig. 5.129

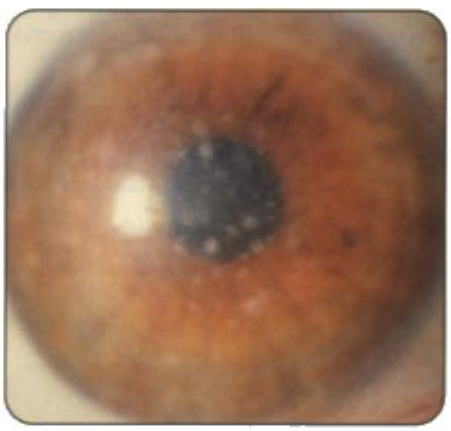


Fig. 5.130

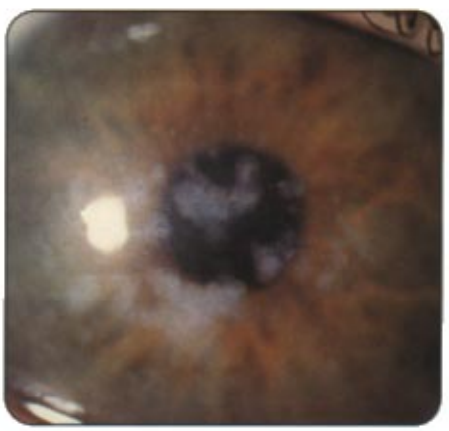


Fig. 5.131

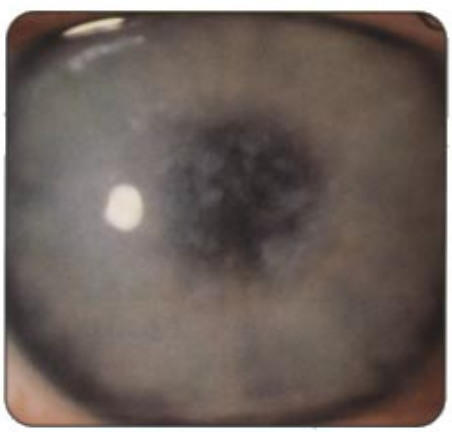


Fig. 5.132

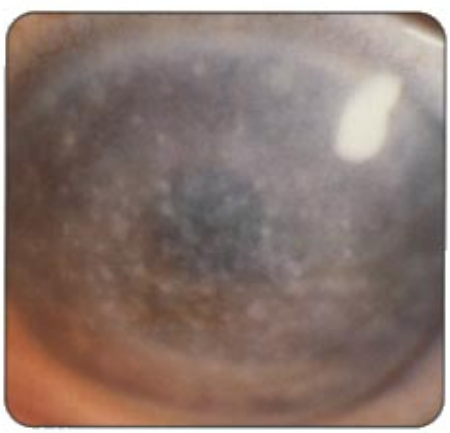


Fig. 5.133

Lattice dystrophy

Lattice dystrophy is an uncommon condition which can be subdivided into three types:

- Type I has autosomal dominant inheritance.
- Type II is associated with systemic amyloidosis (Meretoja syndrome) and has autosomal dominant inheritance.
- Type III has autosomal recessive inheritance.

Signs

- Type I – fine, branching, spider-like refractile lines at different levels within the stroma (**Figs 5.134–5.136**).
- Type II – lattice lines are thicker but less numerous than in type I.
- Type III – lattice lines are coarser than in type I (**Figs 5.137, 5.138**).

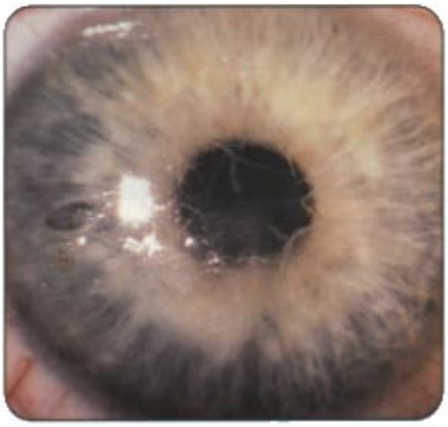


Fig. 5.134

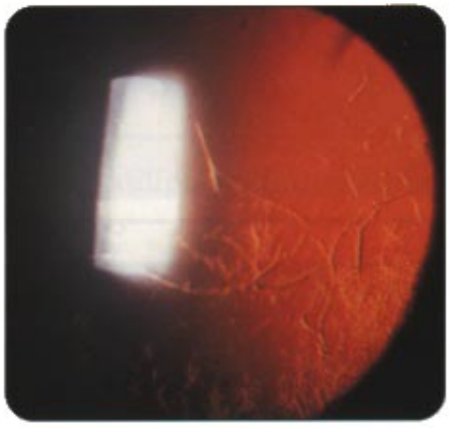


Fig. 5.135

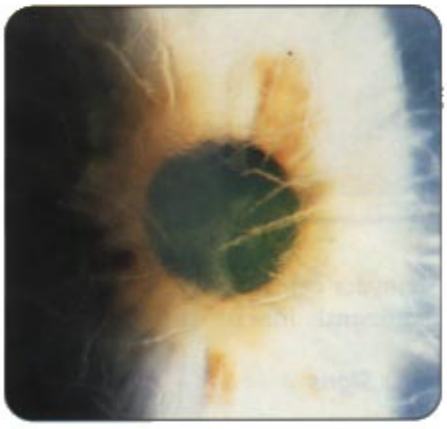


Fig. 5.136

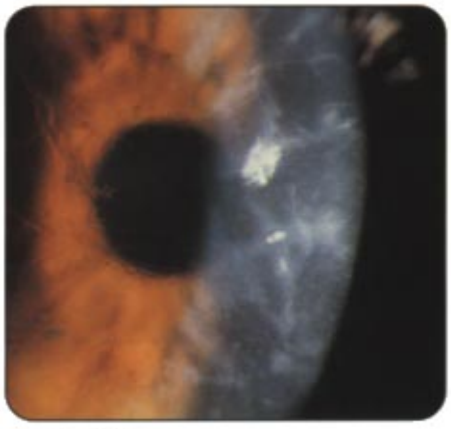


Fig. 5.137

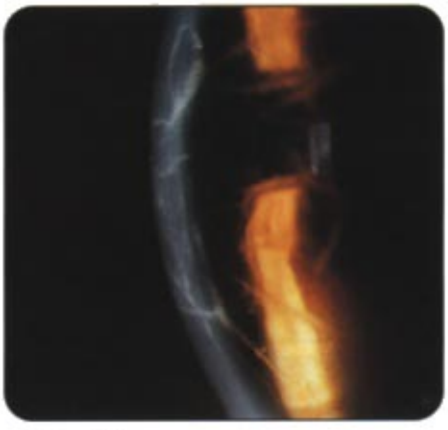


Fig. 5.138

Granular-lattice (Avellino) dystrophy

Granular-lattice dystrophy is a very rare, dominantly inherited condition.

Signs

- Anterior stromal opacities suggestive of granular dystrophy, associated with deeper stromal lesions similar to those found in lattice dystrophy (**Fig. 5.139**).

Fig. 5.139

Schnyder central crystalline dystrophy

Schnyder central crystalline dystrophy is an uncommon, dominantly inherited condition.

Signs

- Needle-shaped crystals mainly involving the central anterior stroma which may be associated with a diffuse central stromal haze (**Fig. 5.140**).

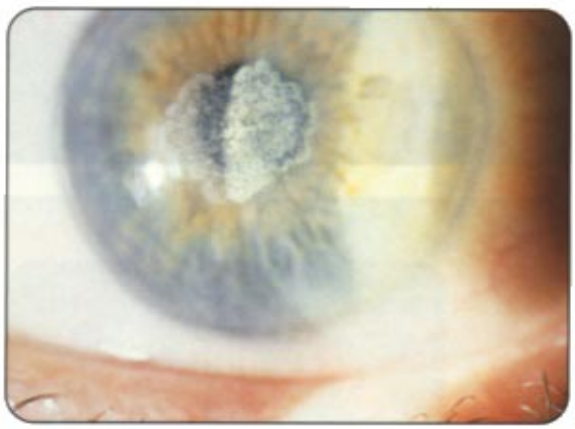


Fig. 5.140

Stromal crystals

Infectious crystalline keratopathy

Infectious crystalline keratopathy is an uncommon condition, usually caused by *Streptococcus viridans*, which is also referred to as 'aborted bacterial keratopathy'. It may develop as a complication of penetrating keratoplasty and the long-term use of topical steroids.

Signs

- Discrete, white, branching crystalline deposits without associated inflammation, in the anterior stroma (**Fig. 5.141**).

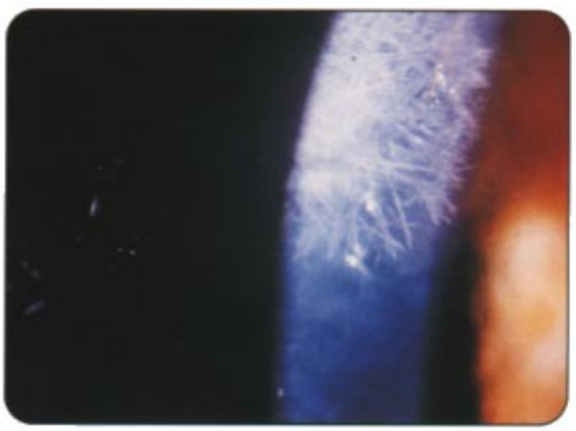


Fig. 5.141

Systemic causes of corneal crystals

- Cystinosis (**Figs 5.142–5.144**).
- Gout.
- Monoclonal gammopathies.
- Lecithin cholesterol acyltransferase (LCAT) deficiency (**Fig. 5.145**).
- Tangier disease.
- Chrysiasis (**Fig. 5.146**).

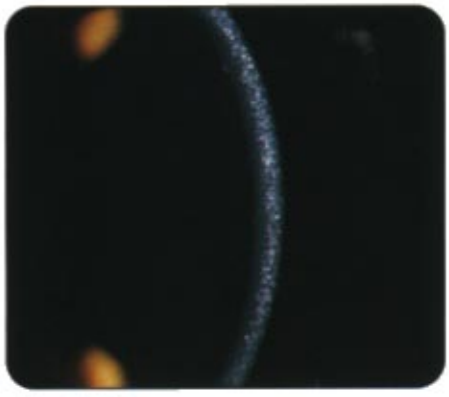


Fig. 5.142

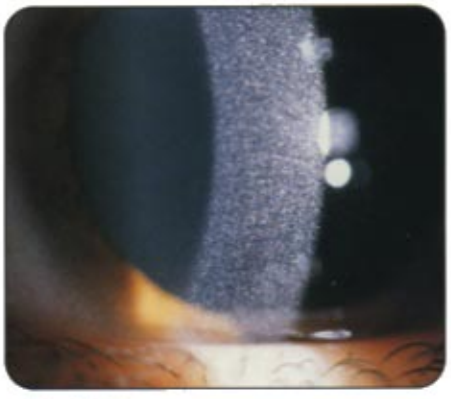


Fig. 5.143

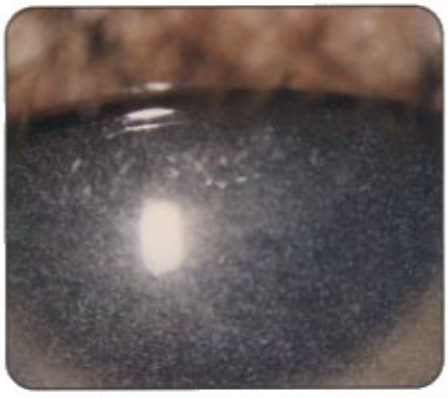


Fig. 5.144

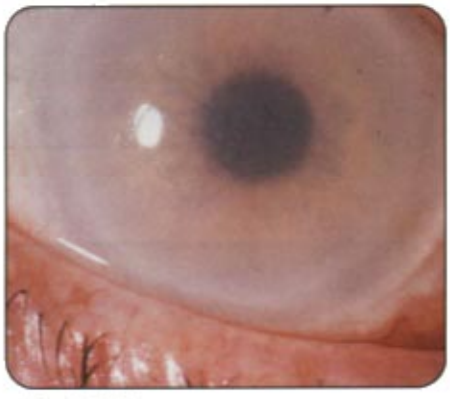


Fig. 5.145

Fig. 5.146

Prominent corneal nerves

Prominent corneal nerves (**Fig. 5.147**) are associated with a variety of ocular and systemic disorders (**Table 5.11**).

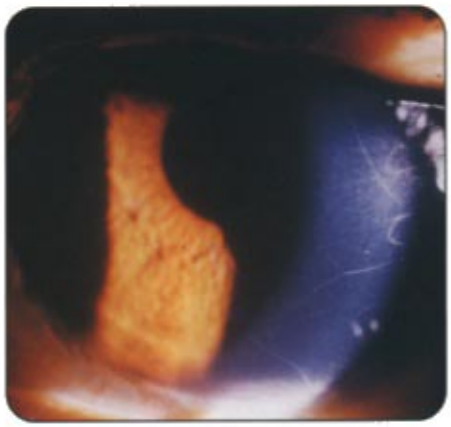


Fig. 5.147

Table 5.11 Causes of prominent corneal nerves

Involving entire cornea
• neurofibromatosis type I
• leprosy
• Refsum syndrome
• primary amyloidosis
• hereditary ichthyosis
• multiple endocrine neoplasia IIb (MEN-IIb)
Localized secondary thickening
• keratoconus
• failed corneal graft
• acanthamoeba keratitis

Diffuse corneal clouding at birth

Figure 5.148 shows corneal clouding associated with buphthalmos.
Figure 5.149 shows corneal clouding in Morquio syndrome.

Fig. 5.148

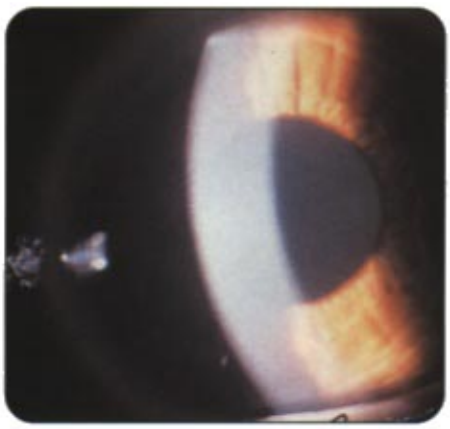


Fig. 5.149

Table 5.12 Causes of diffuse corneal clouding at birth

In a small eye
• complex microphthalmos
In a large eye
• buphthalmos
In a normal-sized eye
• birth trauma
• interstitial keratitis
• rubella keratitis
• glaucoma without buphthalmos
• total sclerocornea
• congenital hereditary endothelial dystrophy
• congenital stromal dystrophy
• mucopolysaccharidosis (Hurler, Scheie, Morquio, and Maroteaux–Lamy)
• mucolipidosis – types I, II, and IV

LESIONS OF DESCEMET MEMBRANE

Tears

Causes

- Infantile glaucoma – horizontal tears (Haab striae) (**Figs 5.150, 5.151**).
- Birth trauma – vertical tears (**Fig. 5.152**).
- Acute hydrops in keratoconus (*see* **Figs 5.89, 5.90**).
- Acute hydrops in keratoglobus (*see* **Fig. 5.91**).

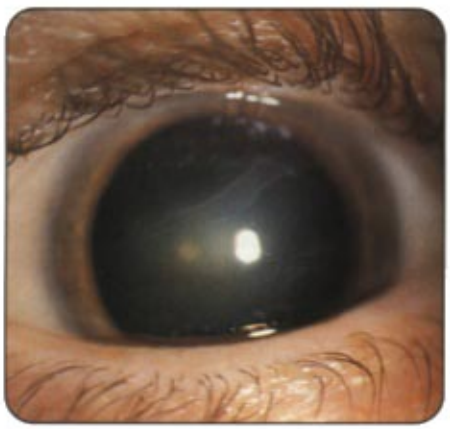


Fig. 5.150

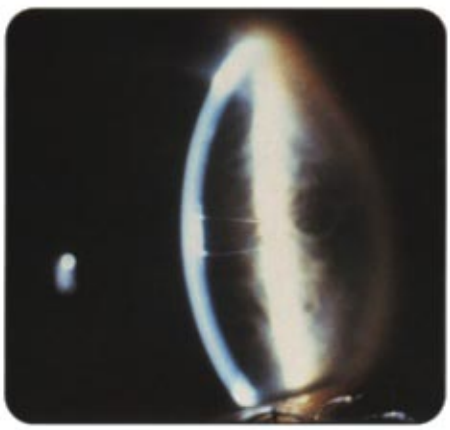


Fig. 5.151

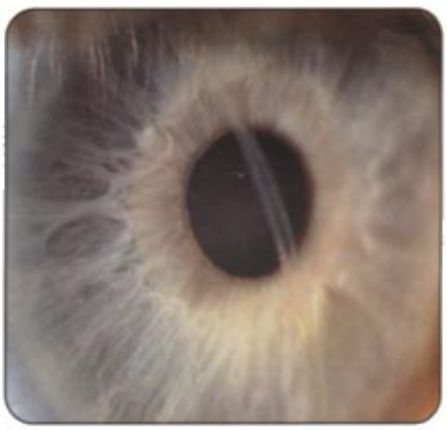


Fig. 5.152

Folds and wrinkles

Causes

- Surgical trauma may cause folds (**Fig. 5.153**).
- Ocular hypotony.
- With disciform keratitis (*see* **Fig. 5.94**).
- Congenital syphilis may cause wrinkles (**Fig. 5.154**).

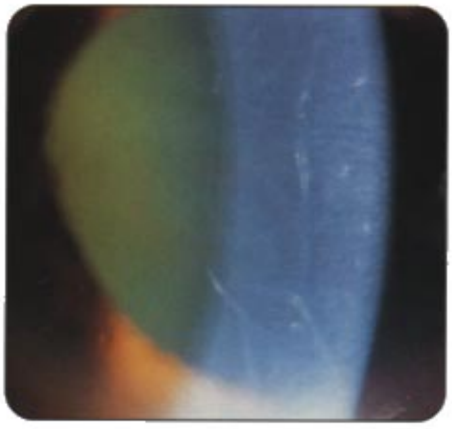


Fig. 5.153

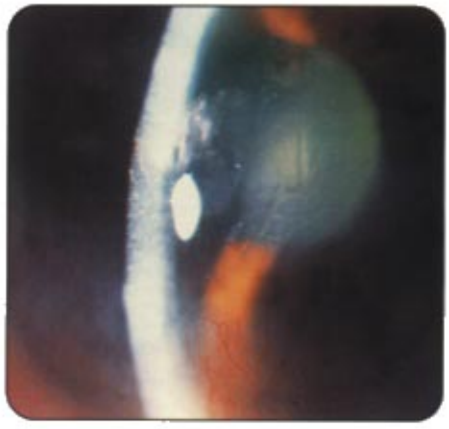


Fig. 5.154

Detachment

Detachment of Descemet membrane (**Fig. 5.155**) is caused by surgical trauma.

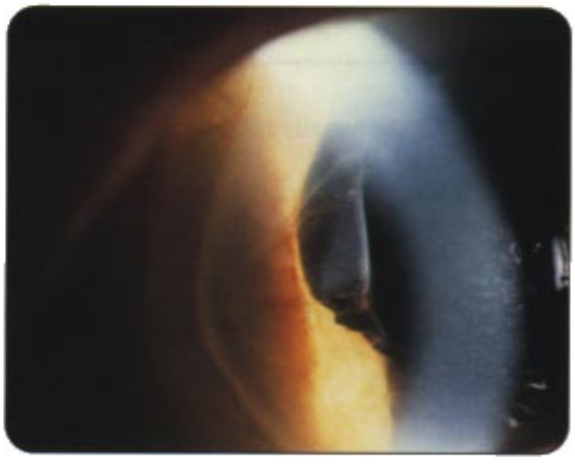


Fig. 5.155

Kayser–Fleischer ring

Signs

- Bilateral, greenish-brown peripheral band 1–3 mm in width at the level of Descemet membrane. The band extends to the limbus and is most prominent in the vertical meridian (**Fig. 5.156**).

Causes

- Wilson disease and other liver disorders.
- Multiple myeloma.
- Carotenemia.

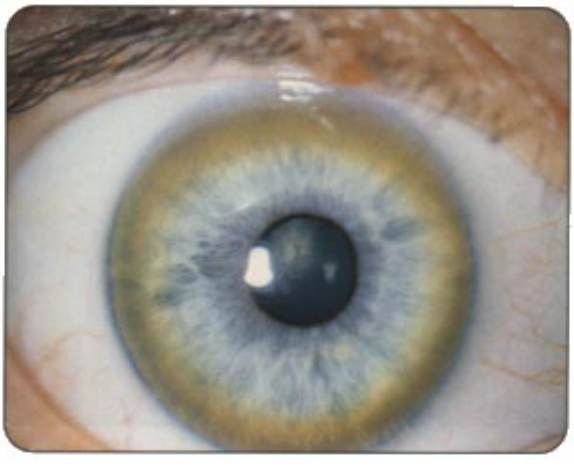


Fig. 5.156

ENDOTHELIAL LESIONS

Keratic precipitates

Keratic precipitates (KP) are cellular deposits on the endothelium which occur in eyes with anterior uveitis (**Fig. 5.157**).

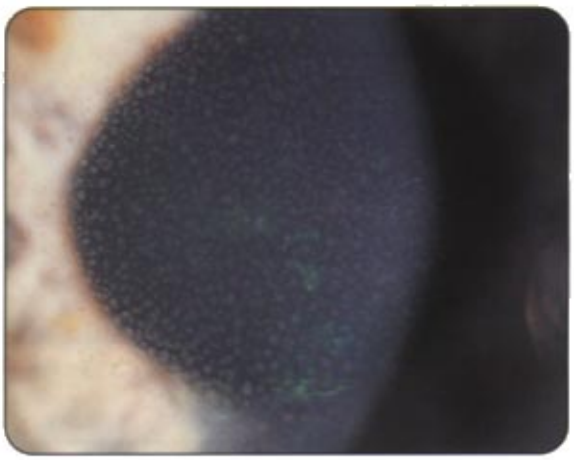


Fig. 5.157

Small KP

- In acute iritis the KP are usually more numerous inferiorly (**Fig. 5.158**).
- In Fuchs heterochromic cyclitis they are characteristically stellate or round and involve the entire endothelium (**Fig. 5.159**). Feathery filaments may be seen between the KP (**Fig. 5.160**).
- In disciform keratitis the KP are confined to the involved area of the cornea (*see* **Fig. 5.93**).

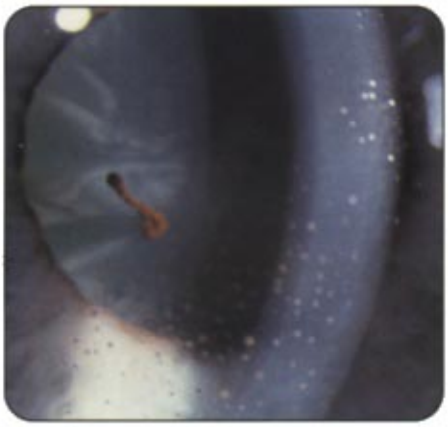


Fig. 5.158

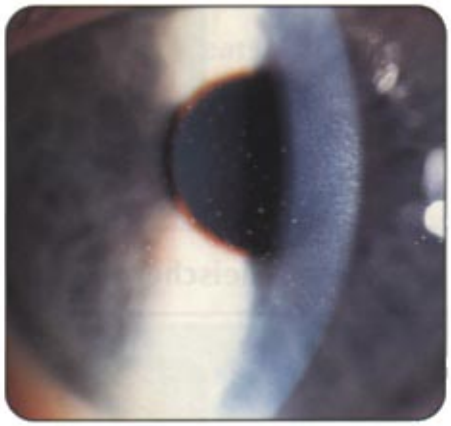


Fig. 5.159

Fig. 5.160

Mutton-fat KP

Mutton-fat KP are medium (**Fig. 5.161**) to large (**Fig. 5.162**) and are characteristic of granulomatous inflammation. They are typically distributed in a base-down triangle, with the larger KP tending to be more numerous at the base and the medium KP more numerous at the apex (**Fig. 5.163**), although this is not always the case (**Fig. 5.164**).

Table 5.13 Causes of granulomatous inflammation

Systemic noninfectious causes
• chronic sarcoidosis
• Vogt–Koyanagi–Harada syndrome
• multiple sclerosis
Systemic infections
• toxoplasmosis
• Lyme disease
• tuberculosis
• syphilis
Local ocular causes
• *Propionibacterium acnes* endophthalmitis
• sympathetic ophthalmitis
• phacoanaphylactic uveitis

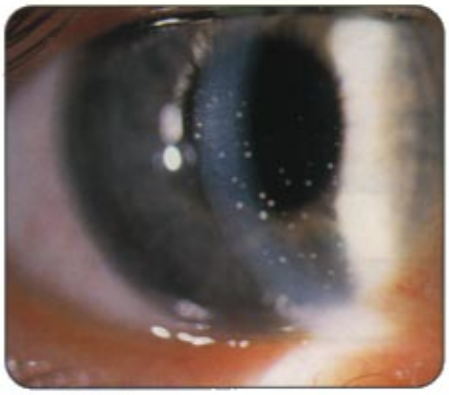


Fig. 5.161

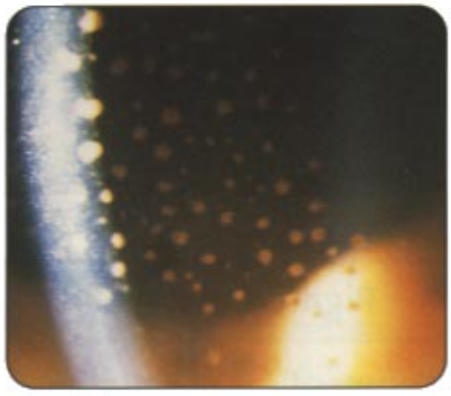


Fig. 5.162

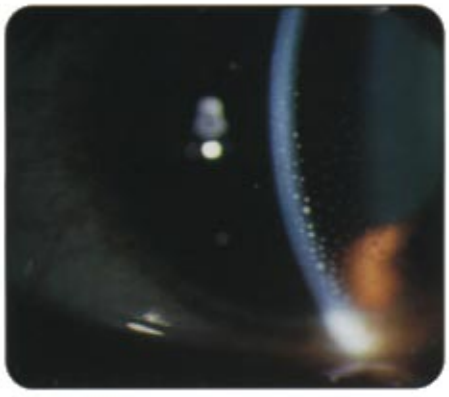


Fig. 5.163

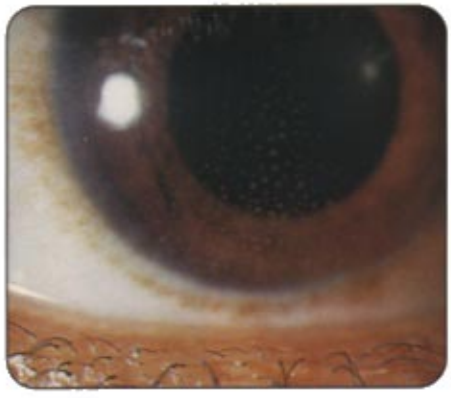


Fig. 5.164

Old KP

Signs

- Pigmented KP (**Fig. 5.165**).
- Ground-glass appearance (**Fig. 5.166**).

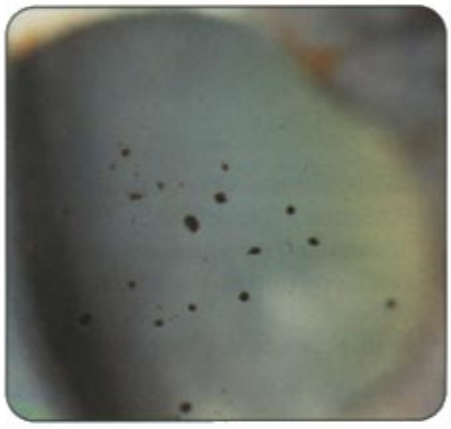


Fig. 5.165

Fig. 5.166

Linear KP

Linear KP occur in association with corneal graft rejection (**Fig. 5.167**).

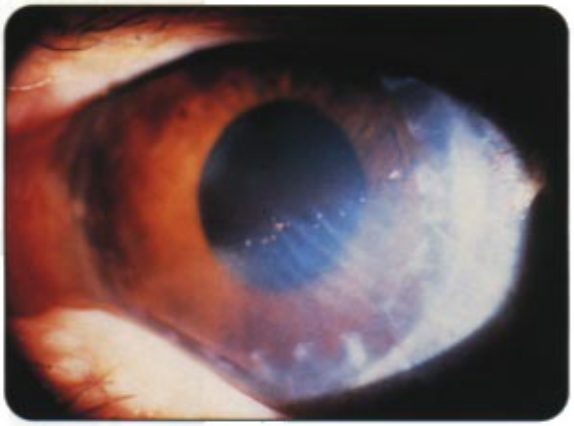


Fig. 5.167

Look for

- Horizontal endothelial rejection line (Khoudadoust).
- Folds in Descement membrane.
- Variable stromal edema.

Pigment deposits

Nonspecific pigment dusting

Nonspecific pigment dusting is a common finding in otherwise normal eyes (**Fig. 5.168**). It may also occur in association with cornea guttata (*see* **Figs 5.172, 5.173**).

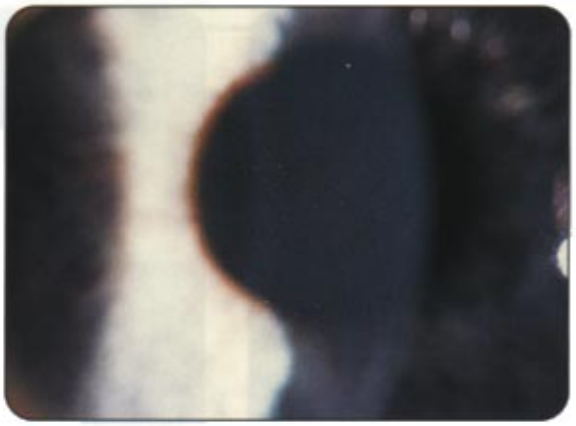


Fig. 5.168

Krukenberg spindle

Signs

- Spindle-shaped deposition of pigment on the endothelium, which is usually vertical (**Fig. 5.169**) but may be oblique or, rarely, horizontal.

Look for pigment dispersion syndrome

- Myopia.
- Deep anterior chamber.
- Midperipheral, radial, slit-like iris transillumination defects.
- Tiny pigment granules on iris surface.
- Midperipheral iris concavity.
- Posterior iris insertion.
- Trabecular hyperpigmentation.
- Predisposition to (pigmentary) glaucoma.
- Retinal lattice degeneration.

Fig. 5.169

Chlorpromazine

Long-term use of chlorpromazine may give rise to diffuse, granular, yellowish-white deposits at the level of the endothelium and deep stroma (**Figs 5.170, 5.171**).

Look for associated

- Anterior capsular lens deposits.
- Pigmentary retinopathy.

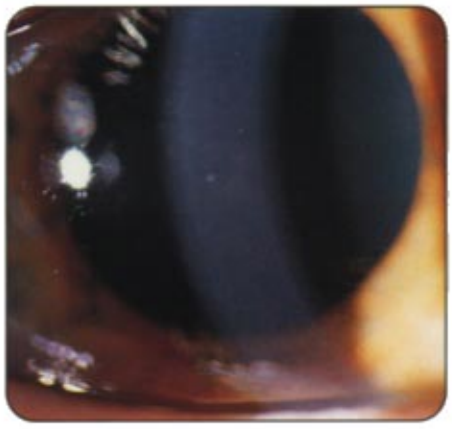


Fig. 5.170

Fig. 5.171

Dystrophies

Fuchs endothelial dystrophy

Fuchs endothelial dystrophy is an uncommon condition which usually occurs after the fifth decade.

Signs (in chronological order)

- Tiny central excrescences of Descemet membrane (cornea guttata) (**Figs 5.172, 5.173**).
- Confluence of lesions, giving rise to a 'beaten-metal' appearance (**Figs 5.174, 5.175**).
- Variable amount of pigment on the endothelium and a gray thickened appearance of Descemet membrane.
- Full-thickness stromal edema, giving rise to a 'ground-glass' appearance (**Fig. 5.176**).
- Epithelial edema and bullae (bullous keratopathy) (*see* **Fig. 5.27**).

Fig. 5.172

Fig. 5.173

Fig. 5.174

Fig. 5.175

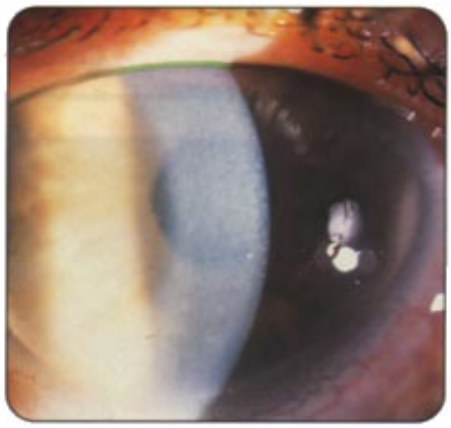


Fig. 5.176

Posterior polymorphous dystrophy

Posterior polymorphous dystrophy is an uncommon, dominantly inherited condition.

Signs

- Band-like (**Fig. 5.177**), vesicular (**Fig. 5.178**) or, geographic configurations on the posterior corneal surface. The lesions may be very subtle and easily overlooked.

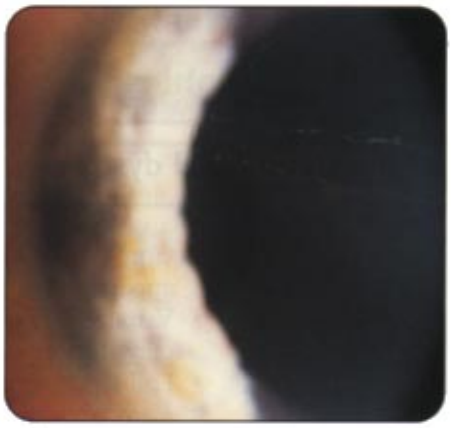


Fig. 5.177

Fig. 5.178

PERIPHERAL LESIONS

Noninflammatory

Vogt white limbal girdle

Vogt white limbal girdle is a very common, bilateral, innocuous, age-related condition.

Signs

- Crescentic, chalky white, linear opacities running in the interpalpebral fissures along the nasal and temporal limbus (**Fig. 5.179**).

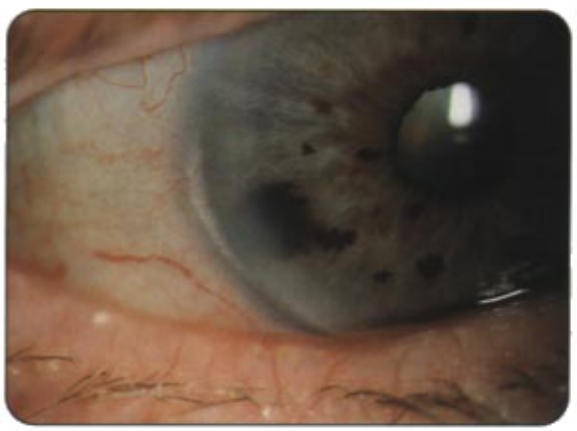


Fig. 5.179

Corneal arcus

Corneal arcus is a very common, bilateral condition which may be either age-related (arcus senilis) or associated with types II and III hyperlipidemia in younger individuals (arcus lipoides).

Signs

- Circumferential white limbal band about 1 mm in diameter with a sharp outline and a more diffuse central boundary (**Figs 5.180, 5.181**).

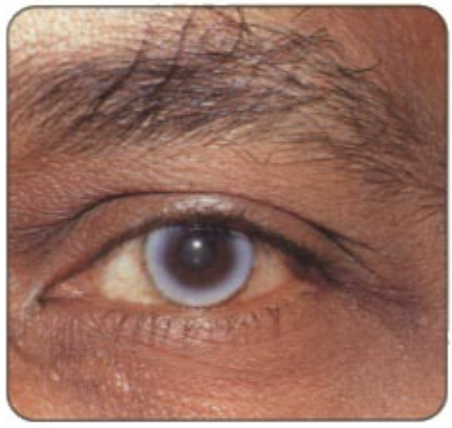


Fig. 5.180

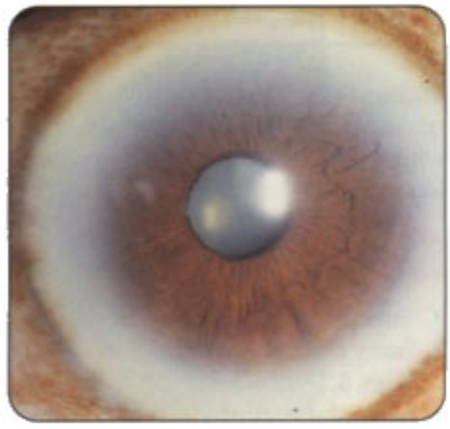


Fig. 5.181

Posterior embryotoxon

Posterior embryotoxon is a bilateral condition which occurs to some extent in about 15% of normals. It is present in 80% of patients with Alagille syndrome and in all patients with Axenfeld–Rieger anomaly.

Signs

- Thickening of an anteriorly displaced Schwalbe's line (**Fig. 5.182**).

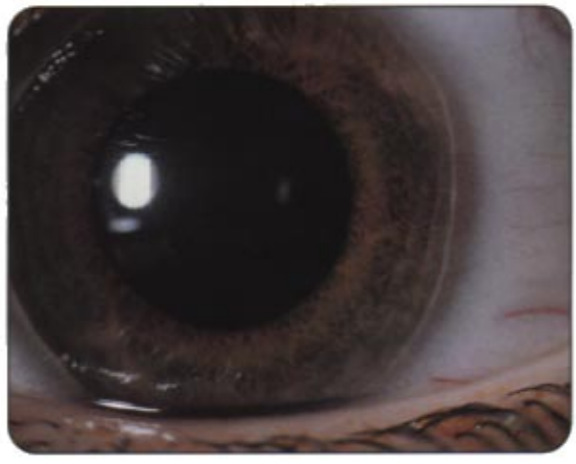


Fig. 5.182

Pseudogerontoxon in vernal disease (anterior embryotoxon)

Anterior embryotoxon is an uncommon, bilateral or unilateral condition which occurs at the site of previous inflammation, most frequently associated with vernal keratoconjunctivitis.

Signs

- Superior cornea opacity with a cupid's-bow configuration (**Fig. 5.183**).

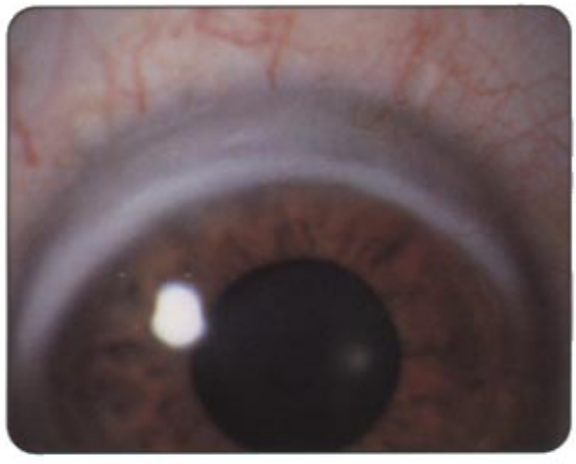


Fig. 5.183

Inflammatory peripheral infiltrates

Marginal keratitis

Marginal keratitis is a very common, transient, usually unilateral condition which may be associated with chronic staphylococcal blepharitis.

Signs

- Subepithelial infiltrate separated from the limbus by clear cornea (**Figs 5.184, 5.185**).
- Secondary epithelial breakdown (**Fig. 5.186**) is common.
- In severe cases the infiltration may spread circumferentially (**Fig. 5.187**).

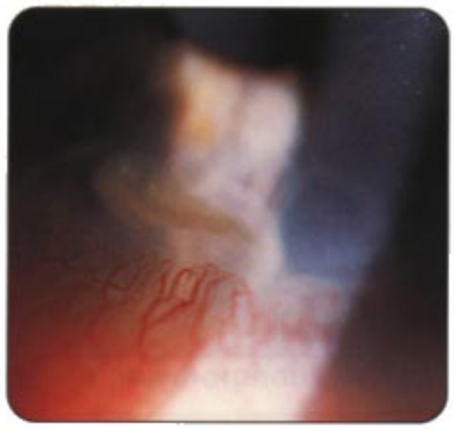


Fig. 5.184

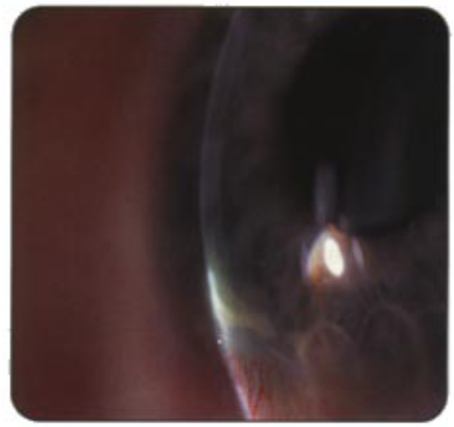


Fig. 5.185

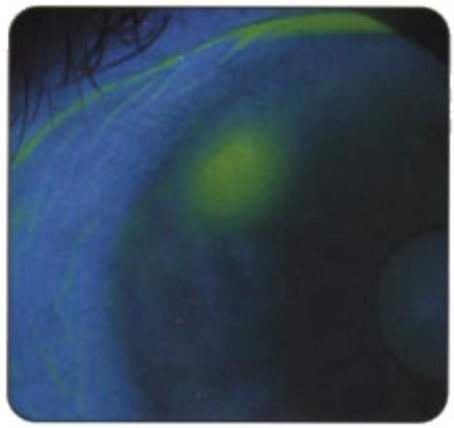


Fig. 5.186

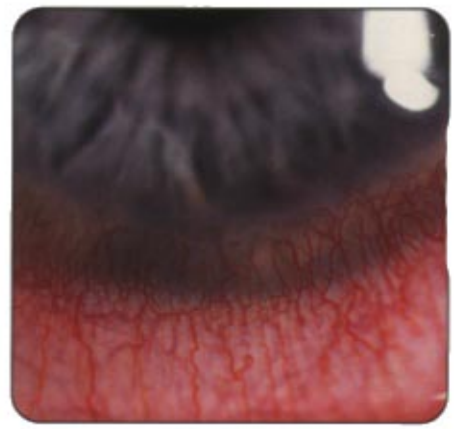


Fig. 5.187

Rosacea keratitis

Rosacea keratitis is a bilateral, chronic condition which affects approximately 5% of patients with acne rosacea.

Signs

- Peripheral inferior vascularization associated with subepithelial infiltrates central to the vessels (**Fig. 5.188**).

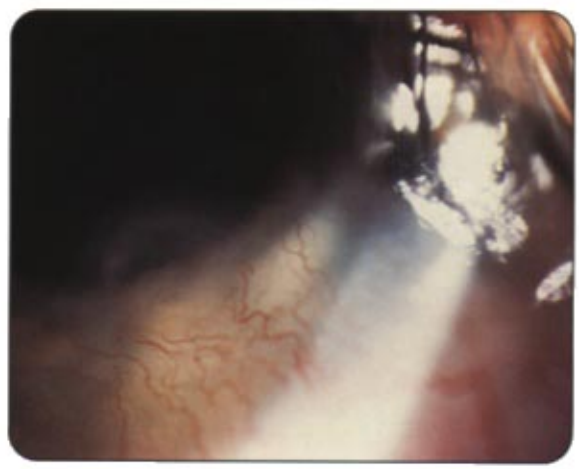


Fig. 5.188

Chlamydial keratitis

Chlamydial keratitis is an uncommon, usually bilateral, chronic condition associated with adult inclusion conjunctivitis.

Signs

- Multiple small peripheral infiltrates which usually involve the superior half of the cornea (**Fig. 5.189**).

Look for associated

- Chronic follicular conjunctivitis.
- Preauricular lymphadenopathy.

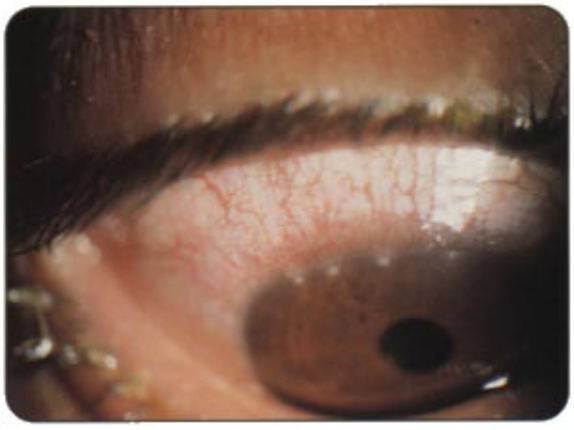


Fig. 5.189

Phlyctenulosis

Phlyctenulosis is an uncommon, usually unilateral condition.

Signs

- Small, pinkish-white limbal nodule (**Fig. 5.190**) which may resolve spontaneously or extend radially onto the cornea.

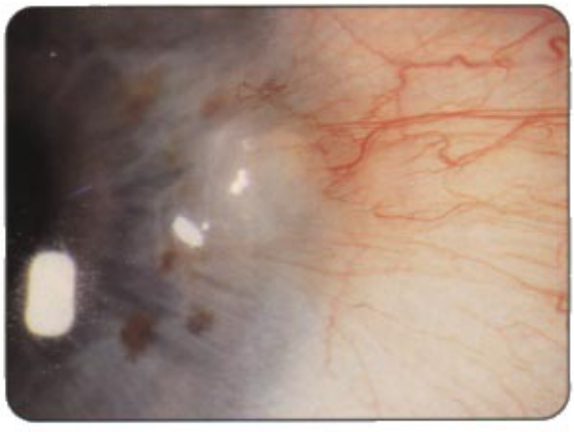


Fig. 5.190

Associated with exposure

These are uncommon, sterile infiltrates similar to those found in marginal keratitis but they only involve the inferior cornea (**Fig. 5.191**).

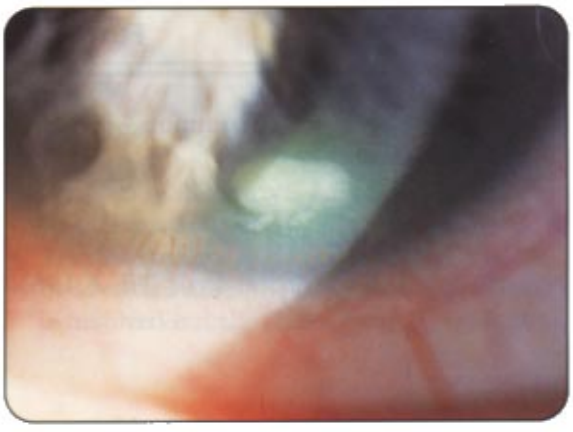


Fig. 5.191

Sclerosing keratitis

Sclerosing keratitis is a rare, chronic condition which may occur either in isolation or adjacent to scleritis.

Signs

- Gradual peripheral stromal thickening and opacification (**Fig. 5.192**).
- Vascularization and lipid deposition may occur.

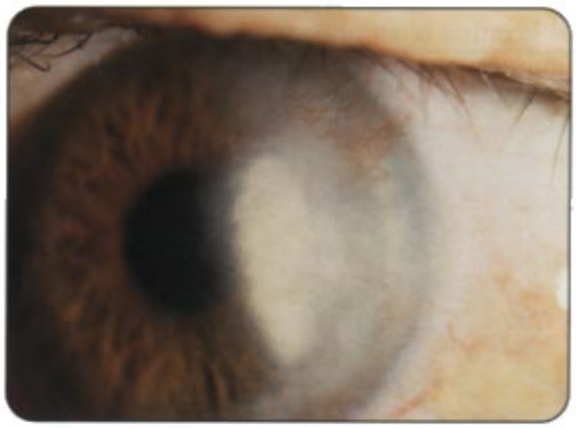


Fig. 5.192

Acute stromal keratitis

Acute stromal keratitis is a rare, acute condition which is usually associated with non-necrotizing scleritis.

Signs

- Peripheral superficial or midstromal infiltration (**Fig. 5.193**) which may be associated with epithelial breakdown (**Fig. 5.194**).

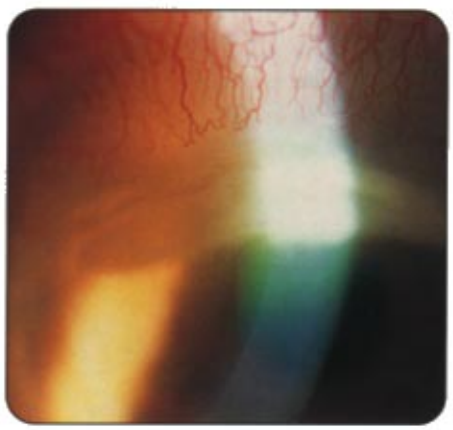


Fig. 5.193

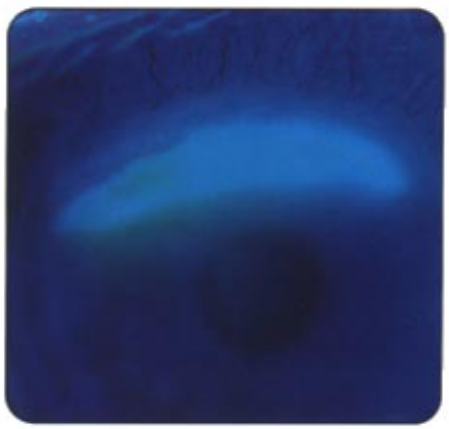


Fig. 5.194

Peripheral thinning not associated with systemic disease

Furrow degeneration

Furrow degeneration is a very common, bilateral, innocuous, age-related condition.

Signs

- Mild thinning peripheral to corneal arcus (**Fig. 5.195**).

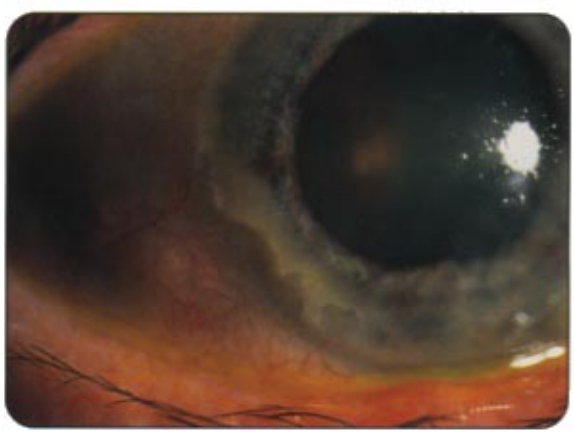


Fig. 5.195

Dellen

Dellen is a common, unilateral, innocuous condition.

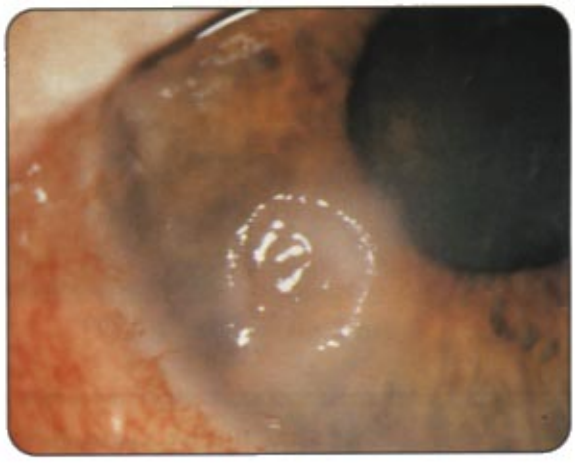


Fig. 5.196

Signs

- Saucer-like thinning secondary to local stromal dehydration (**Fig. 5.196**).
- Because the epithelium is intact the lesion does not stain with fluorescein although there may be pooling.

Causes

- Raised limbal lesions.
- Wear of hard contact lenses.
- Lid abnormalities and abnormal blinking mechanisms.
- Idiopathic in the elderly.

Terrien marginal degeneration

Terrien marginal degeneration is an uncommon, usually bilateral, painless condition.

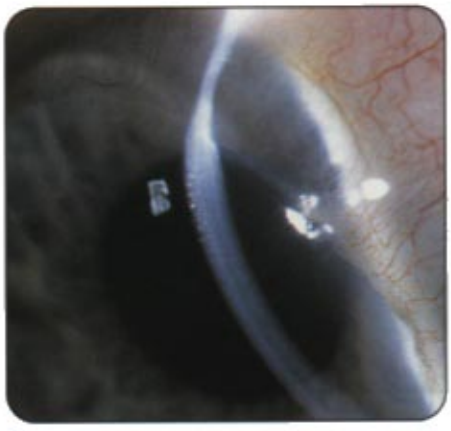


Fig. 5.197

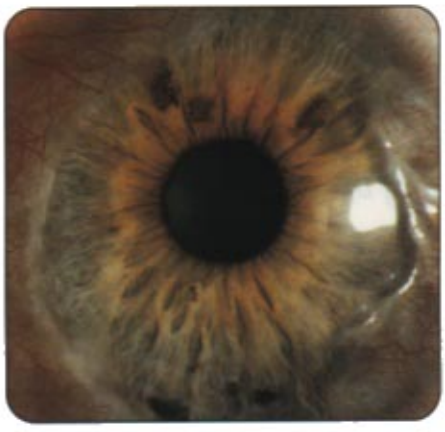


Fig. 5.198

Signs

- Peripheral thinning associated with superficial vascularization and lipid deposits central to the thinned edge (**Fig. 5.197**).
- Severe astigmatism and pseudopterygia (**Fig. 5.198**) develop in long-standing cases.

Mooren ulcer

Mooren ulcer is a very rare, unilateral or bilateral, painful condition.

Signs

- Peripheral ulcerative keratitis that undermines the epithelium and superficial corneal lamellae (**Fig. 5.199**).
- The ulceration spreads centrally (**Fig. 5.200**).
- In advanced cases the entire circumference of the cornea is involved but the sclera spared (**Fig. 5.201**).

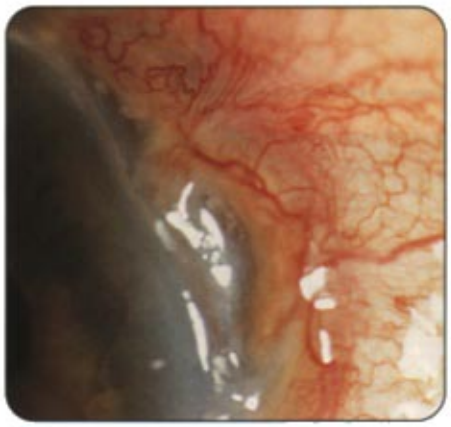


Fig. 5.199

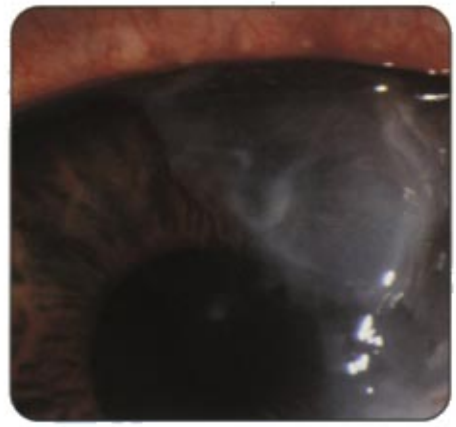


Fig. 5.200

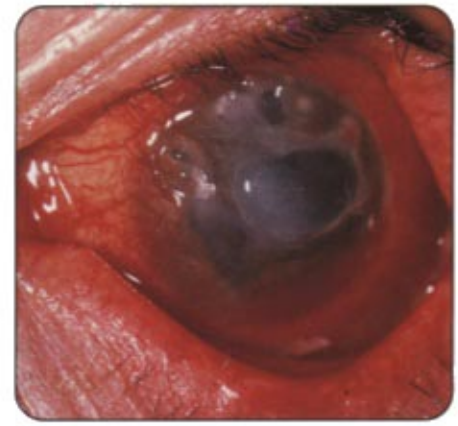


Fig. 5.201

Fuchs superficial marginal keratitis

Fuchs superficial marginal keratitis is an uncommon, unilateral, transient, recurrent, painful condition.

Signs

- Nonstaining, peripheral corneal infiltrates that resolve over a few days, leaving behind thinning, superficial vascularization and, occasionally, pseudopterygia (**Fig. 5.202**).

Differential diagnosis

- This disorder differs from Mooren ulcer because there is no epithelial defect.
- It differs from from Terrien marginal degeneration because there is no lipid line.

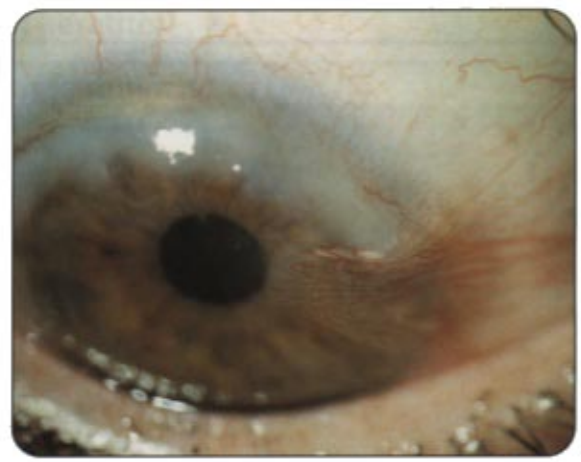


Fig. 5.202

Peripheral thinning associated with systemic diseases

Rheumatoid arthritis

Signs

- Contact-lens cornea is an asymptomatic, gradual, non-inflammatory resorption of peripheral cornea tissue with intact epithelium (**Fig. 5.203**).
- Acute peripheral melting is a severe corneal thinning which may occur in an area of contact-lens cornea unassociated with inflammation (**Figs 5.204, 5.205**).
- Acute peripheral corneal melting associated with inflammation (**Fig. 5.206**).

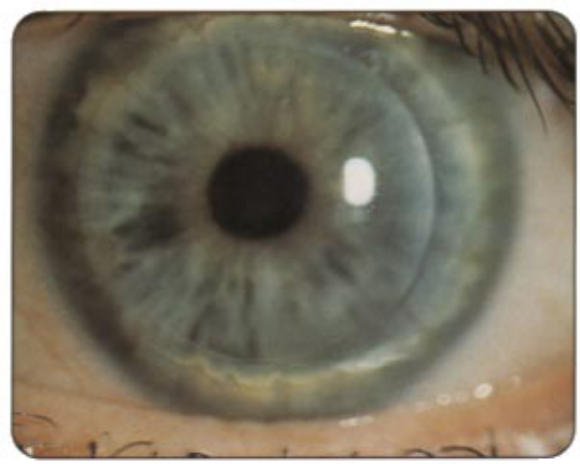


Fig. 5.203

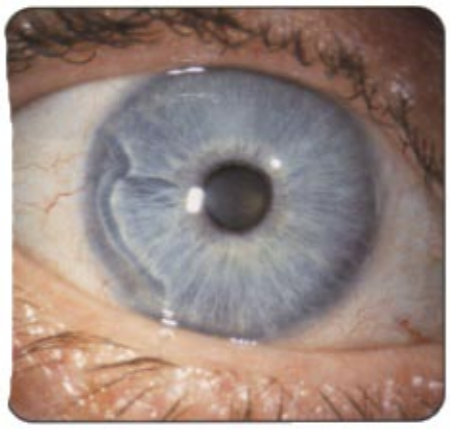


Fig. 5.204

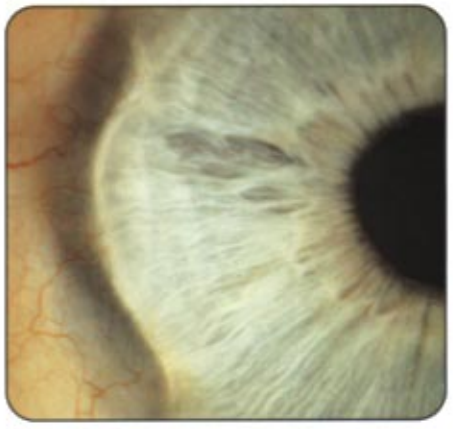


Fig. 5.205

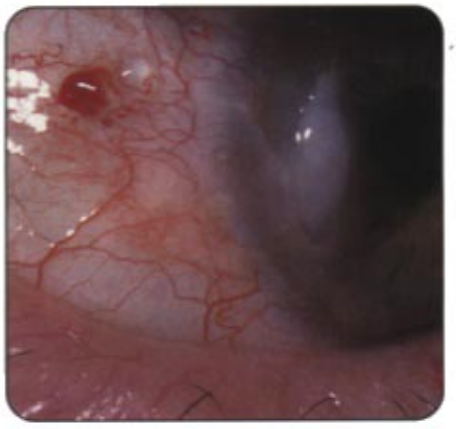


Fig. 5.206

Polyarteritis nodosa and Wegener granulomatosis

In polyarteritis and Wegener granulomatosis, the ulceration is similar to Mooren but the sclera is also involved (**Fig. 5.207**).

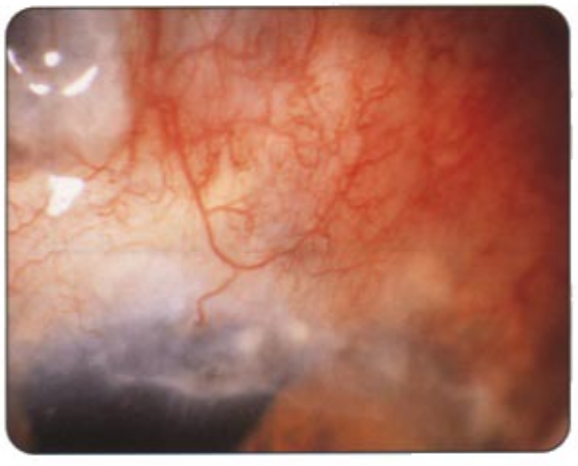


Fig. 5.207

Other diseases

- Systemic lupus erythematosus.
- Inflammatory bowel disease.
- Leukemia.
- Vitamin A deficiency.

6 The anterior chamber

VERY DEEP ANTERIOR CHAMBER 164

SHALLOW ANTERIOR CHAMBER 164

Following glaucoma filtration surgery
- Leaking bleb
- Excessive filtration
- Pupil block
- Malignant glaucoma

Miscellaneous causes

INFLAMMATORY AQUEOUS CELLS 167

Anterior uveitis

Infectious endophthalmitis
- Acute postoperative endophthalmitis
- Chronic postoperative endophthalmitis
- Delayed-onset endophthalmitis associated with a filtering bleb
- Endogenous infectious endophthalmitis

MISCELLANEOUS AQUEOUS CELLS 169
- Red cells (hyphema)
- Other cells

MISCELLANEOUS ANTERIOR CHAMBER SIGNS 171
- Aqueous flare
- Fibrinous exudate
- Hypopyon
- Pseudohypopyon
- Vitreous in anterior chamber
- Lens in anterior chamber
- Foreign bodies in anterior chamber
- Cholesterol crystals

VERY DEEP ANTERIOR CHAMBER

Figure 6.1 shows a very deep anterior chamber in an eye with megalocornea.

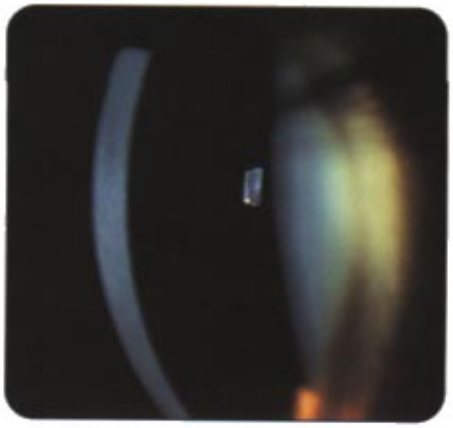


Fig. 6.1

Table 6.1 Causes of very deep anterior chamber

Large globe
• high myopia
• buphthalmos
Large cornea
• advanced keratoconus
• keratoglobus
• megalocornea
Abnormal lens
• aphakia
• pseudophakia
• ectopia lentis
Post-traumatic
• angle recession
• posterior rupture of globe
Pigment dispersion syndrome

SHALLOW ANTERIOR CHAMBER

Following glaucoma filtration surgery

Figure 6.2 shows a moderately shallow anterior chamber.
Figure 6.3 shows a very shallow anterior chamber.
Figure 6.4 shows a completely flat anterior chamber.

Fig. 6.2

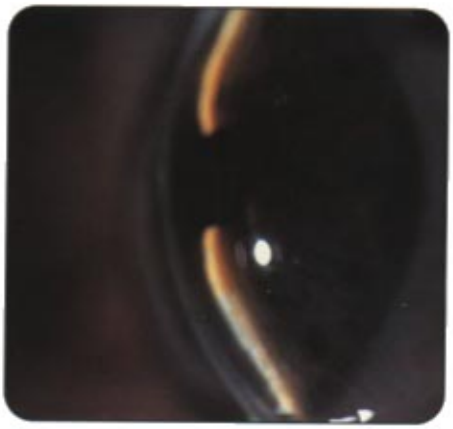


Fig. 6.3

Fig. 6.4

Leaking bleb

Look for

- Low intraocular pressure.
- Poor filtering bleb (**Fig. 6.5**).
- Patent iridectomy.
- Positive Seidel test (**Fig. 6.6**).

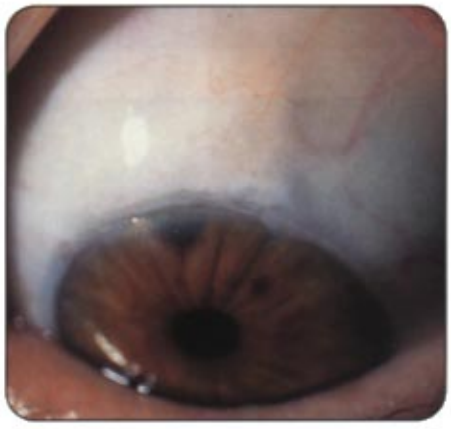


Fig. 6.5

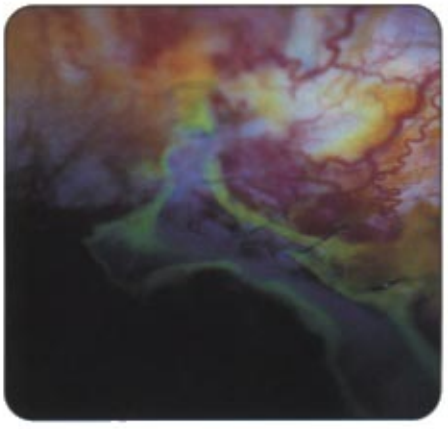


Fig. 6.6

Excessive filtration

Look for

- Low intraocular pressure.
- Large filtration bleb (**Fig. 6.7**).
- Patent iridectomy.
- Choroidal detachment.
- Negative Seidel test.

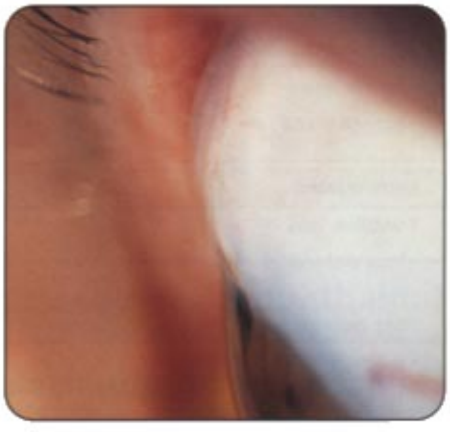


Fig. 6.7

Pupil block

Look for

- High intraocular pressure.
- Flat filtration bleb.
- Nonpatent iridectomy.
- Negative Seidel test.

Malignant glaucoma

Look for

- High intraocular pressure.
- No filtration bleb.
- Patent iridectomy.
- Negative Seidel test.

Miscellaneous causes

Figure 6.8 shows a flat anterior chamber caused by a penetrating injury of the cornea.
Figure 6.9 shows a shallow anterior chamber caused by iris bombè.
Figure 6.10 shows a shallow anterior chamber in Peters anomaly.
Figure 6.11 shows a shallow anterior chamber in an eye with iridoschisis.

Table 6.2 Miscellaneous causes of shallow anterior chamber

Small globe	**Postsurgical**
• hypermetropia	• leaking cataract section
• nanophthalmos	• pupil block by an anterior chamber lens
• microphthalmos	• expansile gas in vitreous cavity
Small cornea	• ciliochoroidal detachments following panretinal photocoagulation
• microcornea	**Other causes**
• cornea plana	• inflammatory pupil block and iris bombè
Lens-related	• ciliary-body tumors and cysts
• swollen lens	• Peters anomaly
• lens dislocated into anterior chamber	• iridoschisis
Post-traumatic	
• external leak	
• cyclodialysis cleft	

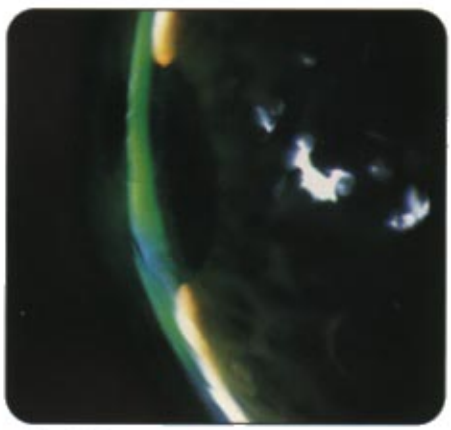


Fig. 6.8

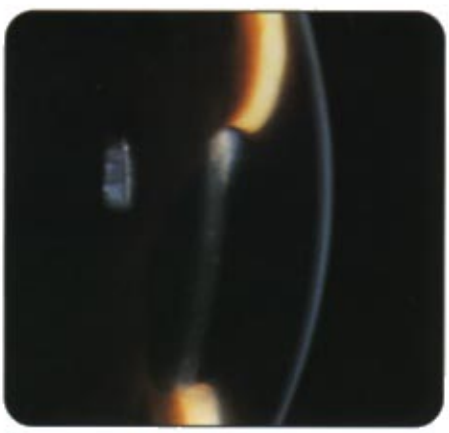


Fig. 6.9

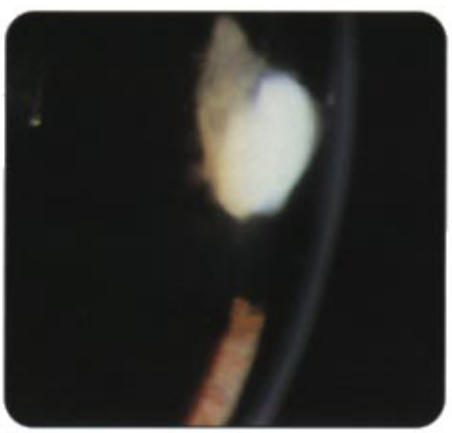


Fig. 6.10

Fig. 6.11

INFLAMMATORY AQUEOUS CELLS

Anterior uveitis

Figure 6.12 shows inflammatory white cells in the aqueous humor.

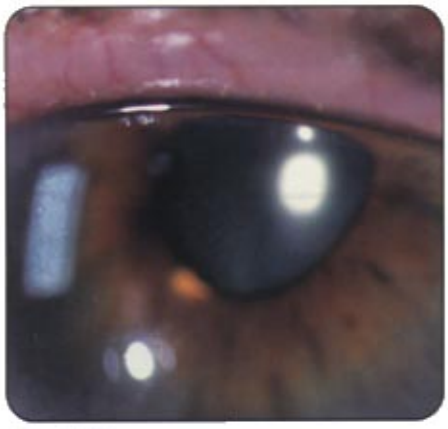


Fig. 6.12

Table 6.3 Associations and causes of anterior uveitis

With arthropathies	**Viral infections**
• ankylosing spondylitis	• herpes zoster
• Reiter syndrome	• herpes simplex
• psoriatic arthritis	**Other infections**
• juvenile chronic arthritis	• tuberculosis
• juvenile spondylitis	• syphilis
• relapsing polychrondritis	• Lyme disease
With bowel disease	**Miscellaneous**
• ulcerative colitis	• Fuchs heterochromic cyclitis
• Crohn disease	• phacoanaphylactic uveitis
• Whipple disease	
Other systemic diseases	
• sarcoidosis	
• Behçet disease	
• interstitial nephritis	

Infectious endophthalmitis

Acute postoperative endophthalmitis

Acute postoperative endophthalmitis usually develops during the first 5 days and is most often caused by Gram-positive bacteria. It is most frequently associated with cataract extraction.

Look for

- Fibrinous exudate.
- Hypopyon (**Fig. 6.13**).
- Vitritis.

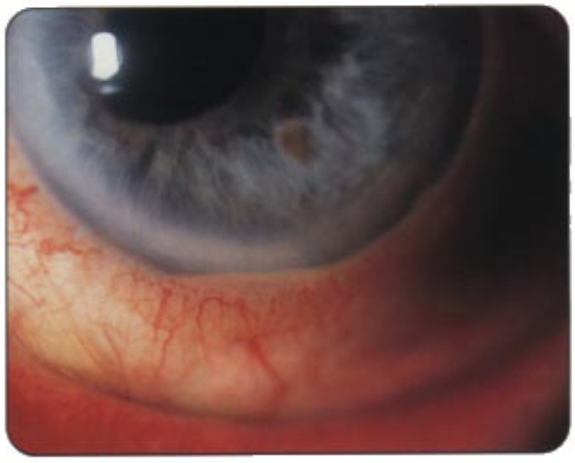


Fig. 6.13

Differential diagnosis

- Sterile fibrinous exudate (**Fig. 6.14**).
- Reaction to suture material (**Fig. 6.15**) which may give rise to anterior uveitis.

Fig. 6.14

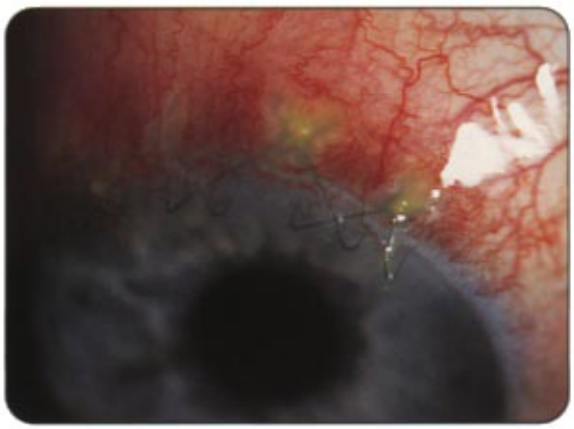


Fig. 6.15

Chronic postoperative endophthalmitis

Chronic postoperative endophthalmitis occurs several weeks or months after cataract surgery. It is caused by bacteria of low virulence such as *Propionibacterium acnes* and *Staphylococcus epidermidis*.

Look for

- Mutton-fat keratic precipitates (**Fig. 6.16**).
- White fluffy plaque on posterior capsule (**Fig. 6.17**).

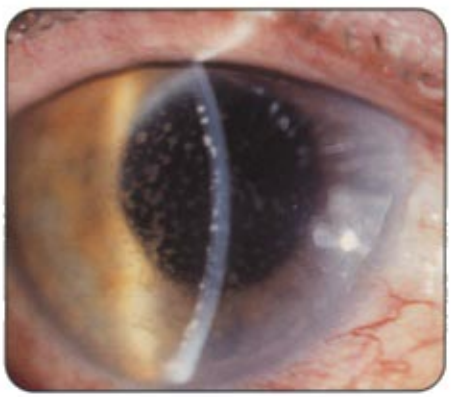


Fig. 6.16

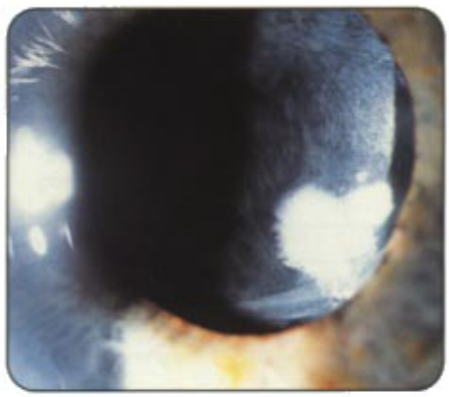


Fig. 6.17

Delayed-onset endophthalmitis associated with a filtering bleb

This most frequently occurs in eyes with thin cystic blebs (**Fig. 6.18**), particularly when associated with the use of adjunctive antimetabolites.

Signs

- White bleb surrounded by hyperemia (**Fig. 6.19, 6.20a**).
- Variable intraocular inflammation (**Fig. 6.20b**).

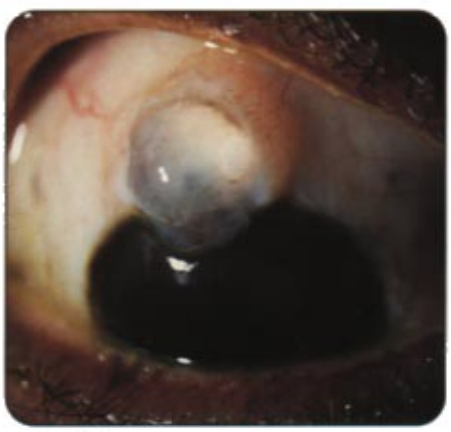


Fig. 6.18

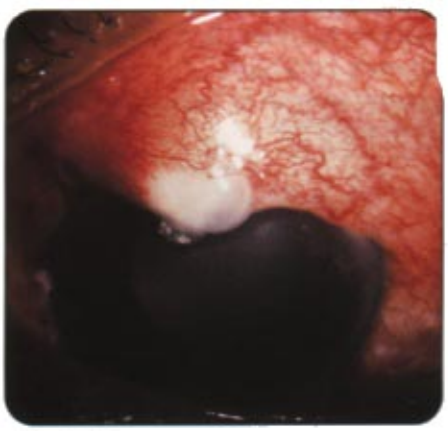


Fig. 6.19

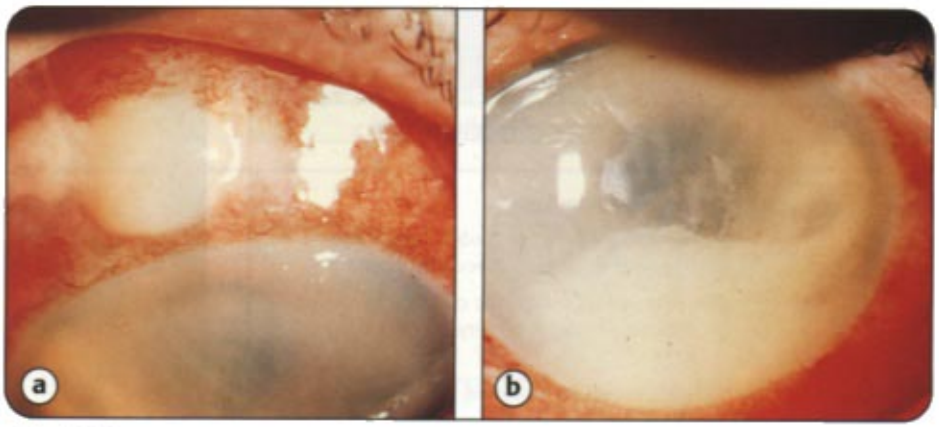


Fig. 6.20

Endogenous infectious endophthalmitis

Endogenous infectious endophthalmitis is rare and is usually associated with immunocompromised status as in the following:

- Following splenectomy.
- AIDS.
- Intravenous drug abuse.
- Diabetes mellitus.
- Septic arthritis.

MISCELLANEOUS AQUEOUS CELLS

Red cells (hyphema)

Figure 6.21 shows blood on the surface of a mature cataract.
Figure 6.22 shows a fresh spontaneous hyphema.
Figure 6.23 shows a hyphema associated with rubeosis iridis.
Figure 6.24 shows a postoperative hyphema.
Figure 6.25 shows a total hyphema caused by severe blunt ocular trauma.

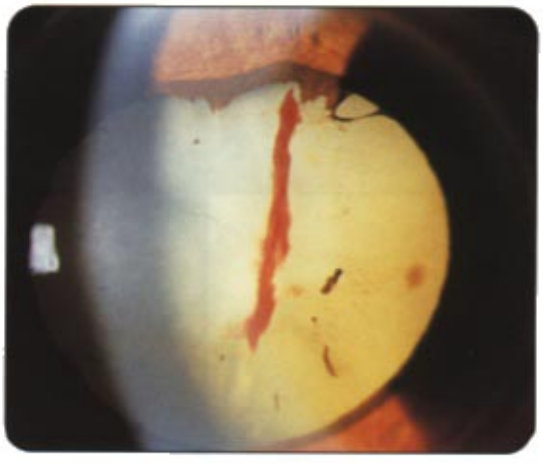


Fig. 6.21

Table 6.4 Causes of hyphema

Ocular trauma	**Iris and angle anomalies**
	• rubeosis
Surgery	• xanthogranuloma
	• pupillary vascular (Cobb) tufts – may give rise to spontaneous hyphema
Iritis	• Fuchs heterochromic cyclitis – hyphema during cataract surgery (Amsler sign)
• herpes simplex	
• herpes zoster	**Ghost cells associated with vitreous hemorrhage**
Blood dyscrasias	

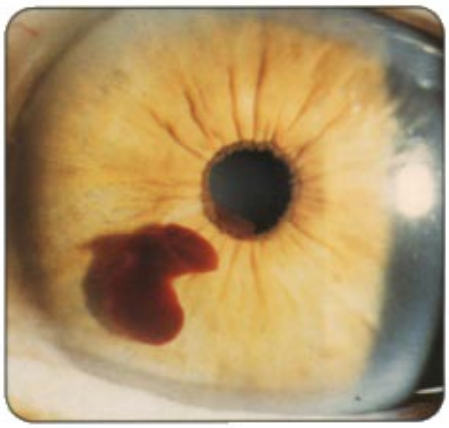


Fig. 6.22

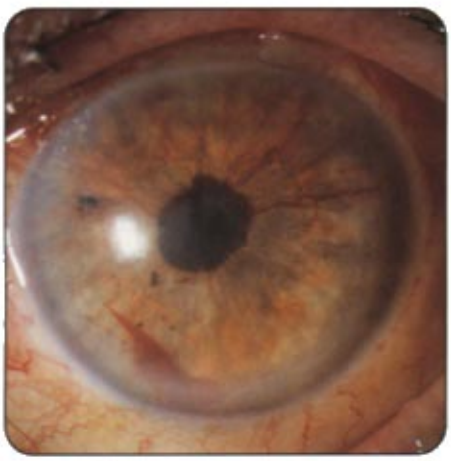


Fig. 6.23

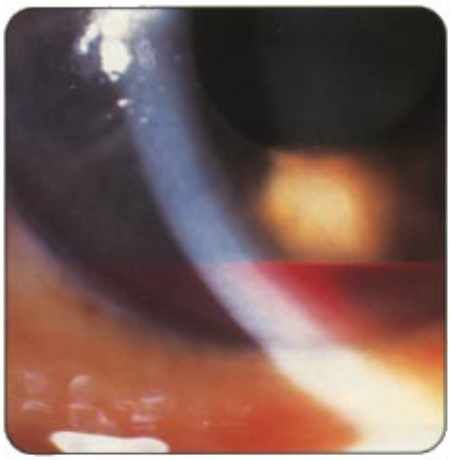


Fig. 6.24

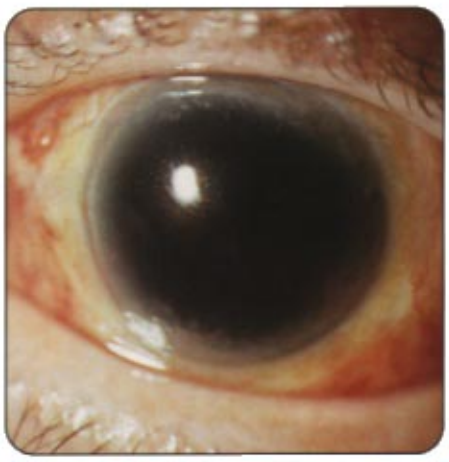


Fig. 6.25

Other cells

Figure 6.26 shows conglomerates of macrophages in phacolytic glaucoma.

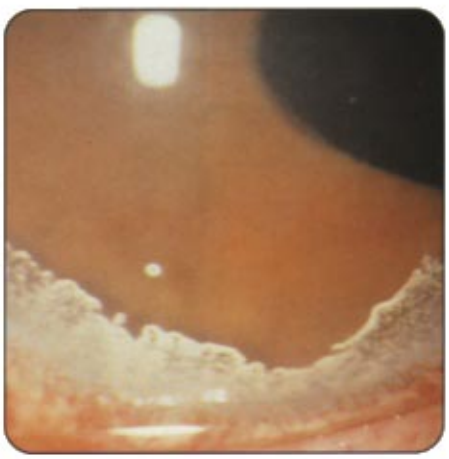


Fig. 6.26

Table 6.5 Miscellaneous aqueous cells

Pigment
• following mydriasis
• pigment dispersion syndrome
Neoplastic
• leukemia
• retinoblastoma
Macrophages in phacolytic glaucoma

MISCELLANEOUS ANTERIOR CHAMBER SIGNS

Aqueous flare

Causes

- Anterior uveitis – aqueous flare is in association with aqueous cells when inflammation is active but not when inactive (**Fig. 6.27**).
- Fluorescein flare following instillation in an eye with an epithelial abnormality.

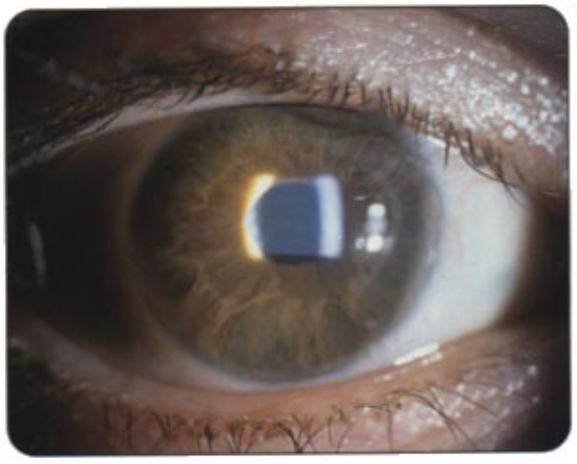


Fig. 6.27

Fibrinous exudate

Causes

- Severe noninfectious anterior uveitis (**Fig. 6.28**).
- Postoperative endophthalmitis.
- Sterile postoperative fibrinous reaction (*see* **Fig. 6.14**).

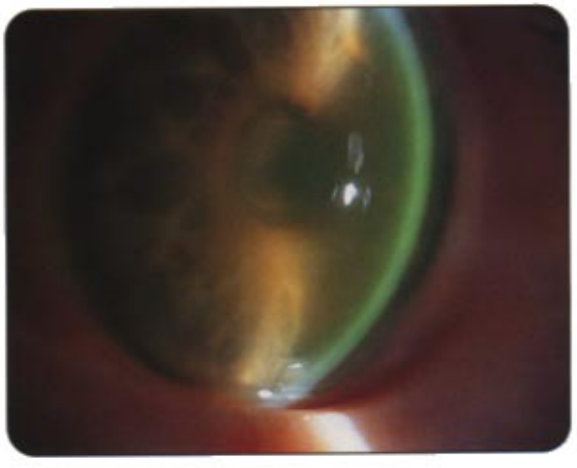


Fig. 6.28

Hypopyon

Causes

- Severe anterior uveitis – particularly associated with Behçet disease (**Fig. 6.29**).
- Endophthalmitis (**Fig. 6.30**).
- Severe infectious keratitis (**Fig. 6.31**).

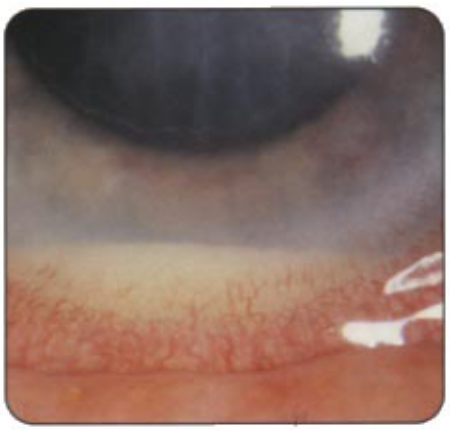


Fig. 6.29

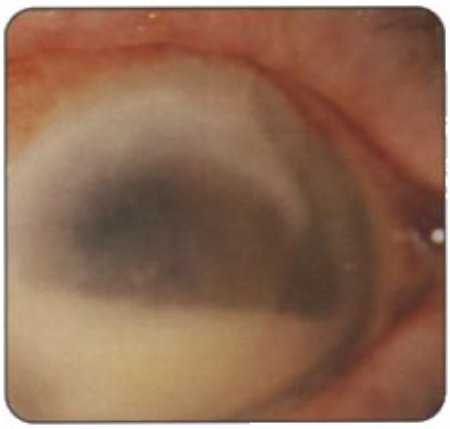


Fig. 6.30

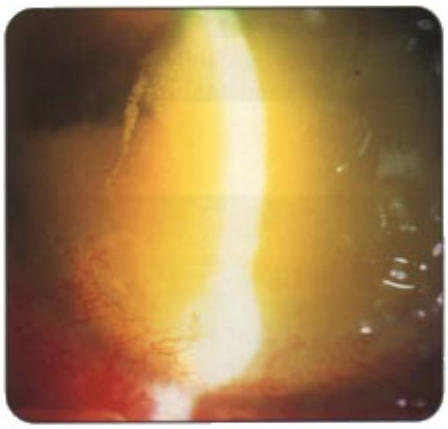


Fig. 6.31

Pseudohypopyon

Causes

- Tumor cells – particularly retinoblastoma in which the pseudohypopyon may be mixed with blood (**Fig. 6.32**).
- Emulsified silicone oil which may occasionally fill the entire anterior chamber (**Fig. 6.33**) or appear as an upside-down pseudohypopyon (**Fig. 6.34**).
- Severe phacolytic glaucoma.

Fig. 6.32

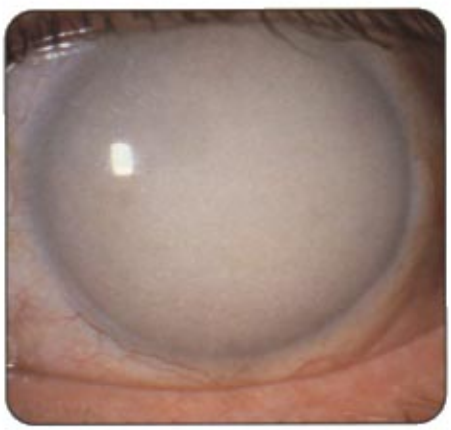


Fig. 6.33

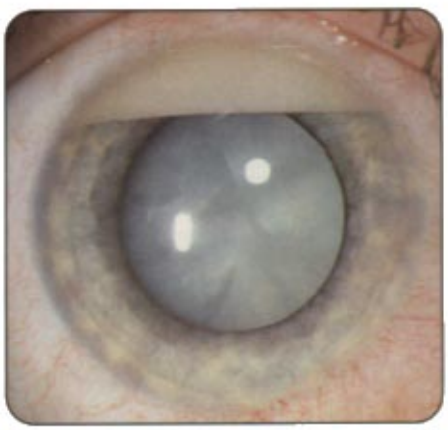


Fig. 6.34

Vitreous in anterior chamber

Causes

- Aphakia (**Fig. 6.35**).
- Lens subluxation – which may be spontaneous or traumatic.
- Following inadvertent vitreous prolapse during cataract surgery.

Fig. 6.35

Lens in anterior chamber

Causes

- Postsurgical retained lens material (**Figs 6.36, 6.37**).
- Dislocation of nearly entire lens into anterior chamber (**Fig. 6.38**).

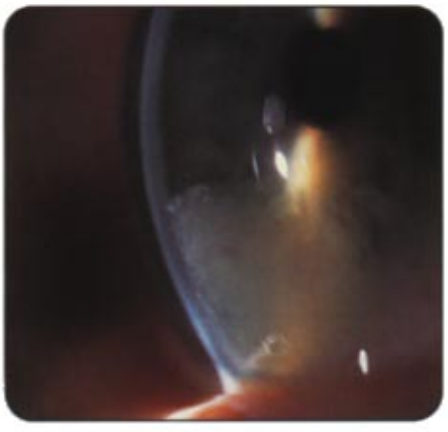


Fig. 6.36

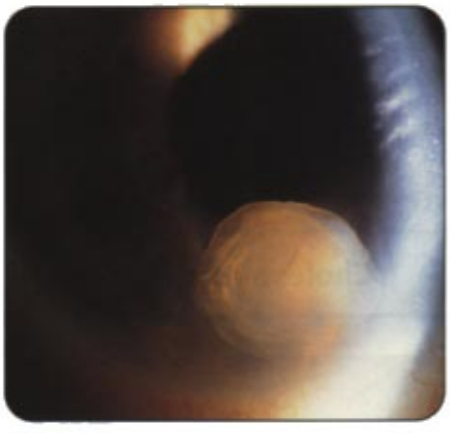


Fig. 6.37

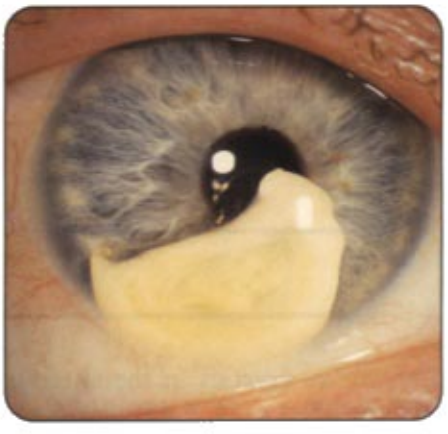


Fig. 6.38

Foreign bodies in anterior chamber

- Associated with penetrating injury such as those caused by glass (**Fig. 6.39**).
- Drainage tube (**Fig. 6.40**).
- Globules of silicone oil (**Fig. 6.41**).
- Dislocated artificial lens implants (**Fig. 6.42**).
- Lashes (**Fig. 6.43**).

Fig. 6.39

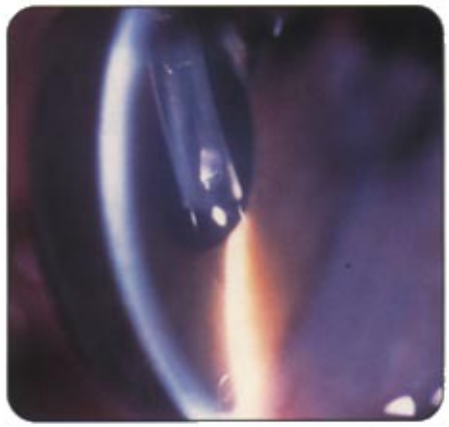


Fig. 6.40

Fig. 6.41

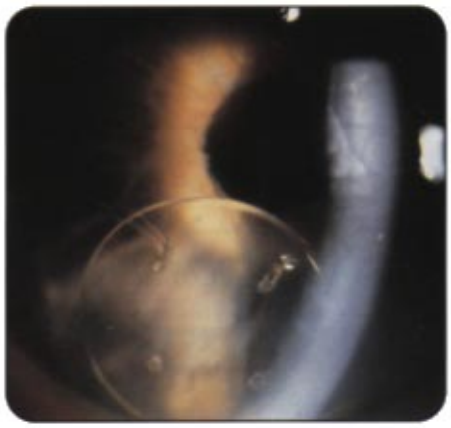


Fig. 6.42

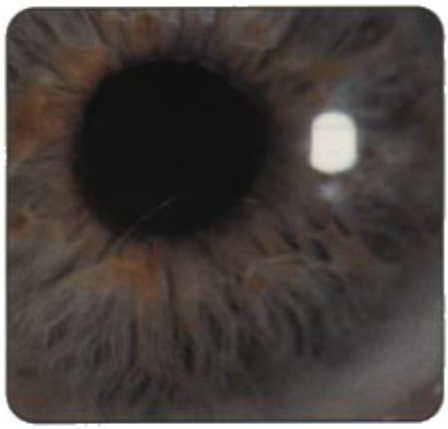


Fig. 6.43

Cholesterol crystals

Cholesterol crystals in the anterior chamber (**Figs 6.44, 6.45**) may occasionally develop following absorption of a hyphema.

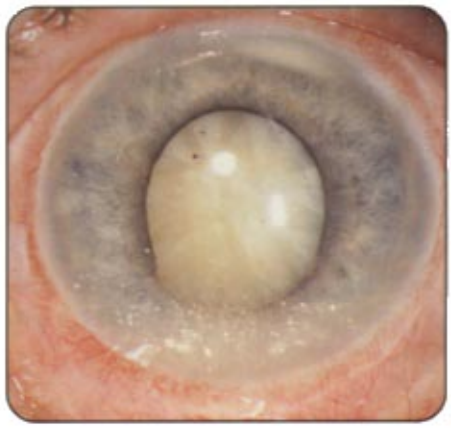


Fig. 6.44

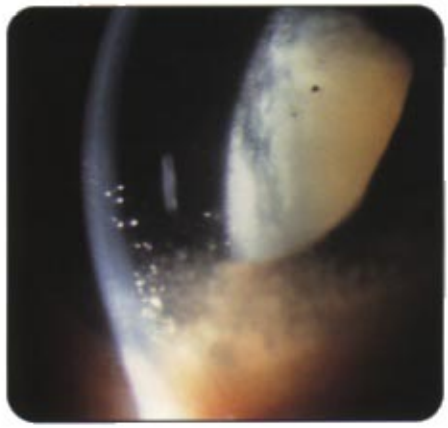


Fig. 6.45

7 The pupil

SMALL PUPIL (MIOSIS) 176

Unilateral miosis
Bilateral miosis

LARGE PUPIL (MYDRIASIS) 178

Unilateral mydriasis
Bilateral mydriasis

LIGHT–NEAR DISSOCIATION OF PUPILLARY REACTIONS 180

ABNORMALITIES OF PUPIL SHAPE 180

Congenital
Acquired

WHITE PUPIL IN CHILDHOOD (LEUKOCORIA) 182

Anterior lesions
Posterior lesions

SMALL PUPIL (MIOSIS)

Unilateral miosis

1. Physiologic anisocoria

Signs

- The difference in pupil size is independent of the level of illumination.

Figure 7.1a and b shows the pupils in bright light.
Figure 7.2a and b shows the pupils in the dark.

2. Sympathetic palsy (Horner syndrome)

Signs

- Miosis and mild ptosis (**Fig. 7.3**).
- The difference in pupil size is most apparent in dim light.
- The pupillary reactions are normal.

Associated features

- Elevation of lower lid.
- Heterochromia iridis – if the palsy is congenital.
- Anhidrosis – if the lesion is below the superior cervical ganglion.

3. Anterior uveitis

- In acute iritis, miosis is associated with pupillary spasm (**Fig. 7.4**).
- In chronic iritis, miosis may be caused by posterior synechiae (**Fig. 7.5**).

4. Other causes

- Longstanding Adie pupil ('little old Adie') which may be associated with diminished or absent deep-tendon reflexes (Holmes–Adie pupil).
- Pharmacologic miosis resulting from the unilateral use of a miotic drop. It may also be bilateral.

Table 7.1 Causes of Horner syndrome

Central lesions (first-order neuron)
• brainstem tumors
• syringomyelia
• lateral medullary (Wallenberg) syndrome
• spinal cord tumors
Preganglionic lesions (second-order neuron)
• neural crest tumors
• neck lesions
• intrathoracic lesions (Pancoast tumor, enlarged glands, aneurysm)
Postganglionic lesions (third-order neuron)
• cluster headache (migrainous neuralgia)
• nasopharyngeal tumors
• otitis media
• cavernous sinus mass
• internal carotid artery disease
Miscellaneous
• congenital
• idiopathic

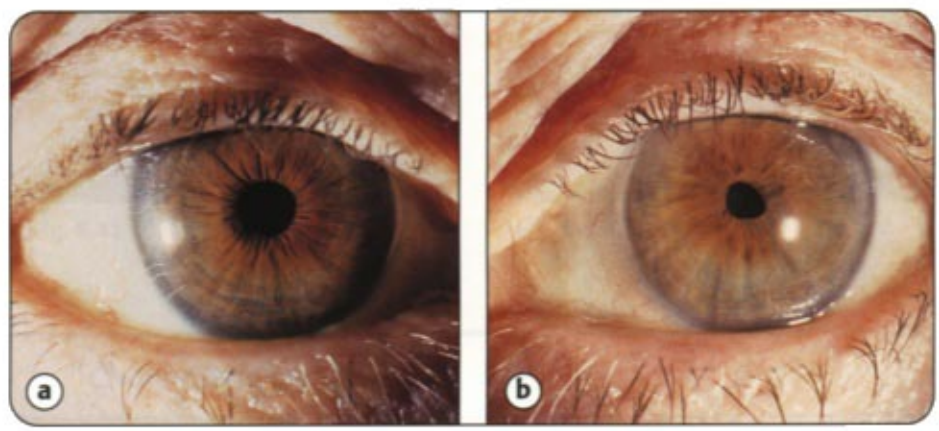


Fig. 7.1

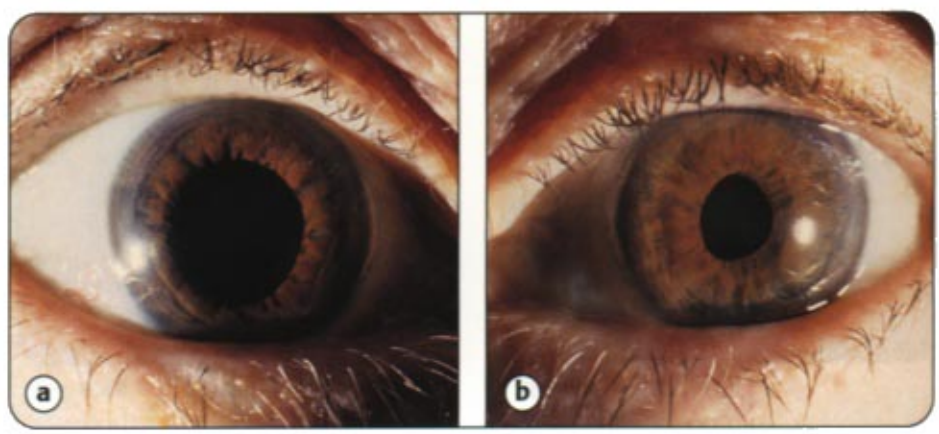


Fig. 7.2

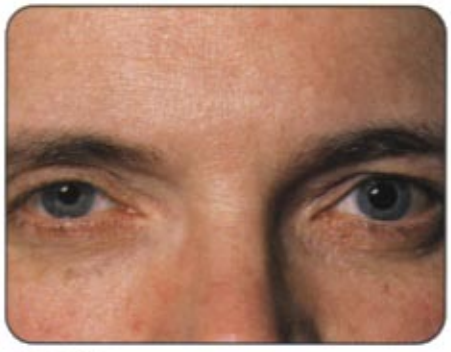


Fig. 7.3

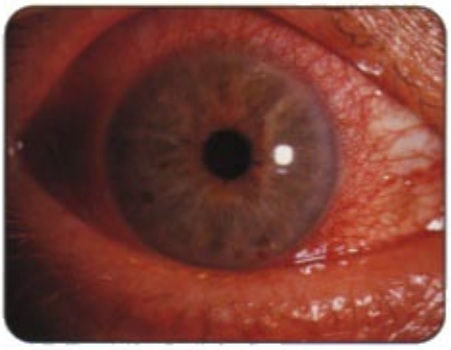


Fig. 7.4

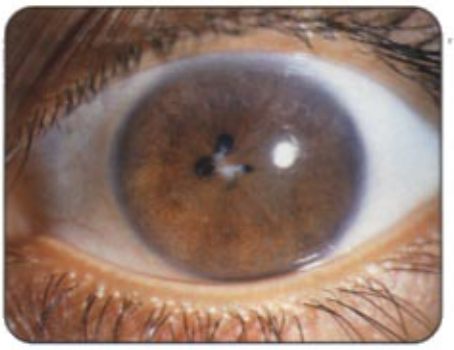


Fig. 7.5

Bilateral miosis

1. Physiologic miosis

This occurs in infancy and old age (senile miosis).

2. Pharmacologic miosis

Causes

- Topical miotics (**Fig. 7.6a and b**).
- Systemic drugs such as morphine, and pesticides.

3. Argyll Robertson (syphilitic) pupil

Look for

- Irregular pupils.
- Light–near dissociation.

4. Spasm of the near reflex

Look for

- Esotropia.
- Myopia.

5. Myotonic dystrophy

Look for

- Light–near dissociation.
- Bilateral symmetric ptosis.
- Symmetric ophthalmoplegia.
- Presenile posterior stellate cataracts.
- Meshlike peripheral pigment clumping.

6. Lepromatous miosis

The pupils may be small in the absence of posterior synechiae (**Fig. 7.7**).

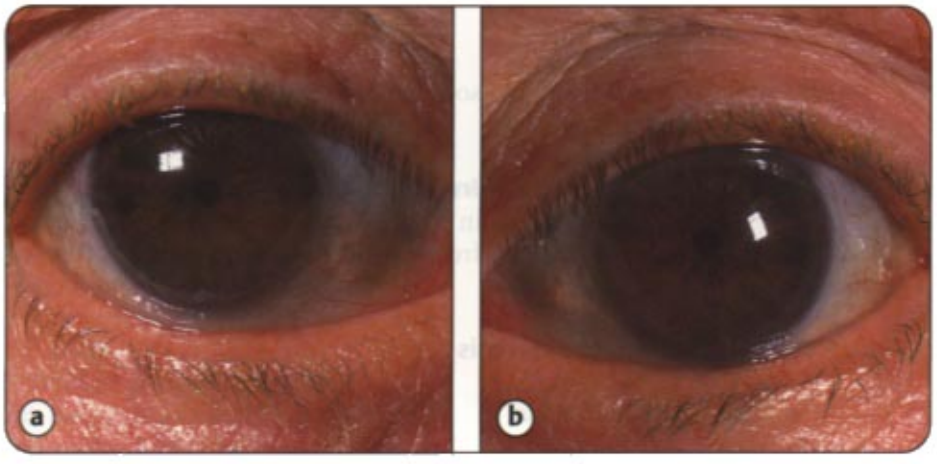


Fig. 7.6

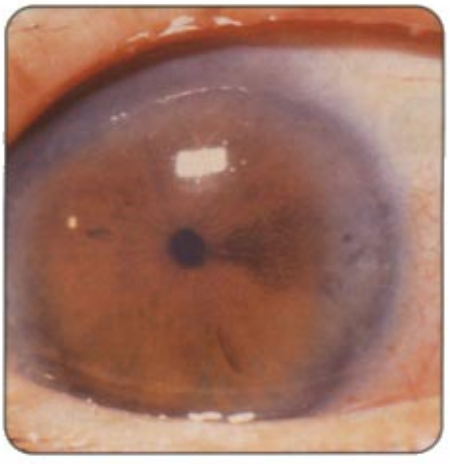


Fig. 7.7

7. Congenital microcoria

Look for

- Shallow anterior chamber.
- Glaucoma.
- Myopia.

8. Ectopia lentis et pupillae

An uncommon, congenital, recessively inherited condition which is not associated with systemic abnormalities.

Signs

- Corectopia (displacement of the pupil) (**Fig. 7.8a and b**).
- The pupil and the lens are displaced in opposite directions.

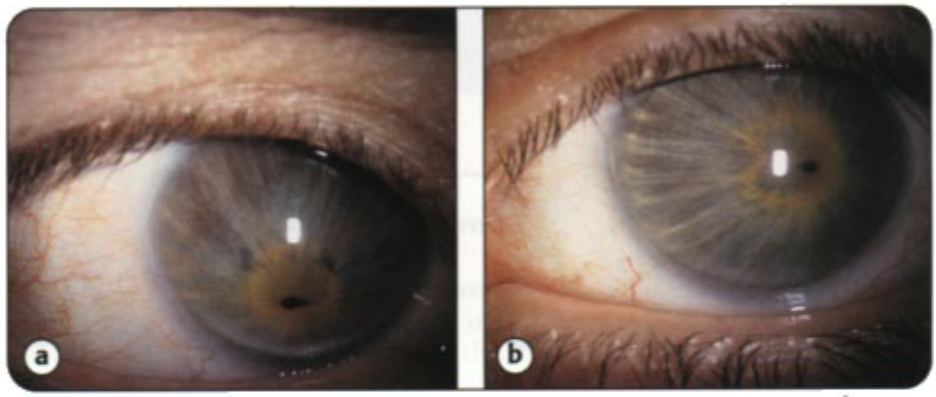


Fig. 7.8

9. Refsum disease

Look for

- Pigmentary retinopathy.
- Cataract.

LARGE PUPIL (MYDRIASIS)

Unilateral mydriasis

1. Physiologic anisocoria – *see* Figs 7.1 and 7.2

2. Pharmacologic mydriasis

This is caused by topical mydriatics. It may also be bilateral.

3. Adie pupil

A common, innocuous, usually unilateral condition.

Look for

- Absent or minimal light reflex.
- Slow reaction to near, with tonic re-dilatation.
- Slow accommodation.
- Vermiform movements of the iris on slit-lamp examination.
- Diminished or absent deep-tendon reflexes (Holmes–Adie pupil).
- Constriction with 0.125% pilocarpine.

4. Third nerve palsy

This is characterized by ptosis and ophthalmoplegia (**Fig. 7.9**).

5. Uncal herniation

This is associated with altered level of consciousness.

6. Traumatic sphincter paralysis

This may be associated with sphincter tears (**Fig. 7.10**) and iridodialysis.

7. Iris ischemia

This is usually associated with iris atrophy (**Fig. 7.11** and **Table 7.2**).

8. Postsurgical (Urrets–Zavalia syndrome)

A rare condition in which the pupil becomes dilated and unreactive, most frequently following corneal grafting for keratoconus.

9. Ocular siderosis

Look for

- Iris hyperchromia and cataract (**Fig. 7.12**).
- Glaucoma.
- Pigmentary retinal degeneration.

10. Idiopathic alternating anisocoria

This is a very rare condition in which the pupils dilate in an alternating fashion and show a poor or absent light reflex during the dilated phase. The dilatation lasts for 1–2 hours.

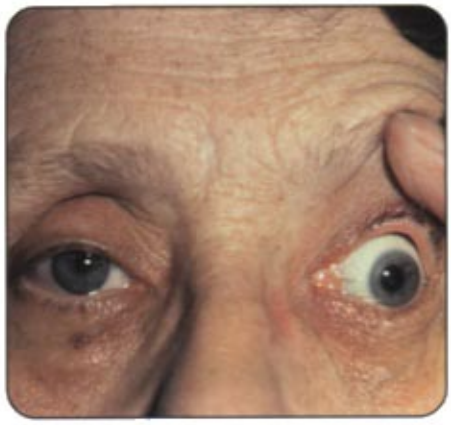


Fig. 7.9

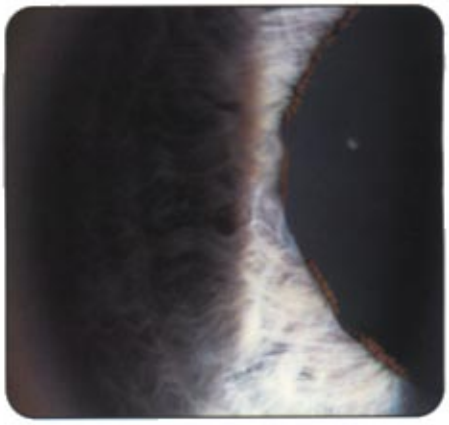


Fig. 7.10

Table 7.2 Causes of iris ischemia

Acute angle-closure glaucoma
Anterior segment ischemia
• squint surgery involving several muscles
• retinal detachment surgery with encirclement
• carotid artery disease

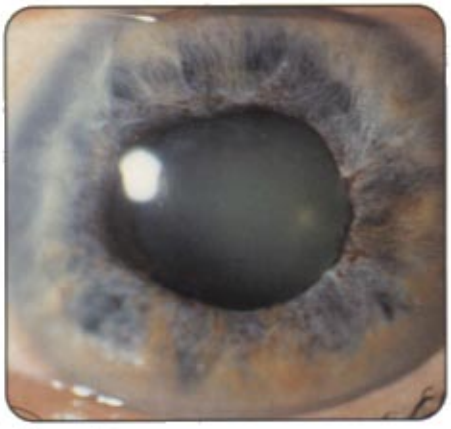


Fig. 7.11

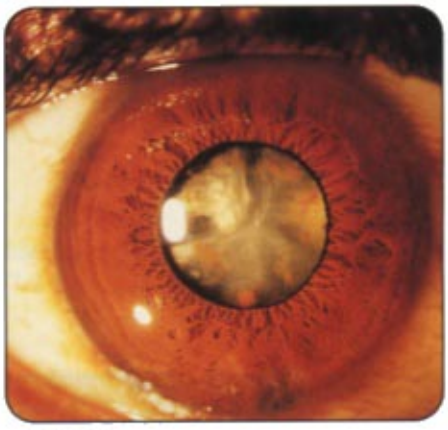


Fig. 7.12

Bilateral mydriasis

1. Pharmacologic mydriasis

Figure 7.13 shows pharmacological mydriasis, which may also be unilateral.

2. Parinaud dorsal midbrain syndrome (see Table 7.3)

Look for

- Light–near dissociation.
- Lid retraction (Collier sign).
- Paralysis of upgaze and convergence.
- Convergence–retraction nystagmus.

3. Benign periodic mydriasis

An uncommon condition that most frequently occurs in young people in which the pupils become dilated for several days.

4. Brainstem death

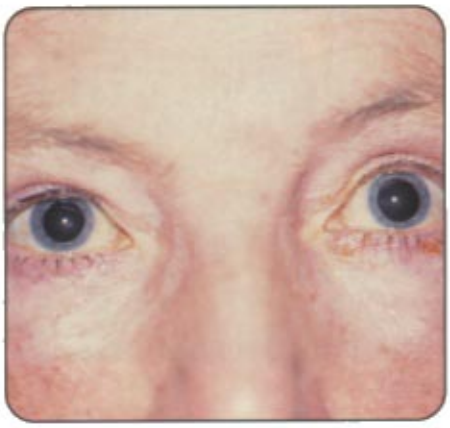


Fig. 7.13

Table 7.3 Causes of Parinaud syndrome

In children
• aqueduct stenosis
• meningitis
• pinealoma
In young adults
• demyelination
• trauma
• arteriovenous malformations
In the elderly
• midbrain vascular accidents
• mass lesions involving periaqueductal gray matter
• giant posterior fossa aneurysms

LIGHT–NEAR DISSOCIATION OF PUPILLARY REACTIONS

In light–near dissociation of pupillary reactions the near response is better than the light reflex. There is no condition in which the reverse is true. The causes are shown in **Table 7.4**.

Table 7.4 Causes of light–near dissociation

Unilateral
• afferent conduction defect
• herpes zoster ophthalmicus
• aberrant regeneration of the third nerve
Bilateral
• juvenile-onset diabetes
• myotonic dystrophy
• Parinaud dorsal midbrain syndrome
• Argyll Robertson pupils
• pituitary tumors
• familial amyloidosis
• encephalitis
• chronic alcoholism

ABNORMALITIES OF PUPIL SHAPE

Congenital

1. Ectopia lentis et pupillae (*see* Fig. 7.8)

2. Axenfeld–Rieger anomaly
Axenfeld–Rieger anomaly is shown in **Figure 7.14**.

3. Persistent pupillary membranes
There are two types:
- Type 1 – cobweb-like strands across the pupil (**Figs 7.15, 7.16**).
- Type 2 – iridolenticular adhesions (**Fig. 7.17**).

4. Iris coloboma
Iris coloboma is shown in **Figure 7.18** (*see* Chapter 8).

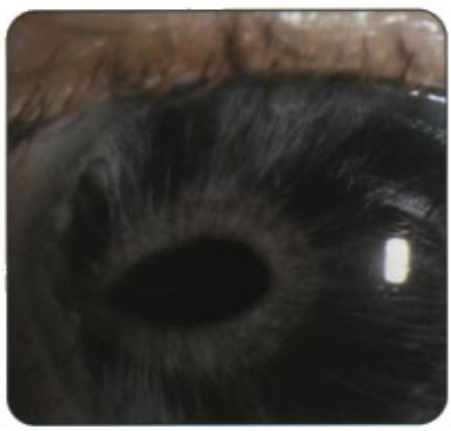


Fig. 7.14

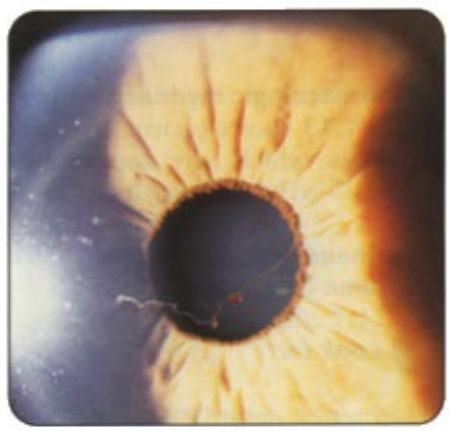


Fig. 7.15

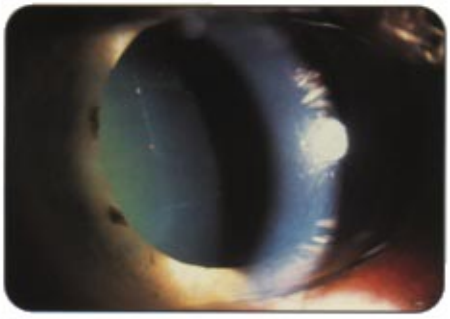


Fig. 7.16

Fig. 7.17

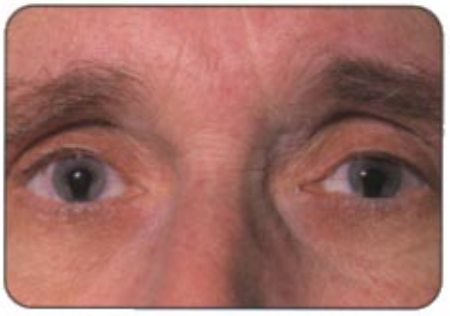


Fig. 7.18

Acquired

1. Inflammatory

The pupil has an irregular shape due to the development of posterior synechiae (**Fig. 7.19**).

2. Surgical

Causes

- Sphincterotomy.
- Iridectomy.
- Iris prolapse (**Fig. 7.20**).
- Updrawn pupil following cataract surgery (**Fig. 7.21**).
- Intraocular implants, particularly anterior chamber (**Fig. 7.22**).
- Sphincter damage during cataract surgery (**Fig. 7.23**).

3. Traumatic

Causes

- Iris prolapse.
- Iris sphincter damage.
- Iridodialysis (**Fig. 7.24**).

4. Iris tumors

Both malignant (**Fig. 7.25**) and, occasionally, benign (**Fig. 7.26**) tumors may alter the shape of the pupil.

5. Iridocorneal–endothelial (ICE) syndrome (*see* Chapter 8)

6. Argyll Robertson pupils

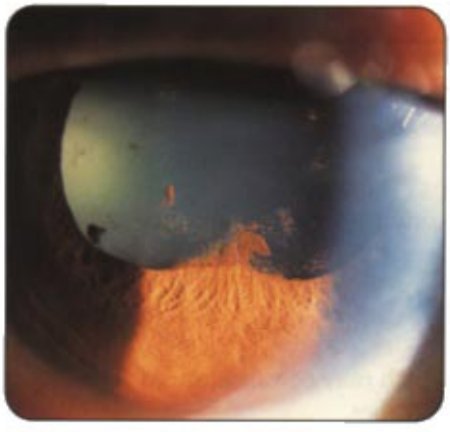


Fig. 7.19

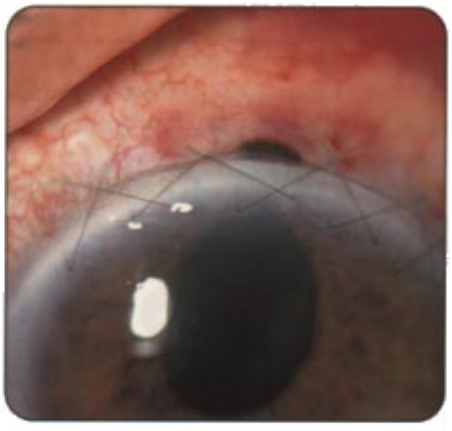


Fig. 7.20

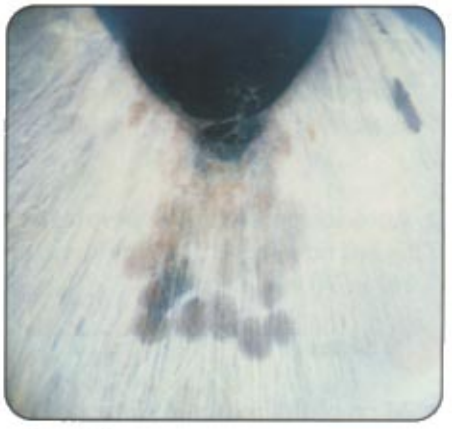


Fig. 7.21

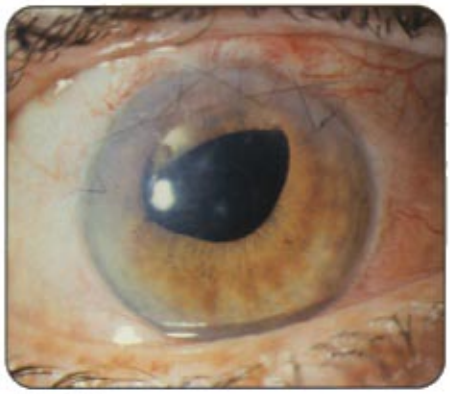


Fig. 7.22

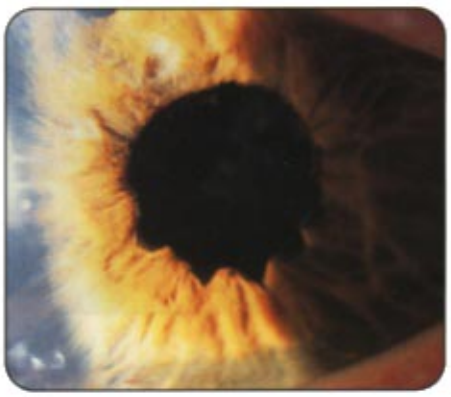


Fig. 7.23

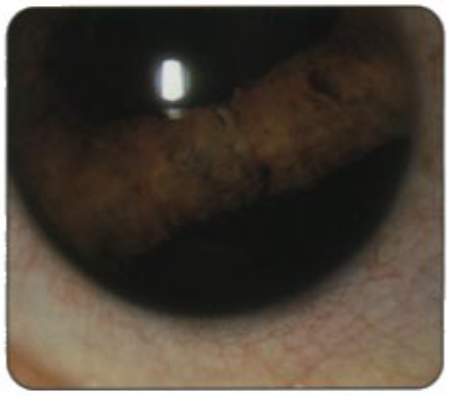


Fig. 7.24

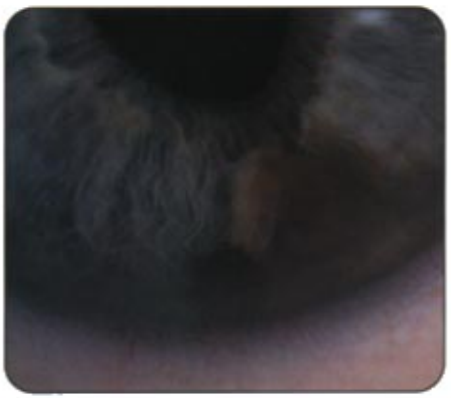


Fig. 7.25

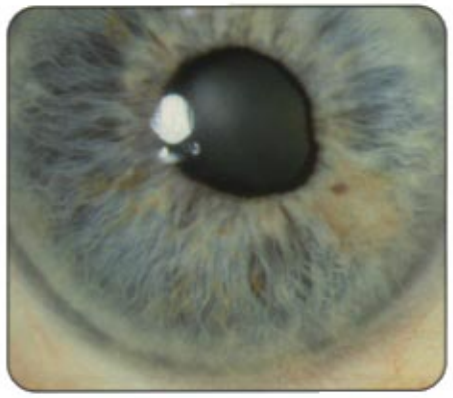


Fig. 7.26

WHITE PUPIL IN CHILDHOOD (LEUKOCORIA)

Anterior lesions

1. Congenital cataract

This may be unilateral or bilateral (**Fig. 7.27**) (*see* Chapter 9).

2. Anterior persistent hyperplastic primary vitreous

This is a rare congenital condition which is unilateral (**Fig. 7.28**) in 90% of cases.

Signs

- Microphthalmos and elongated ciliary processes inserted into a retrolental mass (**Fig. 7.29**).

3. Inflammatory cyclitic membrane

- Chronic toxocaral endophthalmitis (**Fig. 7.30**) is always unilateral and presents between the ages of 2 and 9 years.
- Intermediate uveitis is usually bilateral but may be asymmetric. It is uncommon before the age of 10 years. It may give rise to leukocoria either by cyclitic membrane formation or as a result of severe vitreous inflammation and condensation (**Fig. 7.31**).

4. Retinal dysplasia

This is a very rare condition which presents at birth or soon after.

Signs

- Retrolental mass which may be vascularized (**Fig. 7.32**).
- Shallow anterior chamber.
- Clear lens.

Systemic associations of bilateral cases

- Norrie disease.
- Bloch–Sulzberger syndrome (incontinentia pigmenti).
- Warburg syndrome.
- Patau syndrome (trisomy 13).
- Edward syndrome (trisomy 18).

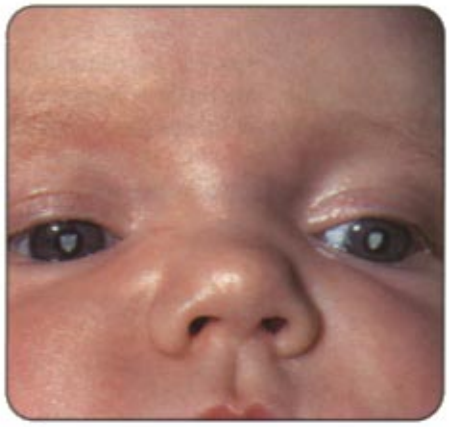


Fig. 7.27

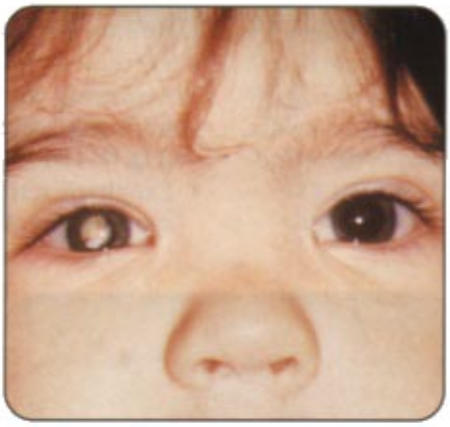


Fig. 7.28

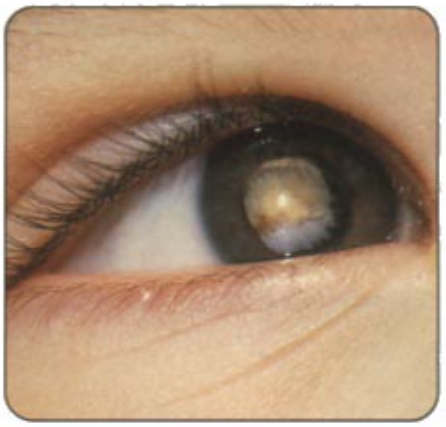


Fig. 7.29

Fig. 7.30

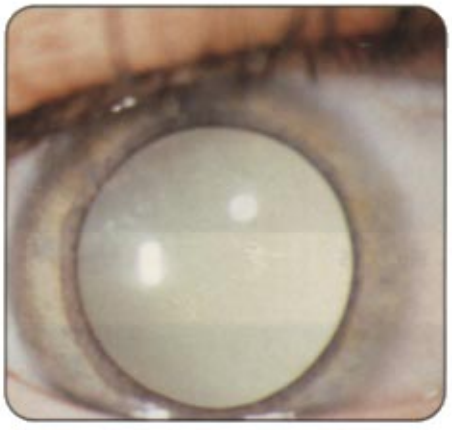


Fig. 7.31

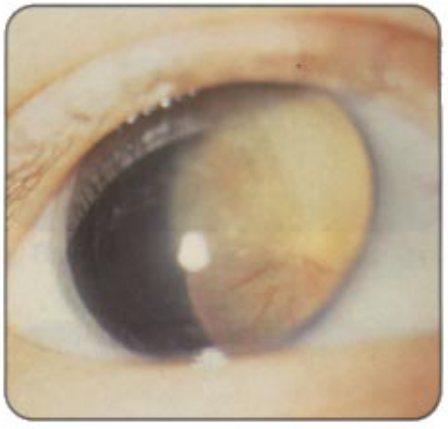


Fig. 7.32

Posterior lesions

1. Tumors and granulomas

- Retinoblastoma is bilateral in 33% of cases and may be multifocal. The average age at diagnosis is 18 months. Leukocoria (**Fig. 7.33**) may be caused by endophytic retinoblastoma which grows into the vitreous cavity (**Figs 7.34–7.36**).

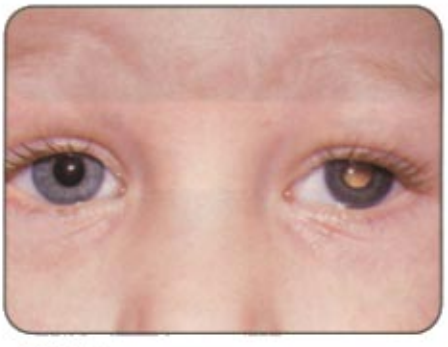


Fig. 7.33

- Retinal astrocytoma (**Figs 7.37, 7.38**) is bilateral in 15% of cases and may be multifocal. It is frequently associated with tuberous sclerosis.
- Posterior-pole toxocaral granuloma (**Fig. 7.39**) is always unilateral. It may mimic (**Fig. 7.40**) endophytic retinoblastoma but presents later (at 6–14 years).

2. Retinal detachment

- Retinopathy of prematurity is always bilateral but frequently asymmetric. Retinal detachment may occur during the acute stage or later as a result of cicatrization (**Fig. 7.41**).
- Exophytic retinoblastoma which grows in the subretinal space (**Fig. 7.42**).

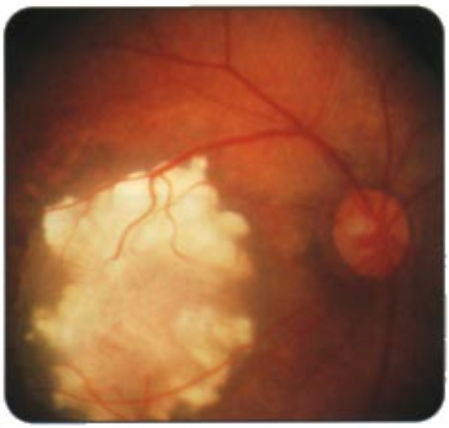


Fig. 7.34

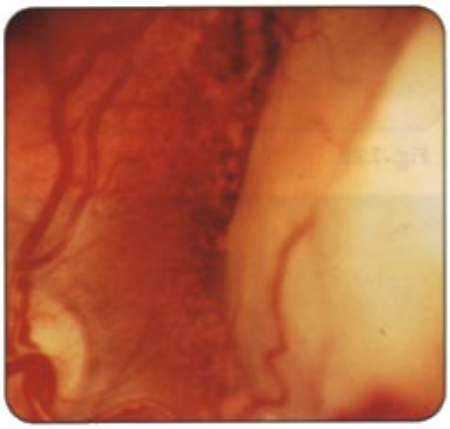


Fig. 7.35

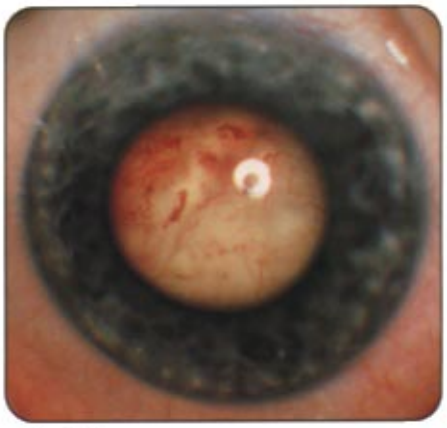


Fig. 7.36

Fig. 7.37

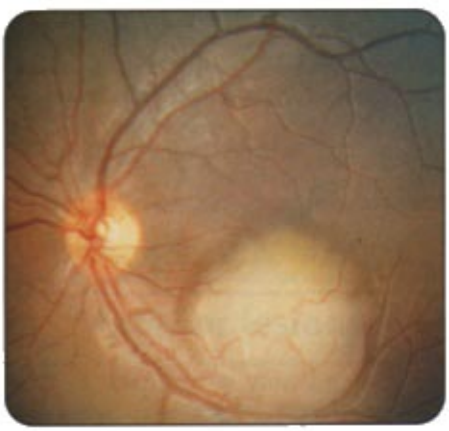


Fig. 7.38

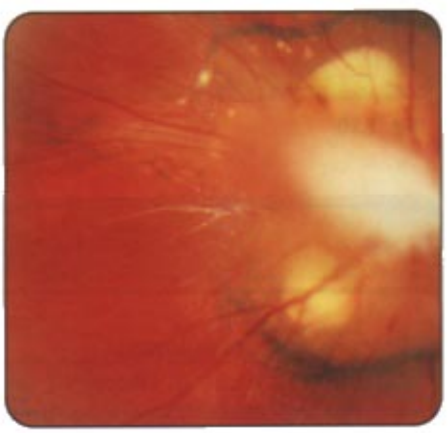


Fig. 7.39

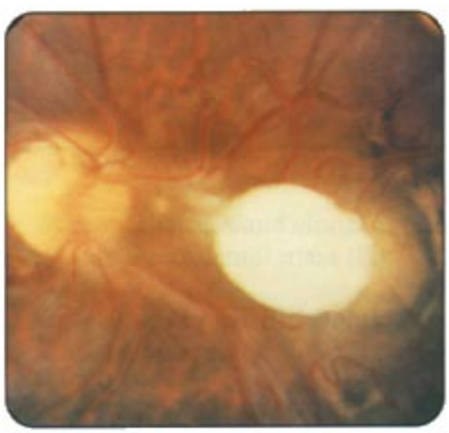


Fig. 7.40

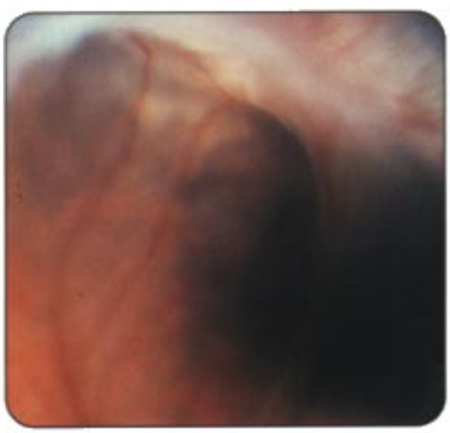


Fig. 7.41

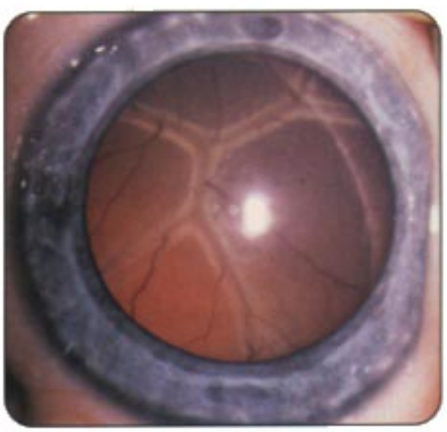


Fig. 7.42

- Coats disease is unilateral and it typically affects boys at about age 8 years (**Fig. 7.43**). It is characterized by massive subretinal exudation (**Figs 7.44, 7.45**), which frequently gives rise to retinal detachment (**Fig. 7.46**).
- Peripheral toxocaral granuloma (**Figs 7.47, 7.48**) may cause tractional retinal detachment.

3. Extensive white fundus lesions

Causes

- Leukocoria (**Fig. 7.49**) may occasionally be caused by extensive myelination of nerve fibers (**Fig. 7.50**) and chorioretinal colobomas (**Fig. 7.51**).

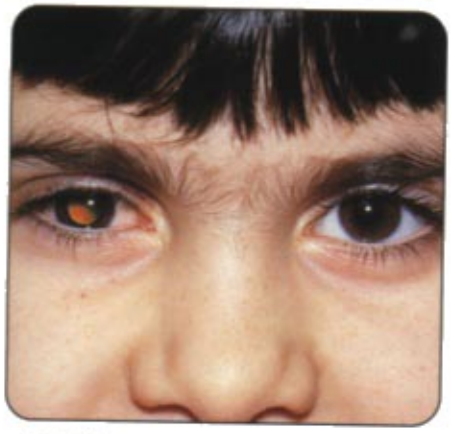


Fig. 7.43

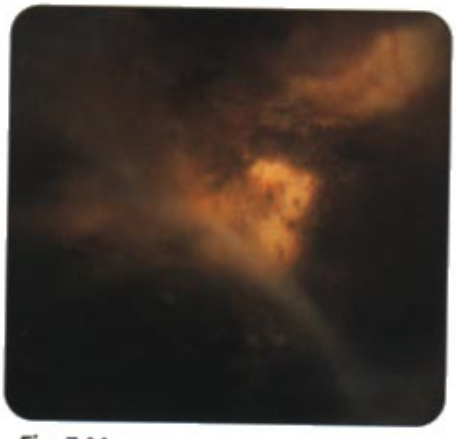


Fig. 7.44

Fig. 7.45

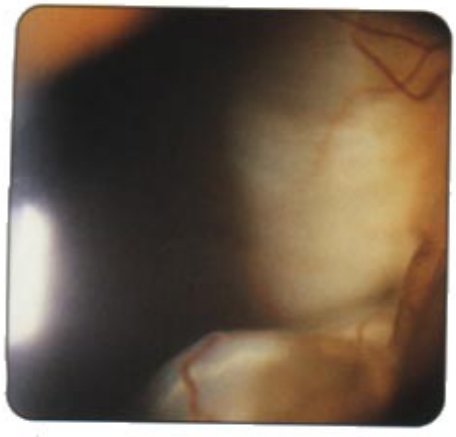


Fig. 7.46

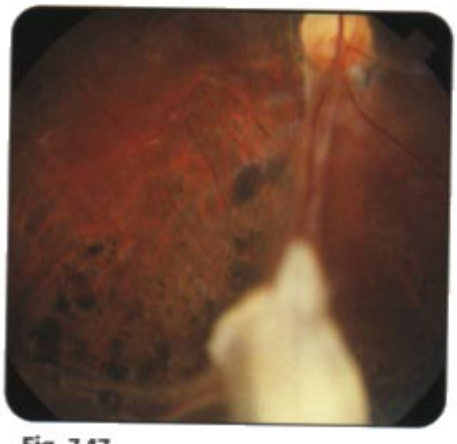


Fig. 7.47

Fig. 7.48

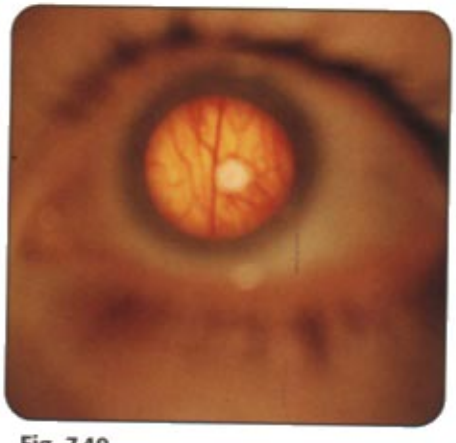


Fig. 7.49

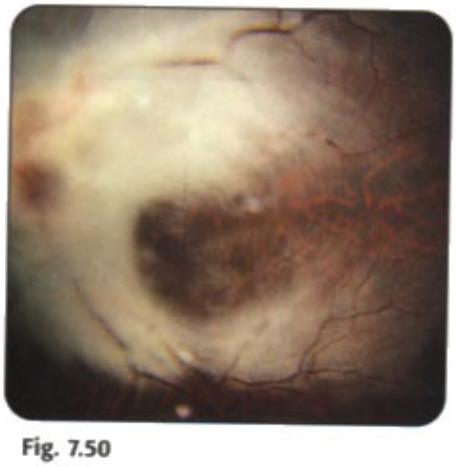


Fig. 7.50

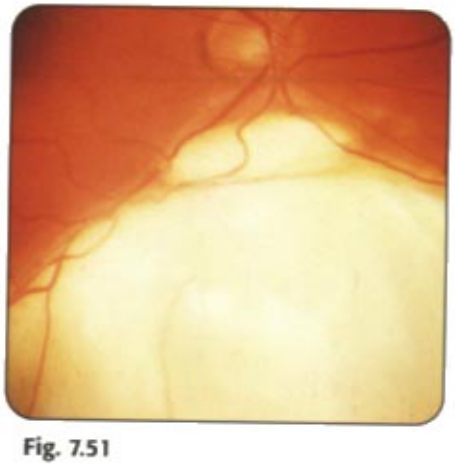


Fig. 7.51

8 The iris

HETEROCHROMIA IRIDIS 188

Hypochromia
Hyperchromia

ECTROPION UVEAE 190

Congenital
Acquired

IRIS ATROPHY 191

Diffuse iris atrophy
Fuchs heterochromic cyclitis
Albinism
Axenfeld–Reiger anomaly
Diabetic iridopathy
Focal iris atrophy
Pigment dispersion syndrome
Pseudophakic pigment dispersion
Pseudoexfoliation syndrome
Postinflammatory
Iridoschisis

FULL-THICKNESS IRIS DEFECTS 196

Congenital
Aniridia
Iris coloboma
Axenfeld–Reiger syndrome
Acquired
Traumatic
Iatrogenic iridectomy or iridotomy
Iridocorneal–endothelial (ICE) syndrome

MULTIPLE SMALL IRIS SPOTS AND NODULES 201

Iris freckles
Brushfield spots
Iris flocculi
Inflammatory nodules
Lisch nodules
Mammillations
Iris nevus (Cogan–Reese) syndrome
Tapioca melanoma
Retinoblastoma invading iris

SOLITARY SOLID IRIS LESIONS 204

Benign
Iris nevus
Iris leiomyoma
Adenoma of iris pigment epithelium
Malignant
Iris melanoma
Forward extension of ciliary body melanoma
Metastatic carcinoma

CYSTIC IRIS LESIONS 208

Primary iris cysts
Primary stromal cysts
Primary epithelial cysts
Secondary iris cysts
Postsurgical cysts
Miotic cysts
Parasitic cysts

VASCULAR IRIS LESIONS 211

Rubeosis iridis
Dilated iris capillaries
Pupillary neovascular (Cobb) tufts
Cavernous hemangioma

HETEROCHROMIA IRIDIS

Heterochromia iridis is a condition in which the two irides have different colors.

Hypochromia

1. Congenital hypochromia

Causes (Table 8.1)

Figure 8.1 shows heterochromia in a patient with Waardenburg syndrome.

2. Horner syndrome

Ipsilateral hypochromia (**Fig. 8.2**) most frequently occurs in congenital cases and occasionally if the lesion is acquired but longstanding.

3. Fuchs heterochromic cyclitis

This is a very common cause of acquired heterochromia.

The affected eye is usually hypochromic (**Figs 8.3, 8.4a and b**) but in 10% of cases it may be hyperchromic (paradoxical heterochromia).

Look for

- Iris atrophy.
- Small, stellate or round keratic precipitates involving the entire endothelium.
- Feathery filaments between the keratic precipitates.
- Absence of posterior synechiae.
- Vitreous cells.
- Complicated cataract – very common and frequently the presenting feature.
- Secondary glaucoma – may occur in longstanding cases.

Table 8.1 Causes of congenital hypochromia

Isolated
Associated with systemic disorders
• Waardenburg syndrome
• Parry–Romberg syndrome
• Hirschprung disease

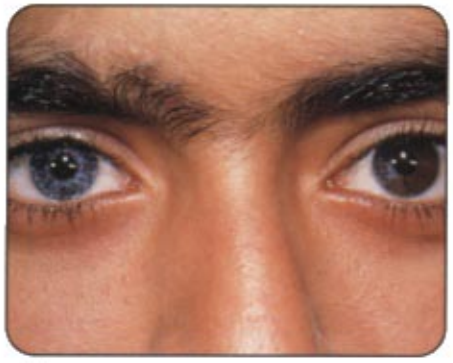


Fig. 8.1

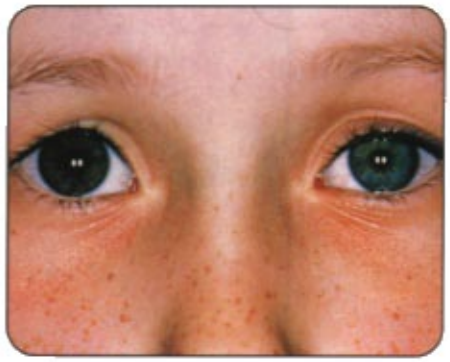


Fig. 8.2

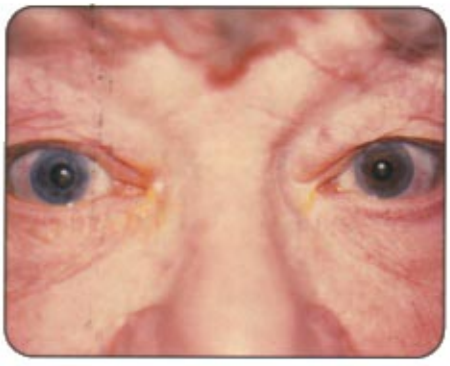


Fig. 8.3

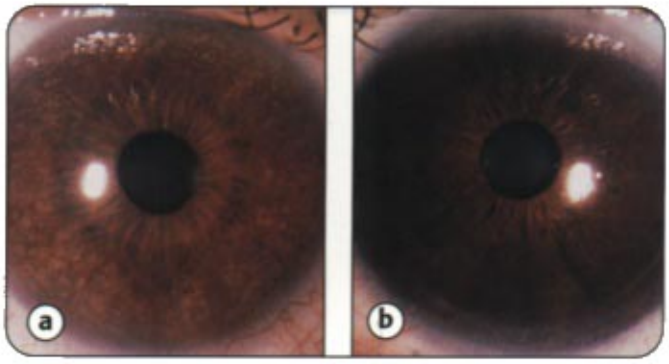


Fig. 8.4

Hyperchromia

1. Oculodermal melancytosis (nevus of Ota)
This is a rare, congenital condition in which iris hyperchromia is associated with ipsilateral skin hyperpigmentation (**Fig. 8.5**).

2. Unilateral use of latanoprost
This drug, which is used for the treatment of glaucoma, may increase iris pigmentation (**Fig. 8.6**), particularly in patients with green–brown or blue–brown irides.

3. Ocular siderosis
Iris hyperpigmentation (**Fig. 8.7**) is a common finding in this condition.

4. Pigmented iris tumors
For example:
- Diffuse iris nevus (**Fig. 8.8**).
- Diffuse iris melanoma (**Fig. 8.9**).

5. Sturge–Weber syndrome
This is a rare phacomatosis which is characterized by nevus flammeus of the face (*see* Chapter 1) and developmental glaucoma. Iris hyperpigmentation is an occasional finding.

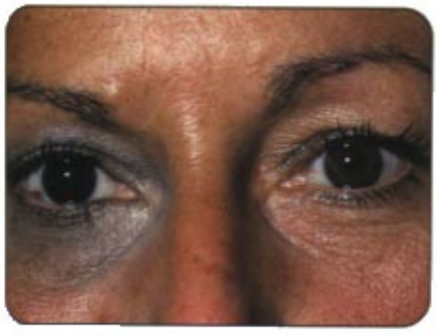


Fig. 8.5

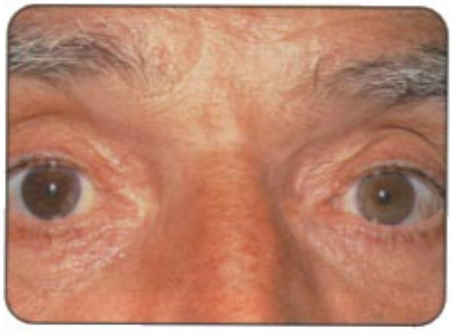


Fig. 8.6

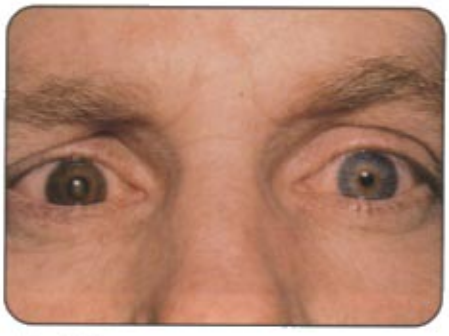


Fig. 8.7

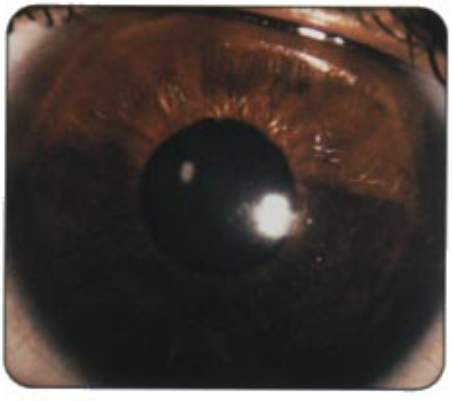


Fig. 8.8

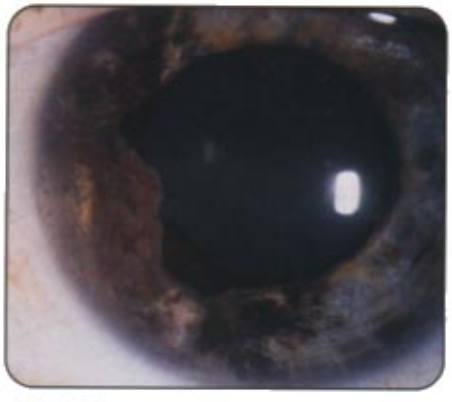


Fig. 8.9

ECTROPION UVEAE

Congenital

1. Isolated ectropion uveae
This is a unilateral, nonprogressive condition with a round and reactive pupil (**Fig. 8.10**). It may be associated with glaucoma due to an angle anomaly. Children with this condition should therefore be followed carefully.

2. Associated with systemic diseases
- Neurofibromatosis-I (NF-I), in which ectropion uvea may coexist with Lisch nodules (**Fig. 8.11**).
- Prader–Willi syndrome.

3. Associated with Axenfeld–Rieger anomaly
This condition is shown in **Figure 8.12**.

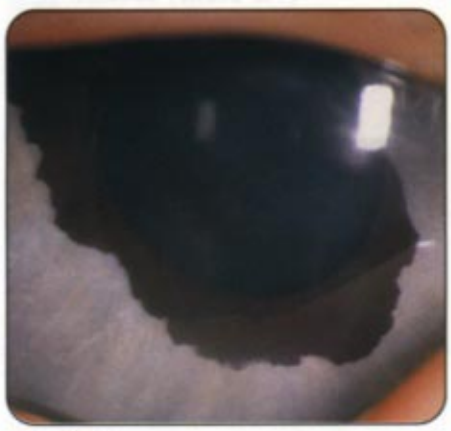


Fig. 8.10

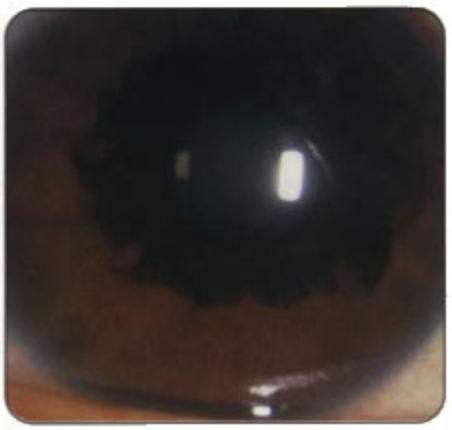


Fig. 8.11

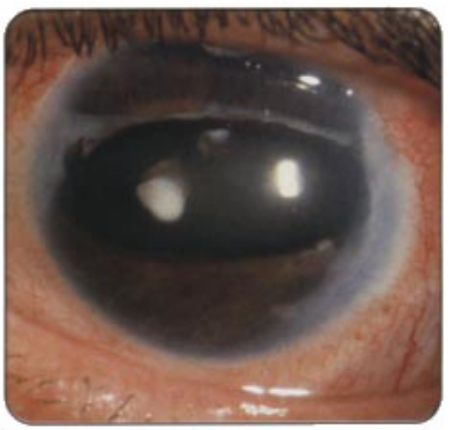


Fig. 8.12

Acquired

1. Associated with rubeosis iridis
Figure 8.13 shows an artificial drainage tube in an eye with neovascular glaucoma.

2. Associated with iris tumors
This is shown in **Figure 8.14**.

Fig. 8.13

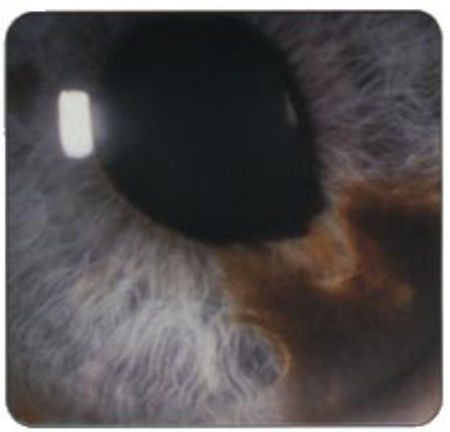


Fig. 8.14

IRIS ATROPHY

Diffuse iris atrophy

Fuchs heterochromic cyclitis

Signs

- Flattening of iris texture resulting in a smooth surface (**Fig. 8.15**).
- Transillumination defects due to atrophy of the pigment epithelium (**Fig. 8.16**).

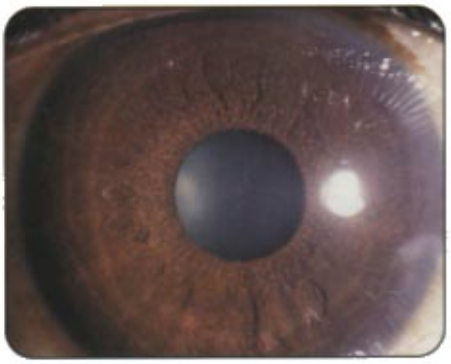


Fig. 8.15

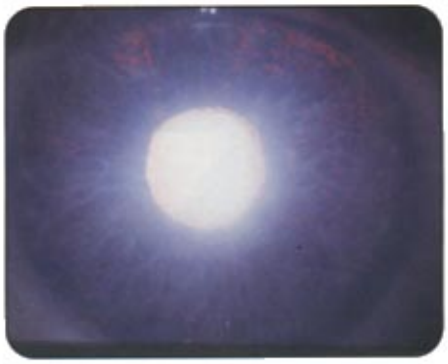


Fig. 8.16

Albinism

Signs

- Patients with oculocutaneous albinism have severe transillumination defects (**Fig. 8.17a and b**) and diaphanous blue irides which give rise to a 'pink-eyed' appearance (**Fig. 8.18a and b**). **Figure 8.19** shows an intraocular lens implant in an albino.
- Patients with ocular albinism may show variable iris transillumination (**Fig. 8.20**).
- Carriers of X-linked ocular albinism may show mild iris transillumination.

Look for

- Photophobia.
- Nystagmus.
- Fundus hypopigmentation.
- Foveal hypoplasia.

Systemic associations

- Chédiak–Higashi syndrome – recurrent infections.
- Hermansky–Pudlak syndrome – easy bruising.

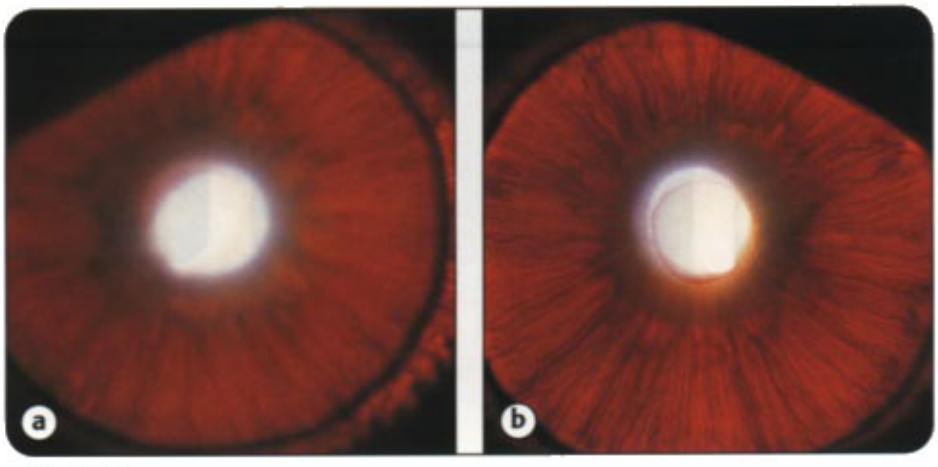


Fig. 8.17

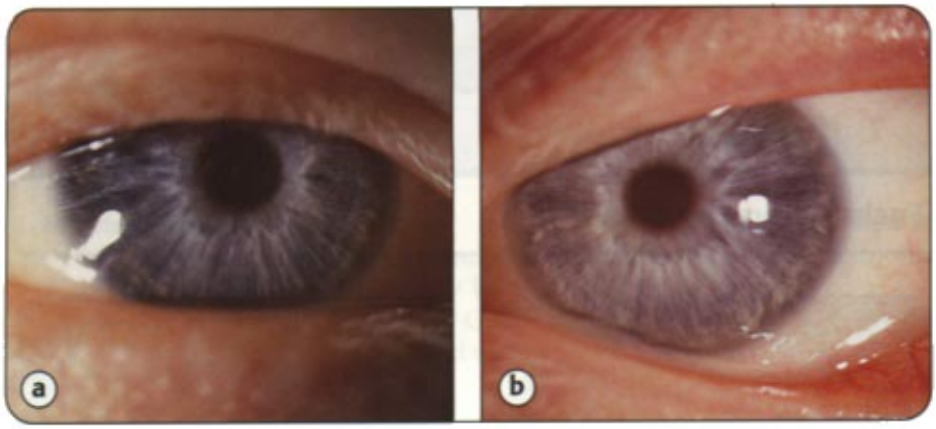


Fig. 8.18

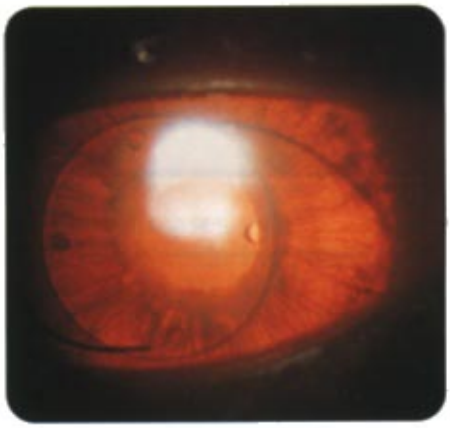


Fig. 8.19

Fig. 8.20

Axenfeld–Rieger anomaly

Axenfeld–Rieger is a rare, bilateral, congenital anomaly in which there is hypoplasia of the anterior leaf of the iris stroma (**Figs. 8.21, 8.22**), as well as other features (*see* **Figs 8.45–8.49**).

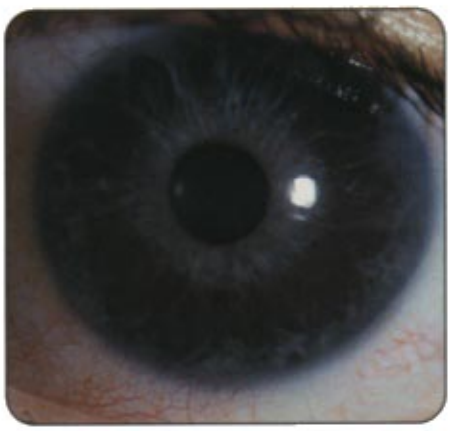


Fig. 8.21

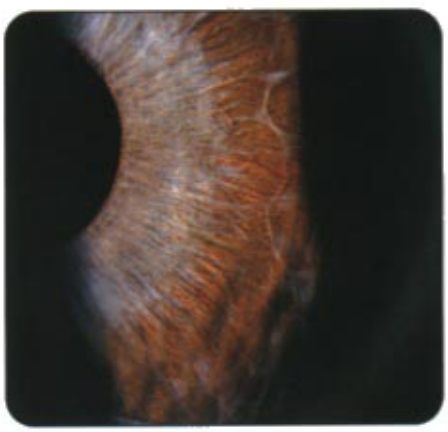


Fig. 8.22

Diabetic iridopathy

Signs

- Mild transillumination defects (**Fig. 8.23**) may be seen due to shedding of the pigment epithelium.

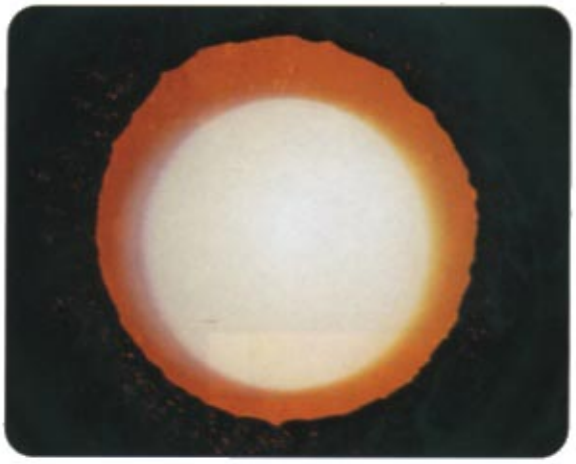


Fig. 8.23

Focal iris atrophy

Pigment dispersion syndrome

Pigment dispersion syndrome is a common, bilateral condition which typically affects young myopic males. Up to 50% of patients with this condition subsequently develop glaucoma.

Signs

- Radial midperipheral iris slits (**Fig. 8.24**) which may be difficult to detect in brown irides and in older patients.
- Fine pigment granules on the anterior surface of the iris and lens (**Fig. 8.25**).
- Pigment on the corneal endothelium either in the form of a (Krukenberg) spindle (**Fig. 8.26**) or distributed diffusely.
- Deep anterior chamber with midperipheral iris concavity (**Fig. 8.27**).
- Posterior iris insertion.
- Circumferential trabecular hyperpigmentation which may in certain places extend onto or anterior to Schwalbe line (Sampaolesi line) (**Fig. 8.28**).
- Increased incidence of lattice degeneration of the retina.

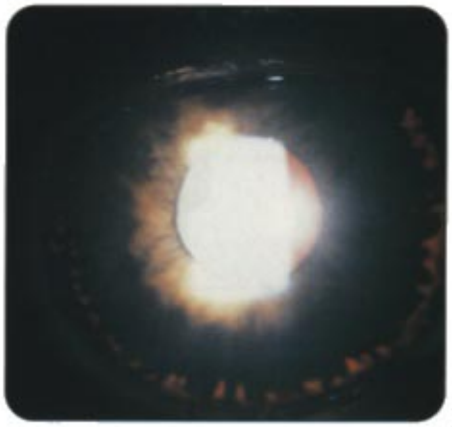


Fig. 8.24

Table 8.2 Causes of trabecular hyperpigmentation

- pigment dispersion syndrome
- pseudophakic pigment dispersion
- pseudoexfoliation syndrome
- after cataract surgery in diabetics
- blunt ocular trauma
- anterior uveitis
- following acute angle-closure glaucoma
- nevus of Ota

Fig. 8.25

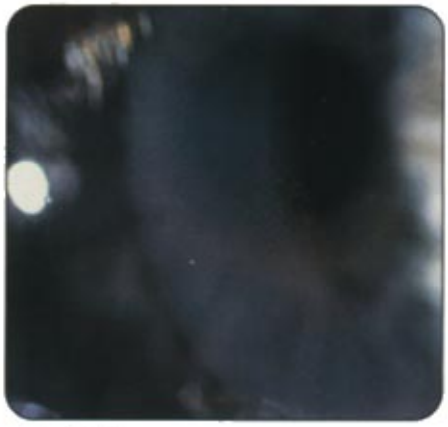


Fig. 8.26

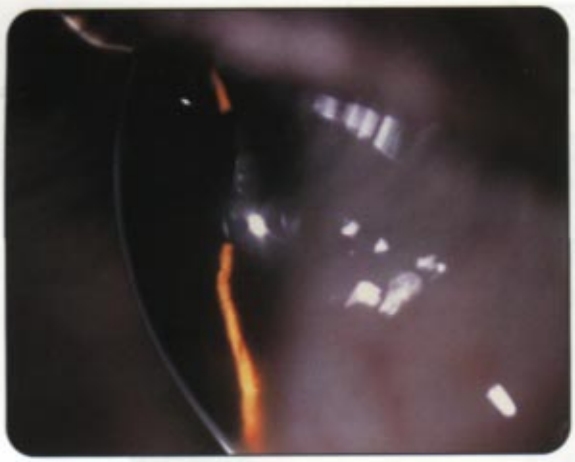


Fig. 8.27

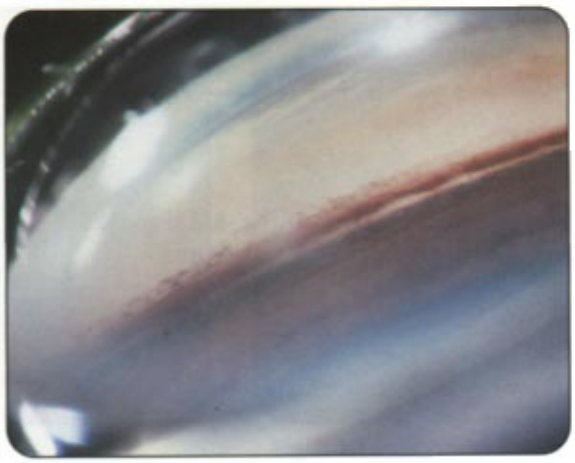


Fig. 8.28

Pseudophakic pigment dispersion

Signs

- Atrophic areas (**Fig. 8.29**) caused by rubbing of a posterior chamber intraocular lens against the back of the iris.

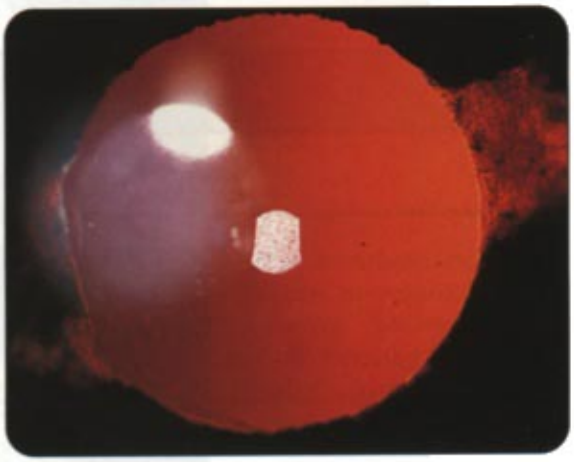


Fig. 8.29

Pseudoexfoliation syndrome

Pseudoexfoliation syndrome is a common condition which at presentation is unilateral in about two-thirds of cases. It typically affects the elderly and is associated with glaucoma.

Signs

- Moth-eaten peripupillary atrophy (**Fig. 8.30**).
- Pseudoexfoliation on the anterior lens surface (**Fig. 8.31**).
- Trabecular hyperpigmentation which is most marked inferiorly, and Sampaolesi line.
- Dandruff-like deposits of pseudoexfoliative material in the trabeculum.

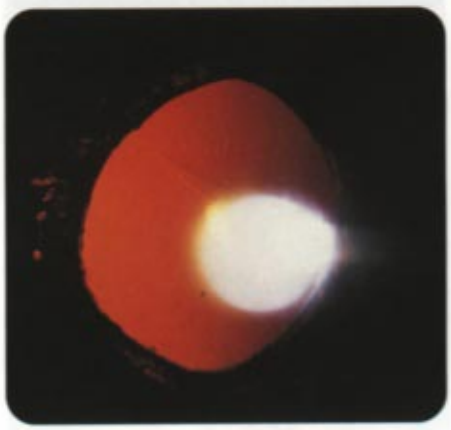


Fig. 8.30

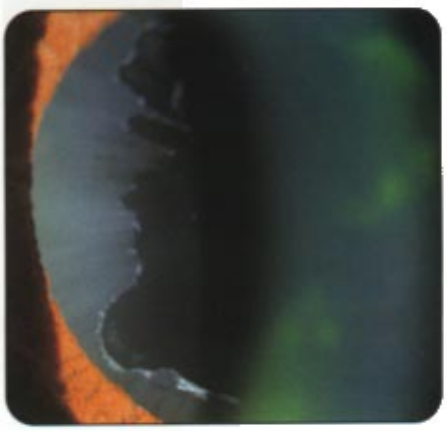


Fig. 8.31

Postinflammatory

Causes

- Herpes zoster (**Fig. 8.32**).
- Herpes simplex.
- Leprosy (**Fig. 8.33**).
- Syphilis.

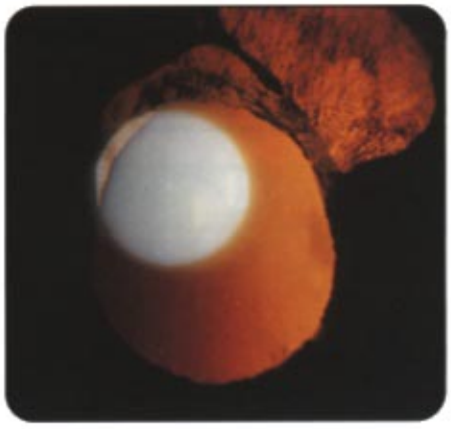


Fig. 8.32

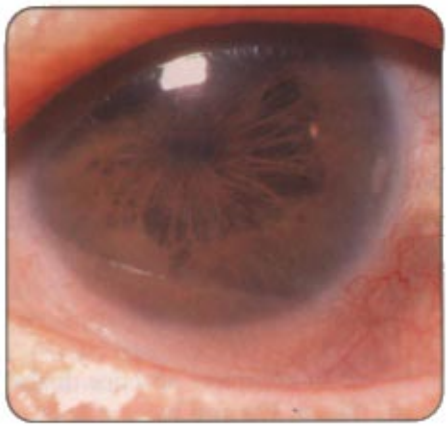


Fig. 8.33

Iridoschisis

Iridoschisis is an uncommon, usually bilateral but asymmetric condition which typically affects elderly individuals.

Signs

- Intrastromal iris atrophy which is frequently inferior (**Fig. 8.34**).
- Splitting of the iris stroma (**Fig. 8.35**).
- Disintegration of stroma into fibrils (**Fig. 8.36**).
- Iris fibrils may float in the anterior chamber (**Fig. 8.37**).
- Shallow anterior chamber (**Fig. 8.38**) and narrow angle.

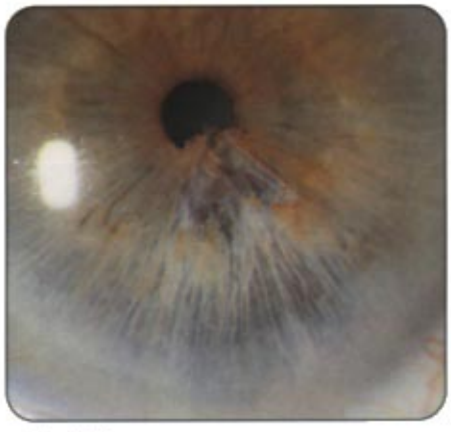


Fig. 8.34

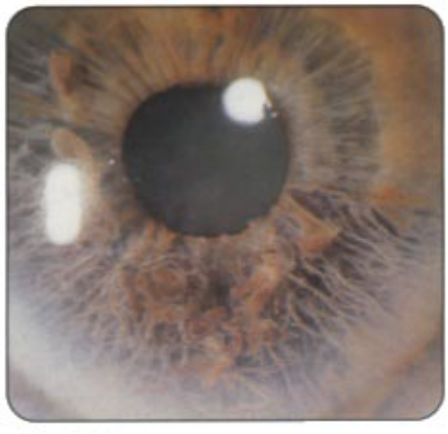


Fig. 8.35

Fig. 8.36

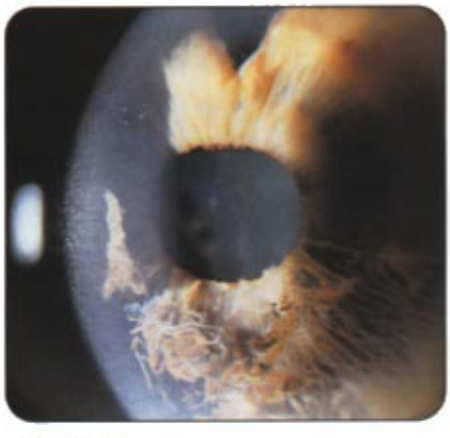


Fig. 8.37

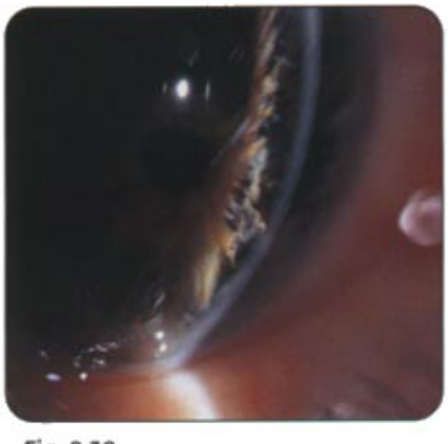


Fig. 8.38

FULL-THICKNESS IRIS DEFECTS

Congenital

Aniridia

Aniridia is a rare, bilateral condition which is associated with glaucoma in 75% of cases.

Signs

- Partial or complete absence of the iris (**Fig. 8.39a and b**).
- Synechial angle-closure glaucoma occurs in 75% of cases as a result of pulling forward of rudimentary iris tissue (**Fig. 8.40**).
- Systemic disorders (**Table 8.3**).
- Many associated ocular malformations (**Table 8.4**).

Figure 8.41 shows the edge of the lens in a phakic eye with aniridia.
Figure 8.42 shows a pseudophakic eye with aniridia.

Table 8.3 Classification of aniridia
AN-1 (85%)
• isolated (autosomal dominant)
AN-2 – Miller syndrome (13%)
• deletion of the short arm of chromosome 11
• Wilm tumor
• genitourinary anomalies
• mental retardation
AN-3 – Gillespie syndrome (2%)
• autosomal recessive
• mental handicap
• cerebellar ataxia

Table 8.4 Ocular associations of aniridia
Glaucoma
Corneal lesions
• pannus
• opacity
• dermoids
• sclerocornea
• keratolenticular adhesions
Lens changes
• absence
• anterior polar cataract
• subluxation
• persistent pupillary membranes
Fundus lesions
• foveal hypoplasia
• disc hypoplasia
• colobomas
Nystagmus

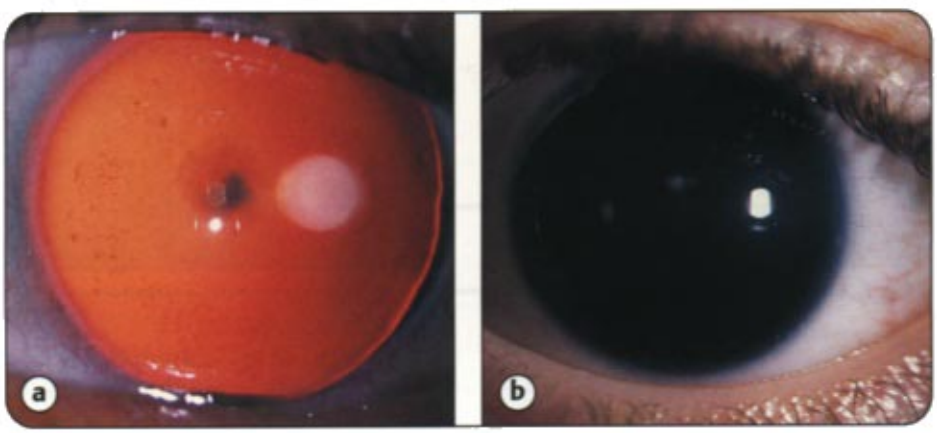


Fig. 8.39

Fig. 8.40

Fig. 8.41

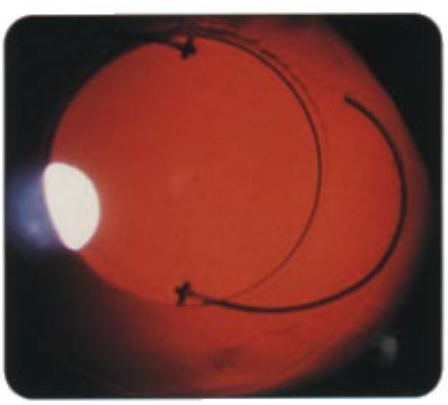


Fig. 8.42

Iris coloboma

Iris coloboma is an uncommon, unilateral or bilateral condition caused by defective closure of the embryonic fissure. Isolated iris colobomas are either sporadic or dominantly inherited.

Signs

- A total coloboma involves the whole iris from pupil to root (**Fig. 8.43**).
- A partial coloboma does not involve the iris root (**Fig. 8.44**).

Ocular associations

- Colobomas of the ciliary body, lens, choroid, and optic nerve.
- Microphthalmos.

Systemic associations

- Patau syndrome (trisomy 13).
- Edward syndrome (trisomy 18).
- Cat-eye syndrome (partial trisomy 22).
- Pagon (CHARGE) syndrome.
- Meckel–Gruber syndrome.
- Lenz microphthalmos syndrome.
- Goltz focal dermal hypoplasia.
- Rubinstein–Taybi syndrome.

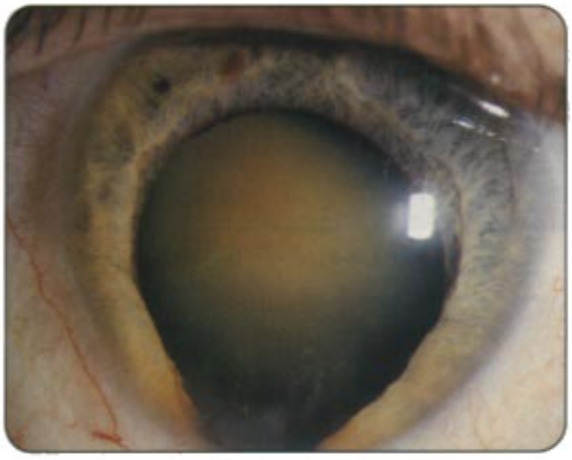


Fig. 8.43

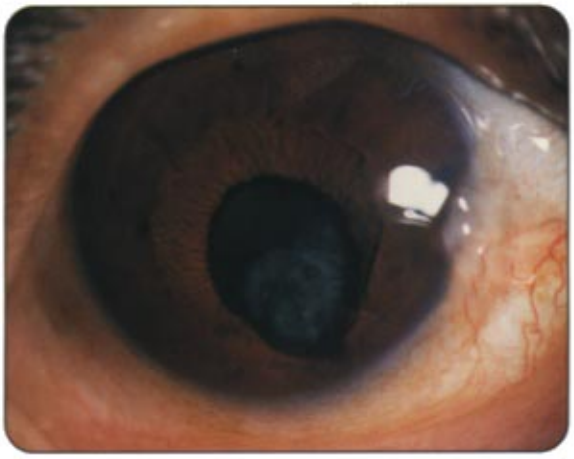


Fig. 8.44

Axenfeld–Rieger syndrome

Axenfeld–Rieger syndrome is a rare, bilateral, congenital disorder which is associated with glaucoma in 50% of cases.

Signs

- Variable diffuse iris stromal atrophy (*see* **Figs 8.21, 8.22**).
- Full-thickness iris defects (**Figs 8.45a and b, 8.46**).
- Corectopia (*see* **Figs 8.45a and b, 8.46**).
- Ectropion uveae (*see* **Fig. 8.12**).
- Posterior embryotoxon (**Fig. 8.47**).
- Angle anomalies (**Fig. 8.48**) and synechial angle closure (**Fig. 8.49**), which are associated with glaucoma in about 50% of cases.
- Iris mammillations in some cases (*see* **Figs 8.68, 8.69, 8.70**).

Systemic associations (Rieger syndrome)

- Dental anomalies (**Fig. 8.50**).
- Facial anomalies.
- Rudimentary paraumbilical skin.
- Defects in the region of the pituitary gland.

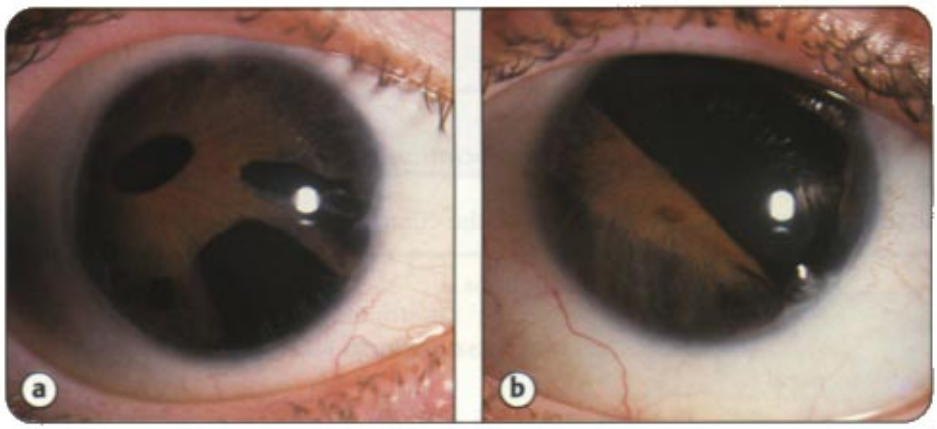


Fig. 8.45

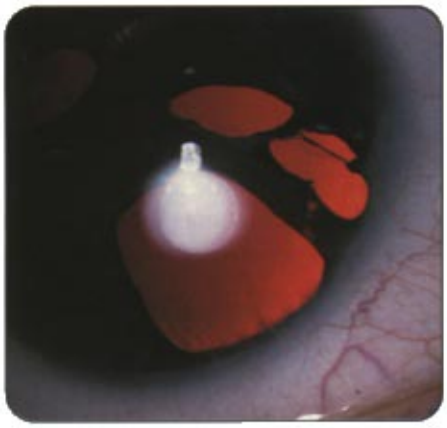


Fig. 8.46

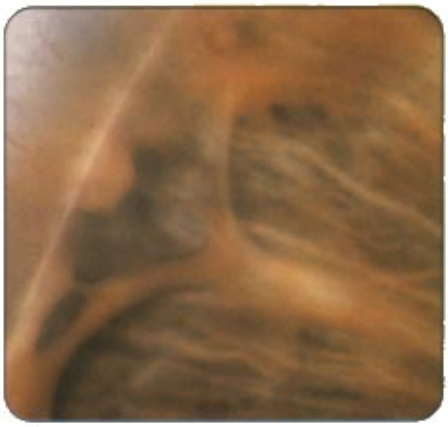


Fig. 8.47

Fig. 8.48

Fig. 8.49

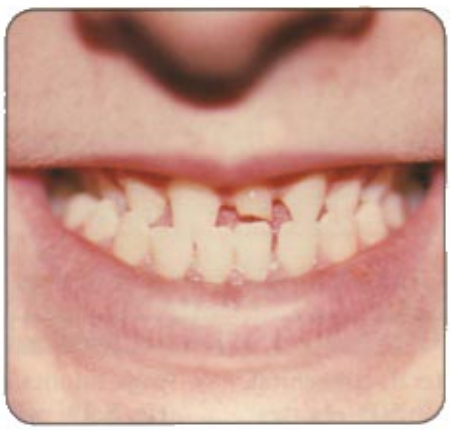


Fig. 8.50

Acquired

Traumatic

- Penetrating trauma.
- Iridodialysis from blunt trauma (**Fig. 8.51**).

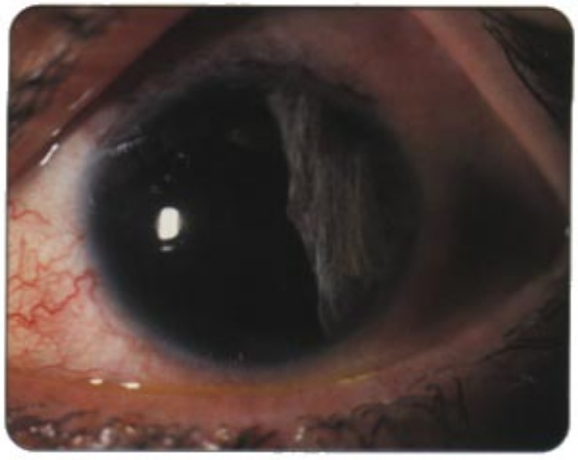


Fig. 8.51

Iatrogenic iridectomy or iridotomy

Figures 8.52 and **8.53** show the appearance following a laser iridotomy.

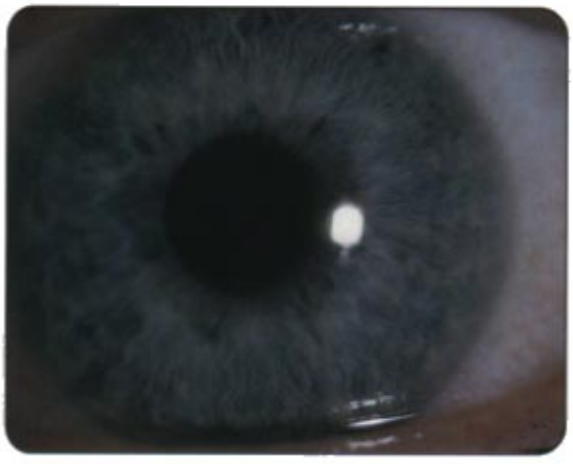


Fig. 8.52

Fig. 8.53

Iridocorneal–endothelial (ICE) syndrome

ICE syndrome is a rare, unilateral condition which typically affects females and is associated with intractable glaucoma.

There are three types:
- Progressive iris atrophy.
- Cogan–Reese syndrome.
- Chandler syndrome.

Signs

- Stromal iris atrophy (**Figs 8.54, 8.55**).
- Pseudopolycoria (**Fig. 8.56**).
- Corectopia (**Fig. 8.57**).
- Broad peripheral anterior synechiae extending beyond Schwalbe line (**Fig. 8.58**).
- Iris nodules (*see* **Fig. 8.71**) or diffuse iris nevus (*see* **Fig. 8.8**) occur in Cogan–Reese syndrome.
- Corneal endothelial 'beaten-silver' appearance (**Fig. 8.59**), which is most severe in Chandler syndrome.

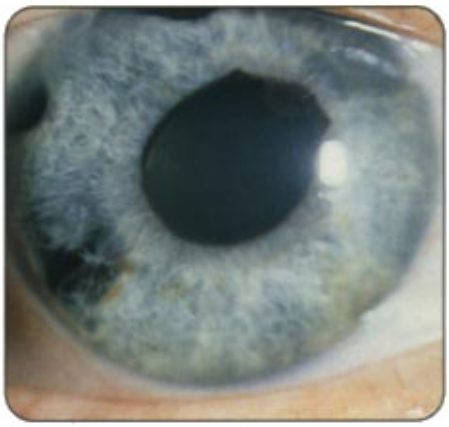


Fig. 8.54

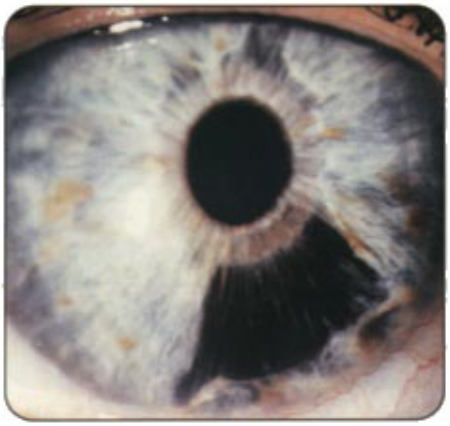


Fig. 8.55

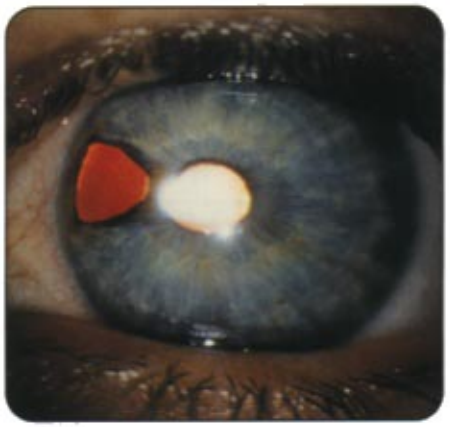


Fig. 8.56

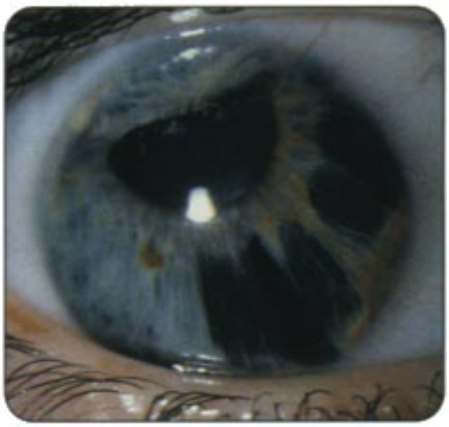


Fig. 8.57

Fig. 8.58

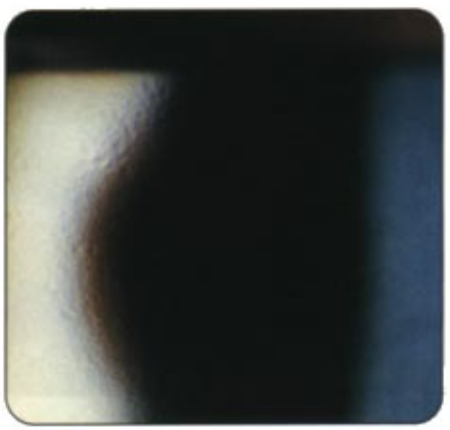


Fig. 8.59

MULTIPLE SMALL IRIS SPOTS AND NODULES

Iris freckles

Iris freckles are bilateral, tiny nevi which are present to some extent in about 50% of adults but are rarely observed before the age of 12 years.

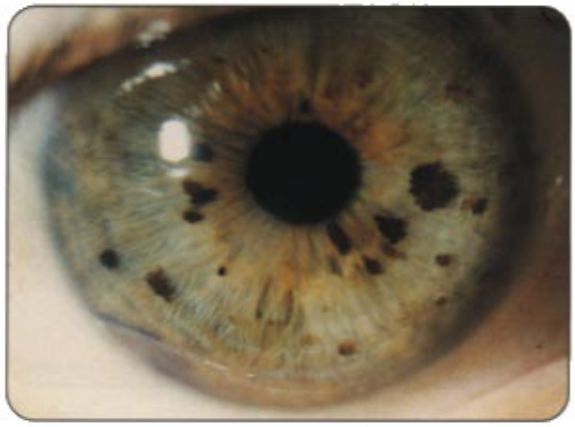


Fig. 8.60

Signs

- Multiple, discrete spots which range in color from yellow-tan to deep chocolate-brown (**Fig. 8.60**).

Brushfield spots

Brushfield spots are bilateral, and are found in 85% of patients with Down syndrome and about 25% of the general population.

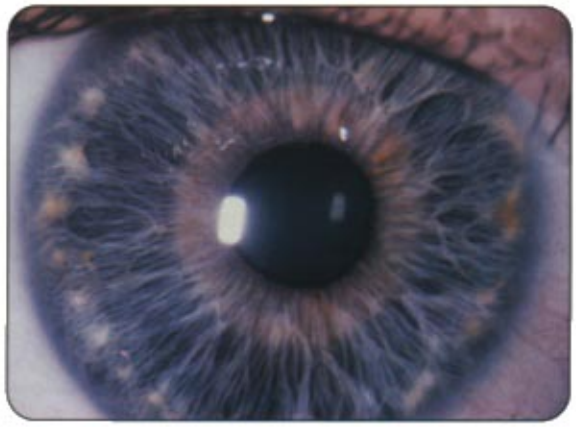


Fig. 8.61

Signs

- Tiny white or yellowish spots arranged in a ring at the junction between the outer and middle third of the iris stroma (**Fig. 8.61**).

Iris flocculi

Iris flocculi are uncommon and bilateral.

Fig. 8.62

Signs

- Pupillary tags or tiny solid nodules (**Fig. 8.62**) of pigment epithelium.

Inflammatory nodules

- Koeppe nodules are common and are located on the border of the pupil. They are a feature of granulomatous inflammation although they may also be seen in Fuchs heterochromic cyclitis.
- Busacca nodules are uncommon and are located on the surface of the iris away from the pupil. They are also a feature of granulomatous inflammation.

Figure 8.63 shows Koeppe and Busacca nodules in an eye with chronic anterior uveitis associated with sarcoidosis.

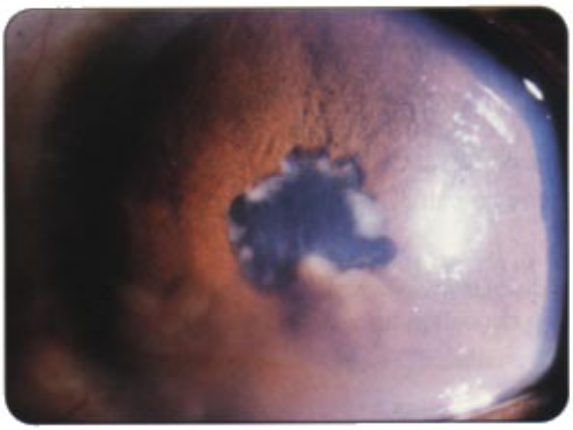


Fig. 8.63

Lisch nodules

Lisch nodules are bilateral melanocytic hamartomas which are found in virtually all patients with neurofibromatosis-I after the age of 16 years.

Signs

- Small, yellow or brown, dome-shaped nodules (**Figs 8.64–8.67**).

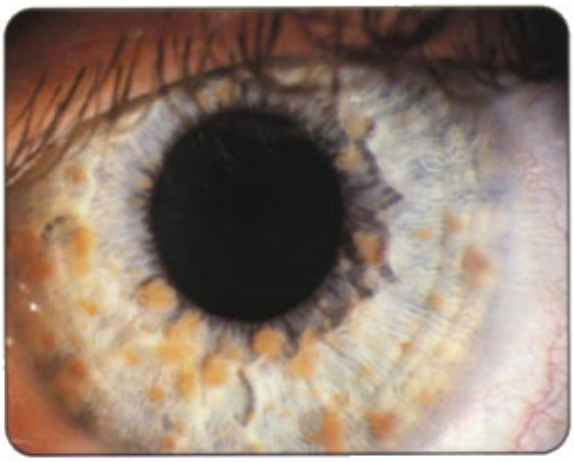


Fig. 8.64

Fig. 8.65

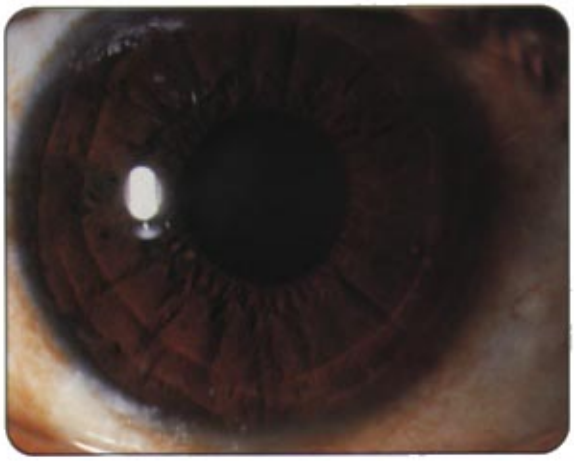


Fig. 8.66

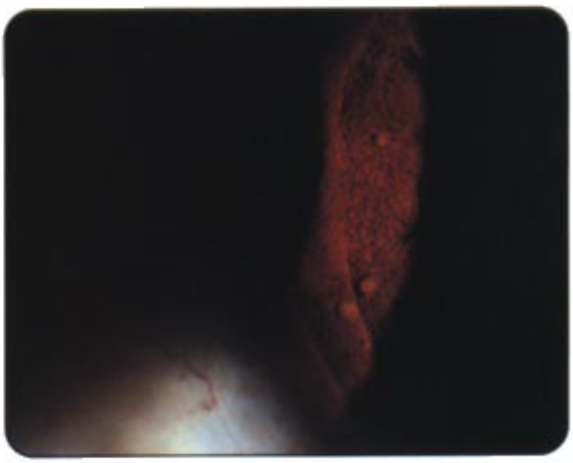


Fig. 8.67

Mammillations

Mammillations are rare and usually unilateral.

Signs

- Regularly spaced, tiny, smooth, villiform lesions (**Figs 8.68–8.70**).

Occasional associations

- Ocular and oculodermal melanocytosis (nevus of Ota).
- Neurofibromatosis-I.
- Axenfeld–Rieger anomaly.
- Peters anomaly.

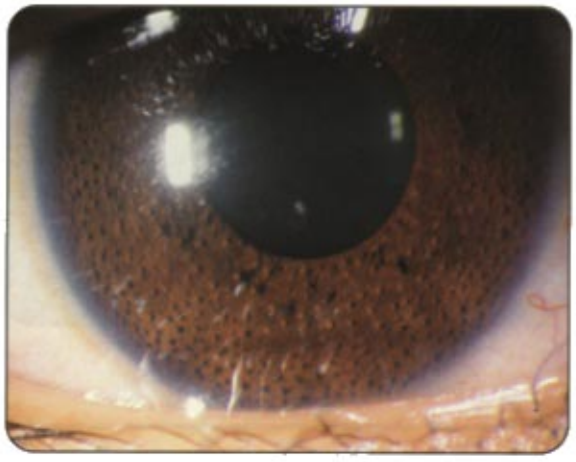


Fig. 8.68

Fig. 8.69

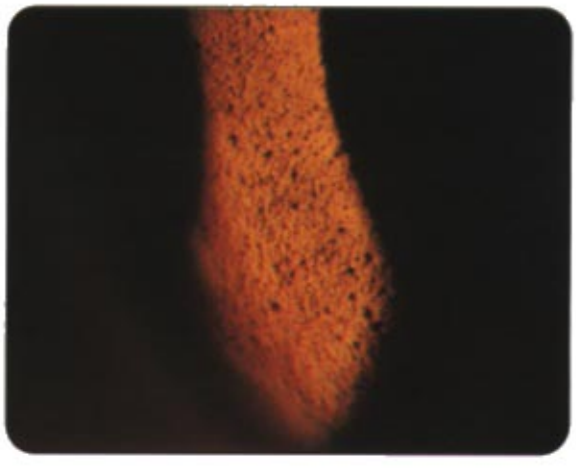


Fig. 8.70

Iris nevus (Cogan–Reese) syndrome

Signs

- Unilateral, small, pedunculated iris nodules which may resemble mammillations (**Fig. 8.71**).

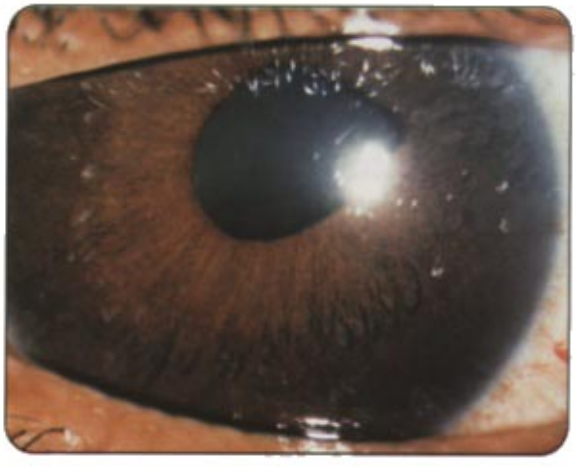


Fig. 8.71

Tapioca melanoma

Signs

- Lightly pigmented or nonpigmented iris nodules (**Fig. 8.72**).

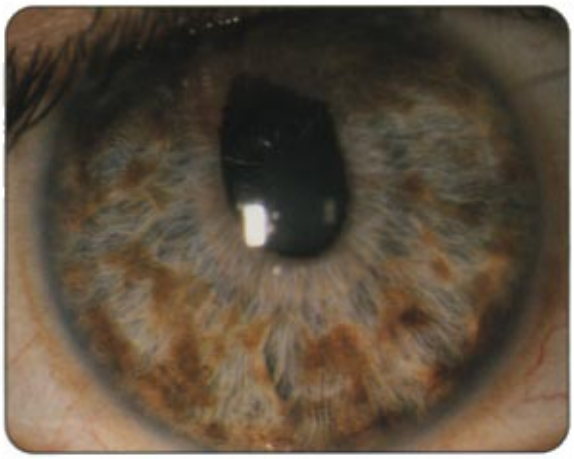


Fig. 8.72

Retinoblastoma invading iris

This very rare presentation of retinoblastoma may give rise to diagnostic difficulties because it occurs at a later age than the more typical type.

Signs

- Pale iris nodules (**Fig. 8.73**) which may be associated with a blood-stained pseudohypopyon (**Fig. 8.74**). Multiple deposits of metastatic carcinoma may give rise to a similar appearance.

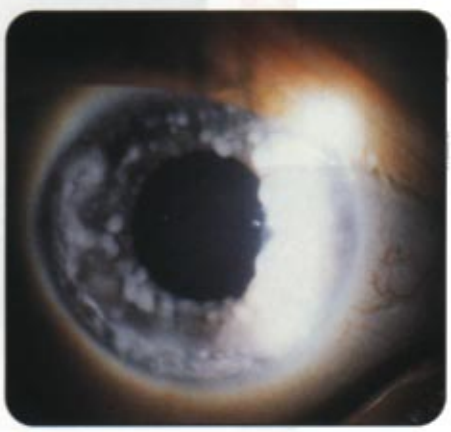


Fig. 8.73

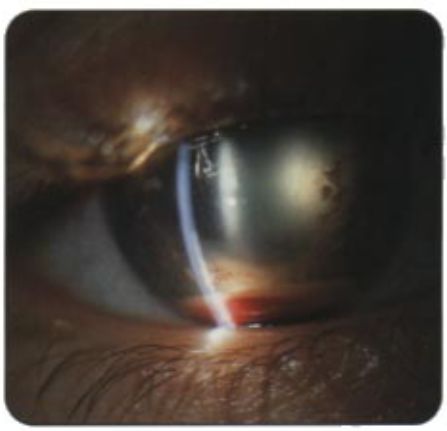


Fig. 8.74

SOLITARY SOLID IRIS LESIONS

Benign

Iris nevus

Iris nevi are common and usually unilateral.

Signs

- Flat or slightly elevated pigmented lesion involving the superficial layers of the iris (**Fig. 8.75**).
- Mild distortion of the pupil and ectropion uveae are occasionally seen (**Fig. 8.76**).
- In the Cogan–Reese syndrome the nevus is more diffuse and obscures the normal pattern of iris crypts and gives rise to hyperchromic heterochromia (*see* **Fig. 8.8**).

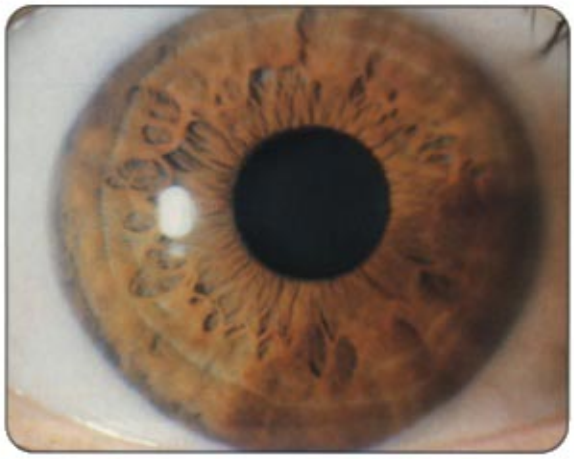


Fig. 8.75

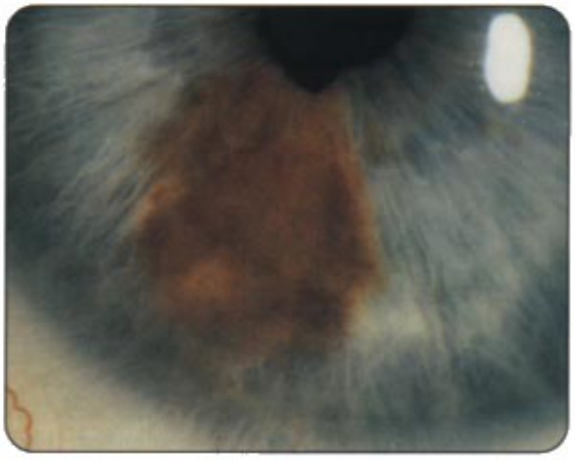


Fig. 8.76

Iris leiomyoma

An iris leiomyoma is a very rare tumor that arises from the sphincter or dilator muscles.

Signs

- The appearance (**Fig. 8.77**) is similar to that of an amelanotic melanoma apart from the fact that an iris leiomyoma may remain stationary in size.

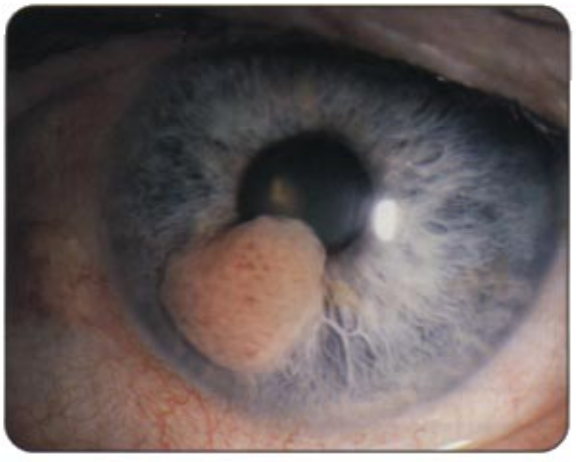


Fig. 8.77

Adenoma of iris pigment epithelium

An adenoma of iris pigment epithelium is a very rare tumor.

Signs

- Black friable nodule in the angle (**Fig. 8.78**) which may be mistaken for a melanoma on initial examination.

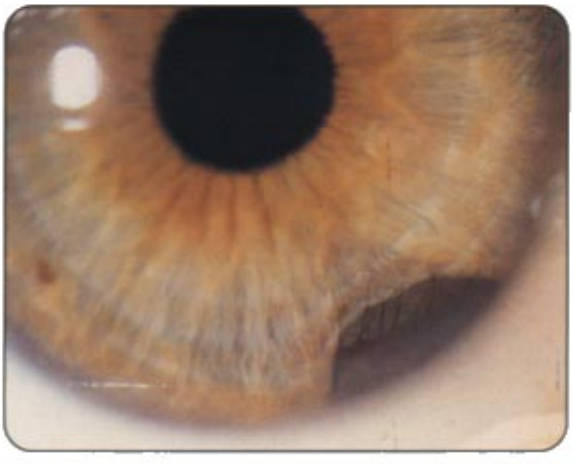


Fig. 8.78

Malignant

Iris melanoma

An iris melanoma is a rare, very slow-growing tumor of relatively low malignancy which typically presents during the fifth decade of life. It occurs with increased frequency in patients with oculodermal melanocytosis (naevus of Ota).

Signs

- Pigmented (**Figs 8.79, 8.80**) or nonpigmented (**Figs 8.81–8.83**) nodule, at least 3 mm in diameter and 1 mm in thickness, which is invariably located in the inferior half of the iris.
- Prominent vascularity which is more easy to detect in a nonpigmented tumor (**Figs 8.84, 8.85**).
- Pupillary distortion, ectropion uveae, and secondary lens opacities (**Fig. 8.86**).
- Angle involvement (**Figs 8.87, 8.88**) which, if extensive, may give rise to raised intraocular pressure.
- Documented growth.

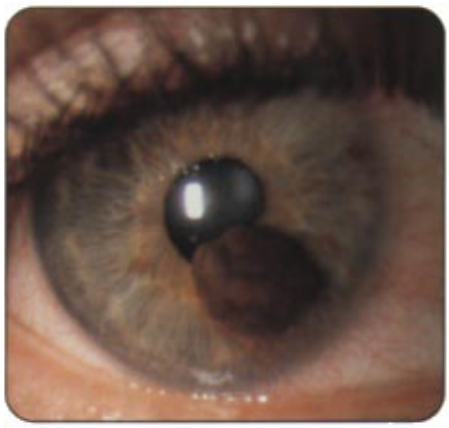


Fig. 8.79

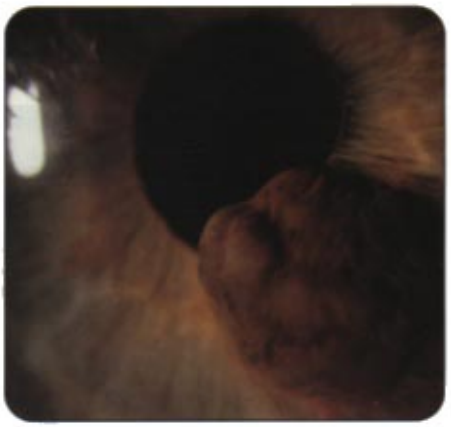


Fig. 8.80

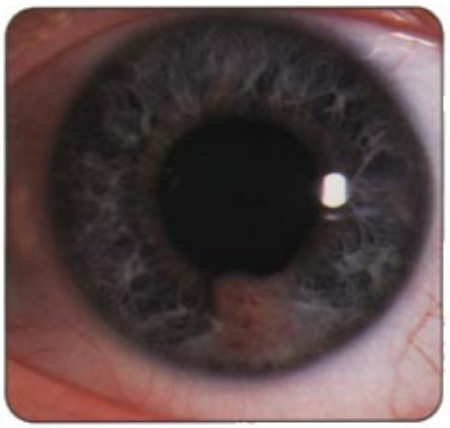


Fig. 8.81

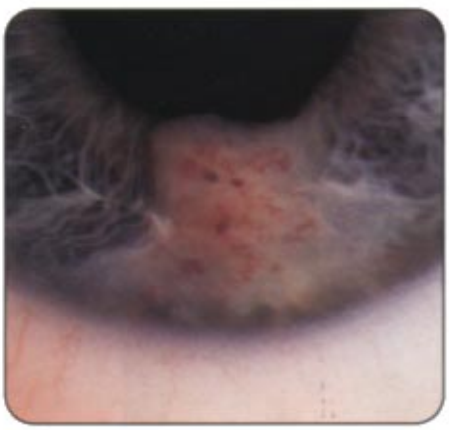


Fig. 8.82

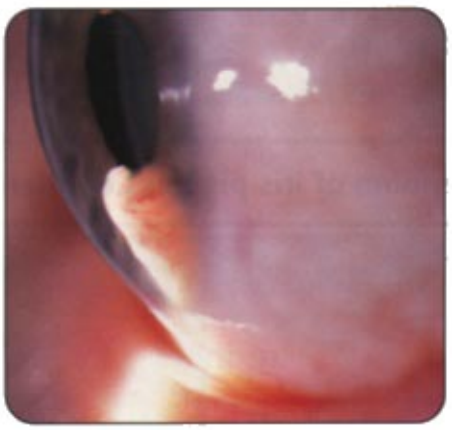


Fig. 8.83

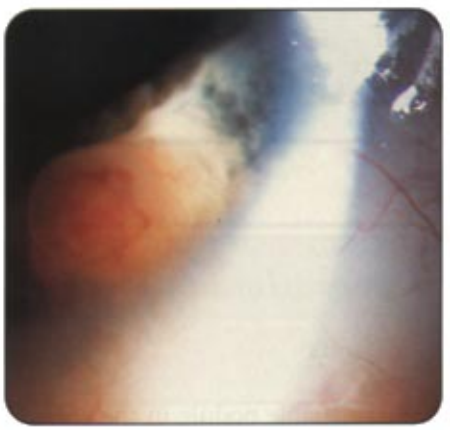


Fig. 8.84

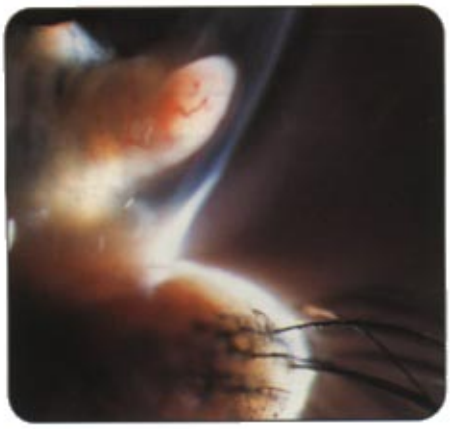


Fig. 8.85

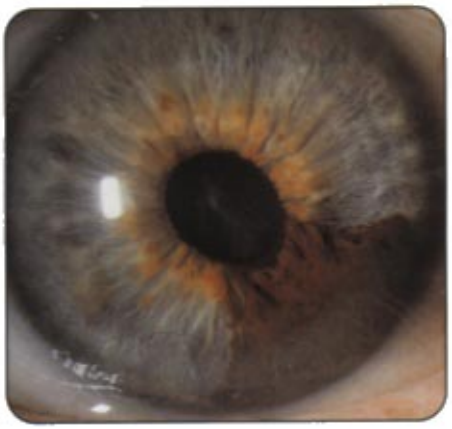


Fig. 8.86

Fig. 8.87

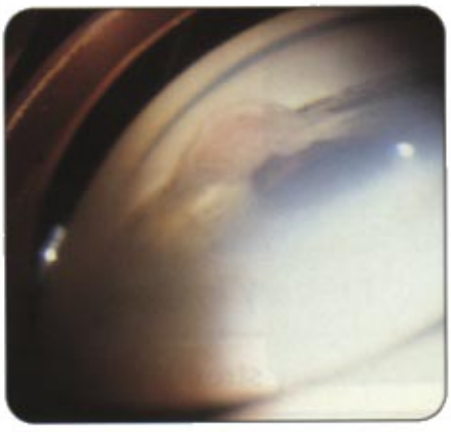


Fig. 8.88

Forward extension of ciliary body melanoma

Signs

- This condition may mimic an iris melanoma but is not confined to the inferior half (**Fig. 8.89**).
- Examination through a dilated pupil will reveal the ciliary body tumor (**Fig. 8.90**).
- Secondary lens opacity is common.

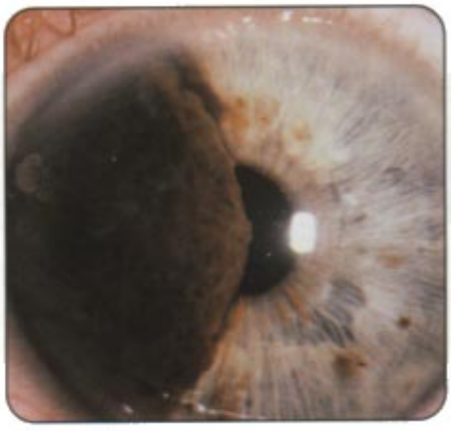


Fig. 8.89

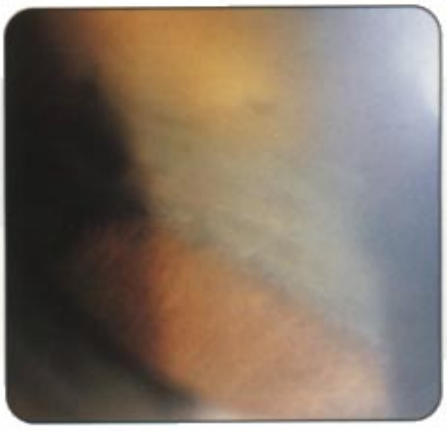


Fig. 8.90

Metastatic carcinoma

As in choroidal metastates, bilateral involvement is common. The most frequent primary sites are the bronchus and breast.

Signs

- Amelanotic lesion (**Fig. 8.91**) which may be associated with pain, inflammation, and secondary glaucoma.
- Multiple deposits in the same eye may also occur.

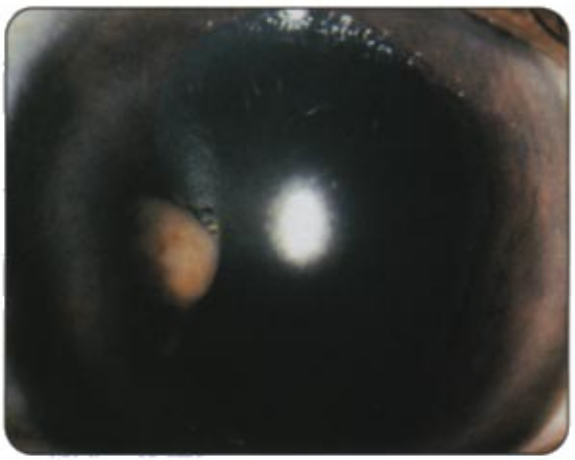


Fig. 8.91

CYSTIC IRIS LESIONS

Primary iris cysts

Primary stromal cysts

Primary iris stromal cysts are very rare and usually occur in young children.

Signs

- Fluid-filled cyst with a clear anterior wall (**Figs 8.92–8.94**).

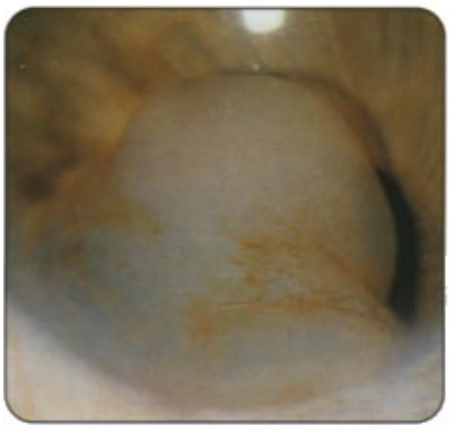


Fig. 8.92

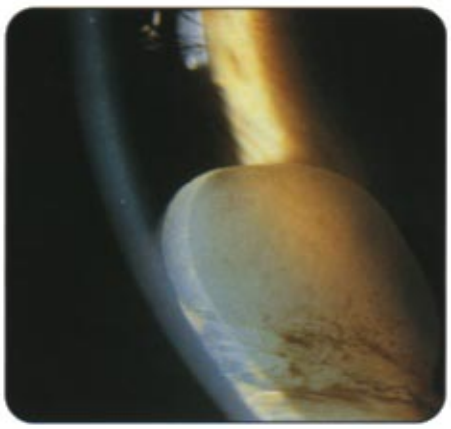


Fig. 8.93

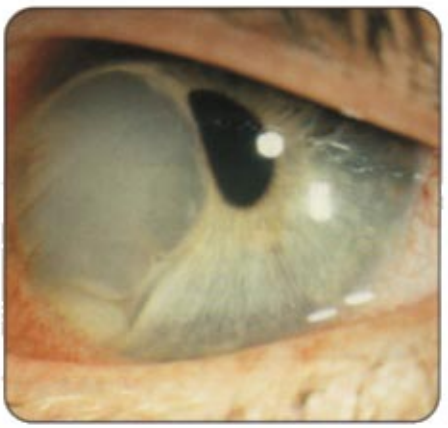


Fig. 8.94

Primary epithelial cysts

Primary iris epithelial cysts are uncommon but clinically important because they may be confused with melanomas.

Signs

- Globular, dark-brown lesion which transilluminates.
- Occasionally, the cyst becomes dislodged into the anterior chamber.

Classification

- Pupillary cysts are rare (**Figs 8.95, 8.96**).
- Midzonal cysts have an elongated fusiform shape (**Fig. 8.97**).
- Peripheral (iridociliary) cysts are the most common (**Fig. 8.98**).

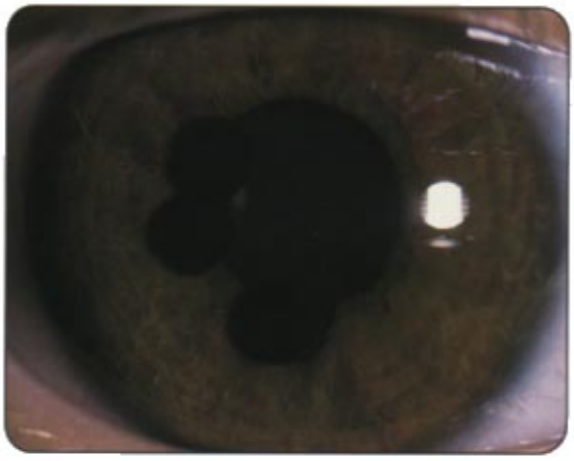


Fig. 8.95

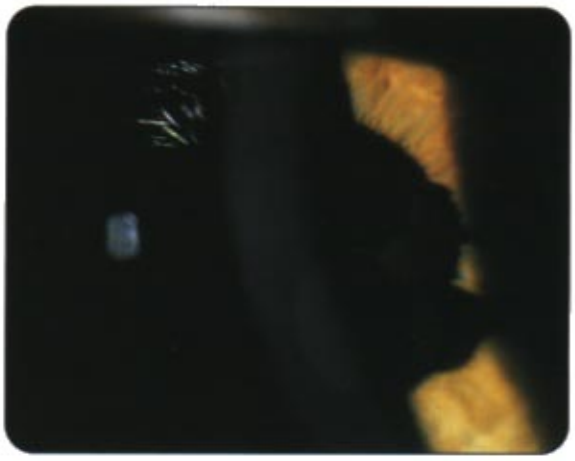


Fig. 8.96

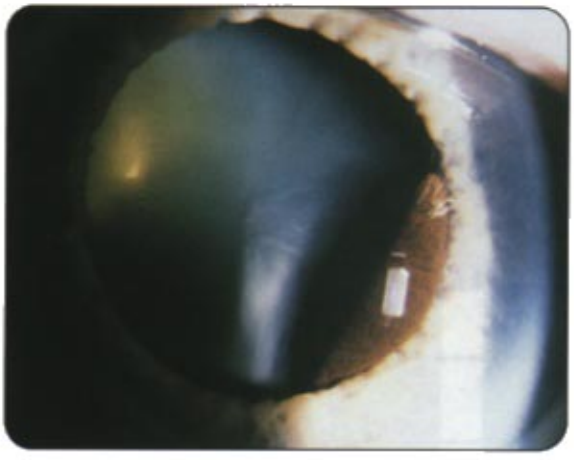


Fig. 8.97

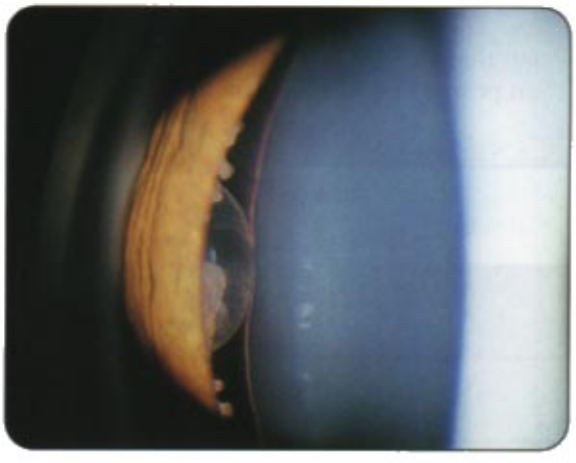


Fig. 8.98

Secondary iris cysts

Postsurgical cysts

Postsurgical iris cysts represent a form of epithelial downgrowth and occur following intraocular surgery.

Signs

- Fluid-filled cyst with a clear anterior wall (**Fig. 8.99**), similar to a primary stromal cyst.

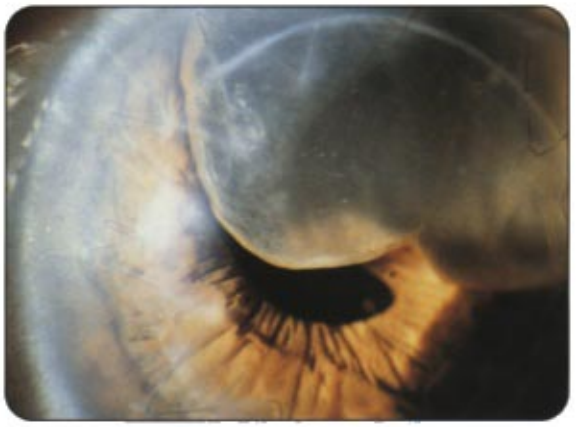


Fig. 8.99

Miotic cysts

Miotic iris cysts are caused by the prolonged use of miotics, particularly ecothiopate iodide.

Signs

- Multiple pupillary cysts (**Fig. 8.100**) similar to but smaller than primary pupillary epithelial cysts.

Fig. 8.100

Parasitic cysts

Parasitic iris cysts are extremely rare and associated with encysted parasites such as those found in cysticercosis (**Fig. 8.101**).

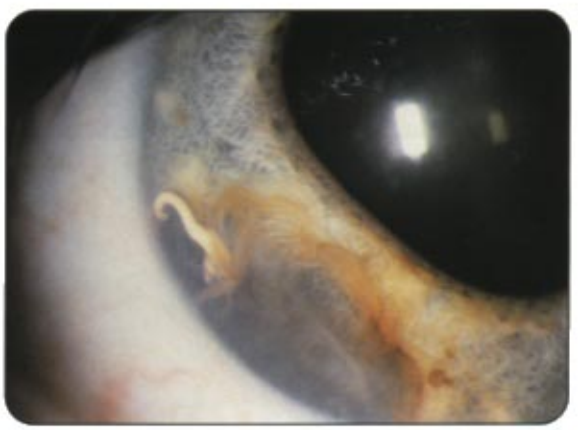


Fig. 8.101

VASCULAR IRIS LESIONS

Rubeosis iridis

Signs (in chronologic order)

- Tiny dilated capillary tufts at the pupil border (**Figs 8.102, 8.103**).
- Radial iris neovascularization (**Figs 8.104, 8.105**).
- Angle neovascularization (**Fig. 8.106**).
- Progressive angle closure by peripheral anterior synechiae and raised intraocular pressure.
- Enlargement and increase in number of iris vessels (**Fig. 8.107**).
- Growth of new vessels across the anterior lens surface may occur (**Fig. 8.108**).
- Ectropion uveae (*see* **Fig. 8.13**).
- Spontaneous hyphema is an uncommon complication (**Fig. 8.109**).

Causes

- Ischemic central retinal vein occlusion.
- Diabetes mellitus.
- Chronic intraocular inflammation.
- Central retinal artery occlusion.
- Ocular ischemic syndrome.
- Intraocular tumors.
- Longstanding retinal detachment.
- Retinopathy of prematurity.

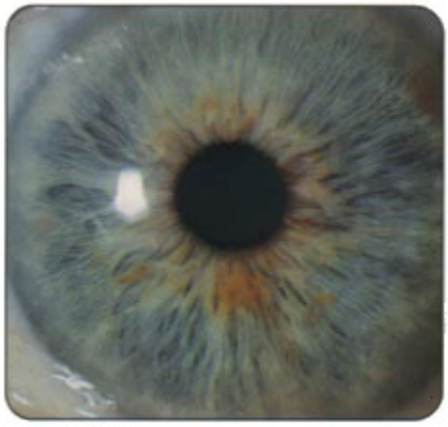


Fig. 8.102

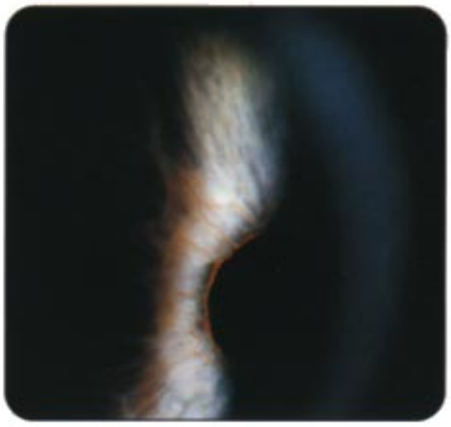


Fig. 8.103

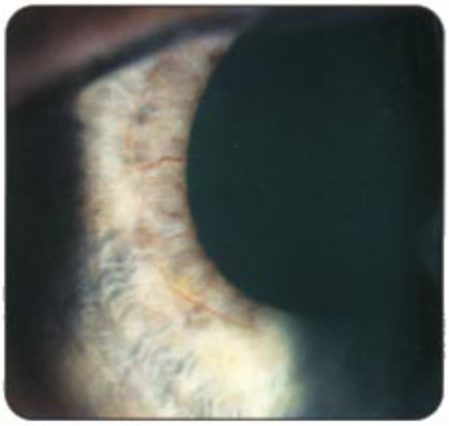


Fig. 8.104

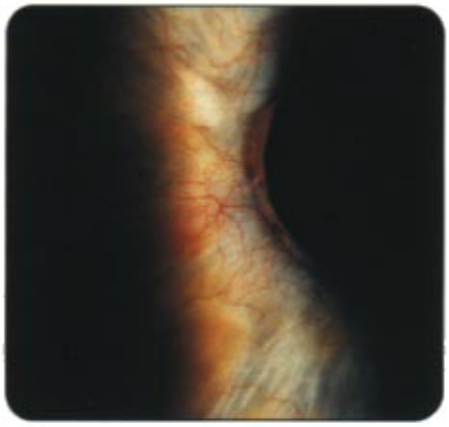


Fig. 8.105

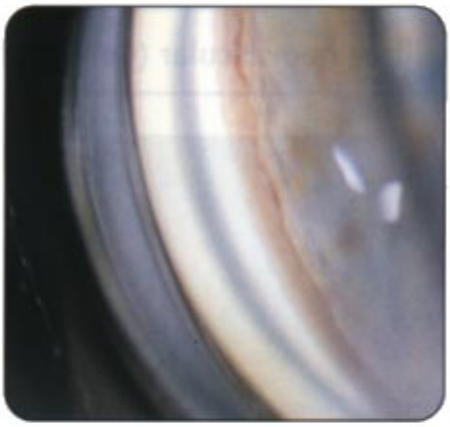


Fig. 8.106

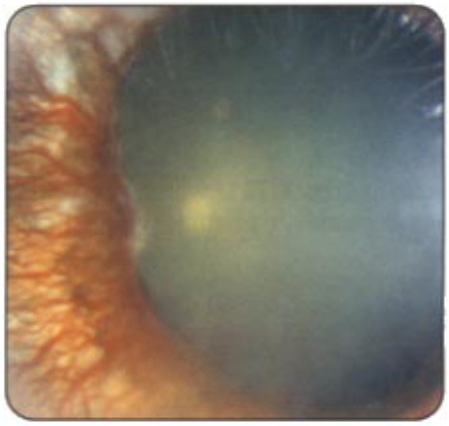


Fig. 8.107

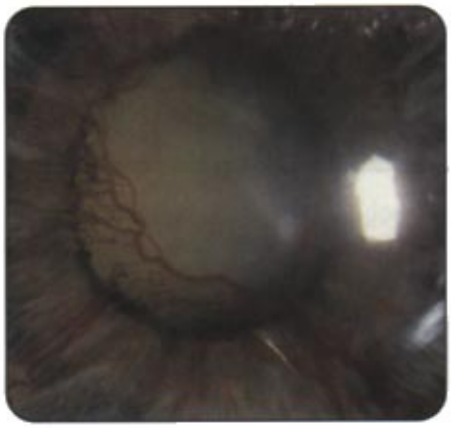


Fig. 8.108

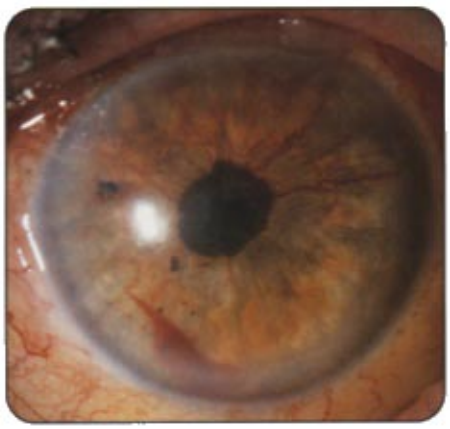


Fig. 8.109

Dilated iris capillaries

Dilated iris capillaries (**Fig. 8.110**) may be associated with various types of anterior uveitis. When seen in patients with secondary syphilis they are referred to as roseolae.

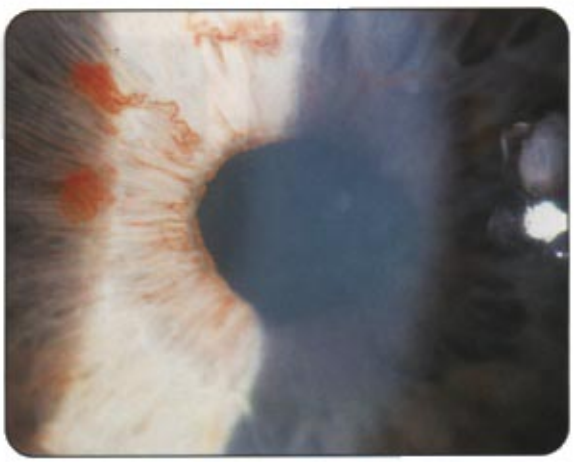


Fig. 8.110

Pupillary neovascular (Cobb) tufts

Pupillary neovascular tufts are tiny lesions (**Fig. 8.111**) which may cause spontaneous hyphemas (**Fig. 8.112**).

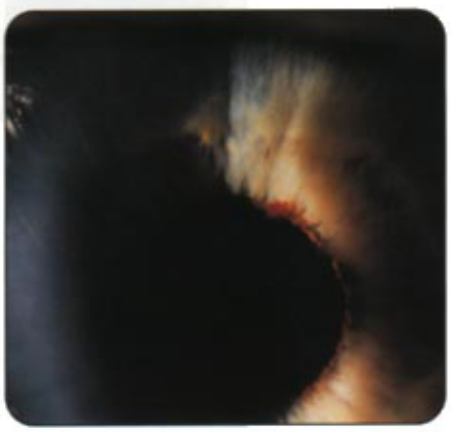


Fig. 8.111

Fig. 8.112

Cavernous hemangioma

Cavernous hemangioma is an extremely rare tumor which resembles a cluster of grapes (**Fig. 8.113**), just like its more common retinal counterpart.

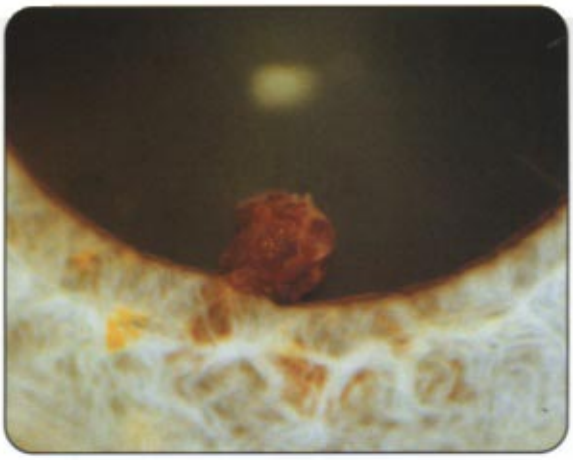


Fig. 8.113

9 The Lens

APHAKIA 216

ABNORMALITIES OF SHAPE 216

DISPLACED LENS 218

- Developmental
- Acquired

CONGENITAL AND INFANTILE CATARACT 220

Cataract types

- Anterior polar cataract
- Posterior polar cataract
- Coronary (supranuclear) cataract
- Cortical spoke-like cataract
- Lamellar cataract
- Central pulverulent cataract
- Sutural cataract
- Focal dot opacities

Systemic associations

DIFFERENTIAL DIAGNOSIS OF CHILDHOOD CATARACT 225

- In a healthy neonate
- In an unwell neonate
- In a healthy child
- In an unwell child

TYPES OF ACQUIRED CATARACT 227

- Anterior capsular
- Anterior subcapsular
- Posterior subcapsular
- Cortical
- Nuclear
- Nonopaque lens defects

OPACITIES IN PSEUDOPHAKIA 233

- Capsular
- Lenticular

1. Iatrogenic aphakia

This is the result of cataract extraction and no lens implantation. **Figure 9.1** shows an aphakic eye with vitreous herniating into the anterior chamber.

2. Spontaneous absorption

This is a very rare occurrence. The opaque capsule usually remains and is called a membranous cataract (**Fig. 9.2**).

Systemic associations

- Lowe syndrome.
- Hallermann–Streiff–François syndrome.

3. Congenital aphakia

This is very rare and is usually associated with microphthalmos.

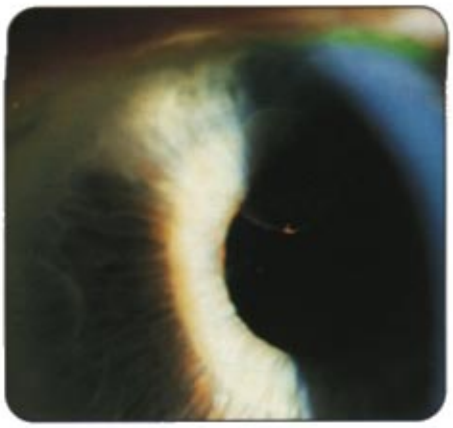


Fig. 9.1

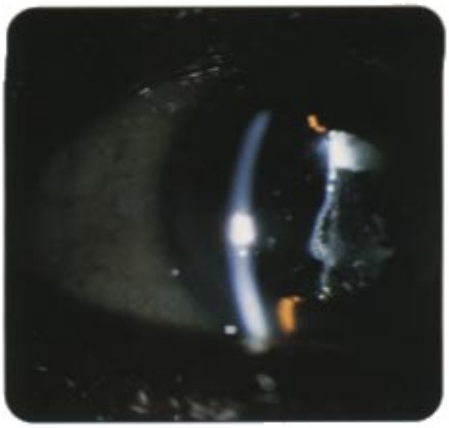


Fig. 9.2

ABNORMALITIES OF SHAPE

1. Coloboma

This is a rare condition which may be associated with other colobomatous defects of the iris, ciliary body, and choroid as well as microphakia and spherophakia.

Signs

- Notching of the inferior equator of the lens (**Fig. 9.3**) associated with absent zonules.

2. Tear-drop lens

This is a very rare form of coloboma where the inferior portion of the lens adheres to the site of fetal fissure closure.

Signs

- Tear-drop-shaped lens which is displaced inferiorly.

3. Microphakia (small lens)

This disorder is very rare and occurs in association with conditions that cause arrest of ocular development such as microphthalmos. It may, however, be an isolated finding (**Fig. 9.4**) or found in association with Lowe syndrome.

4. Microspherophakia

The systemic associations of microspherophakia are shown in **Table 9.1**.

Signs

- Lens with a small diameter and spherical shape, resulting in lenticular myopia (**Fig. 9.5**).
- Displacement, usually downward, may occur.
- Pupil-block glaucoma is a common complication.

5. Lentiglobus

This is a very rare, usually unilateral condition which causes induced myopia.

Signs

- Generalized hemispheric lens deformity which may be associated with posterior polar lens opacity.

6. Anterior lenticonus

This is a rare, bilateral condition which is associated with Alport syndrome.

Signs

- Localized anterior conoid projection of the center of the lens (**Fig. 9.6**).
- On direct ophthalmoscopy, early cases give rise rise to an 'oil-droplet' sign similar to that seen in keratoconus (**Fig. 9.7**).
- Lenticular opacification may develop later.

7. Posterior lenticonus

This is a rare, bilateral condition which may occur in isolation or in association with Lowe syndrome.

Signs

- Localized posterior conoid projection of the lens (**Figs 9.8, 9.9**).
- It may be associated with posterior lens opacity (*see* **Figs 9.33, 9.34**).

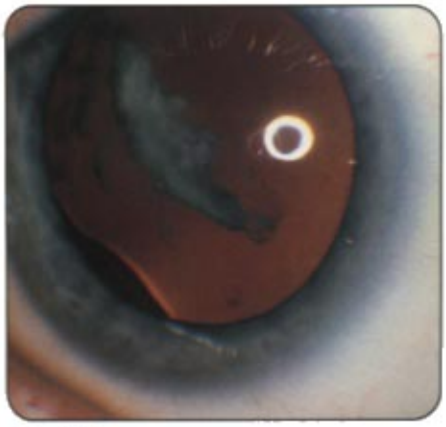


Fig. 9.3

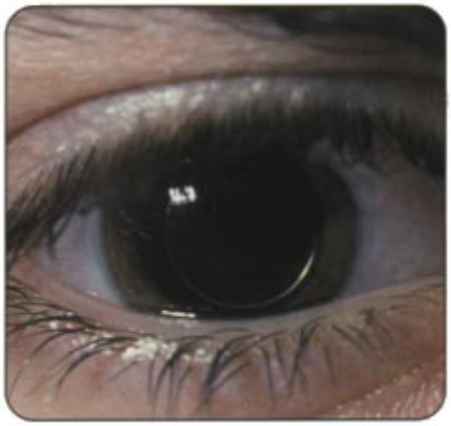


Fig. 9.4

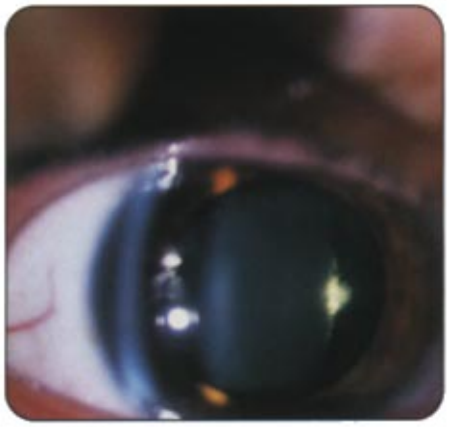


Fig. 9.5

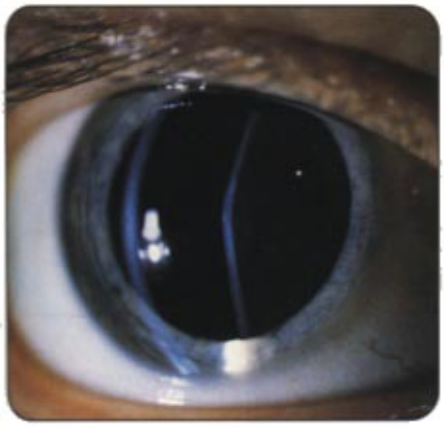


Fig. 9.6

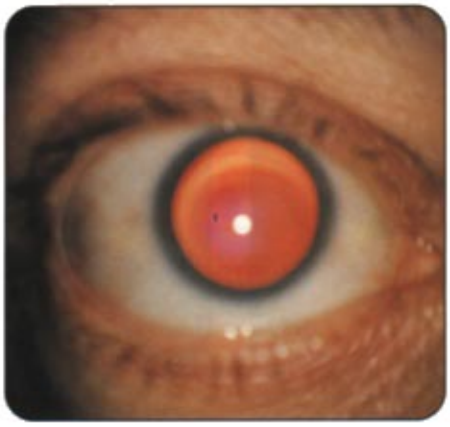


Fig. 9.7

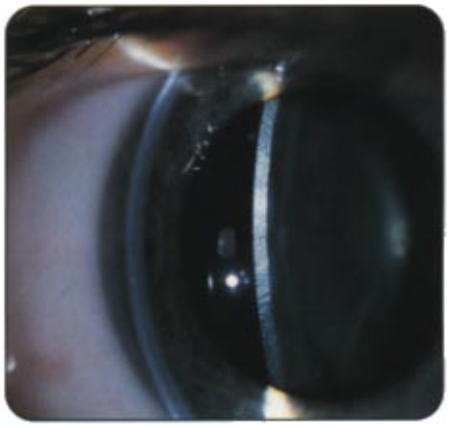


Fig. 9.8

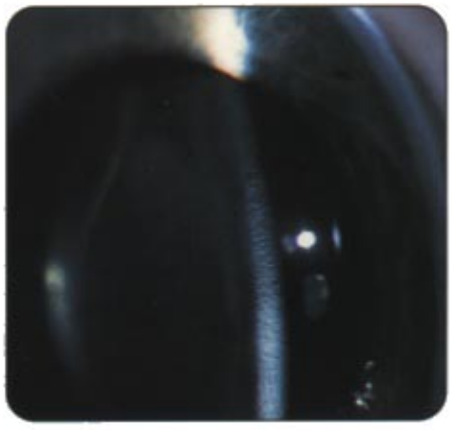


Fig. 9.9

Table 9.1 Associations of microspherophakia

Systemic syndromes
• Weill–Marchesani
• Marfan
• Alport
Miscellaneous
• familial (dominant)
• hyperlysinemia
• familial ectopia lentis et pupillae
• Peters anomaly
• congenital rubella

DISPLACED LENS

Developmental

Ocular associations

- Megalocornea (**Figs 9.10, 9.11**).
- Severe buphthalmos (**Fig. 9.12**).
- Very high myopia.
- Aniridia (**Figs 9.13, 9.14**).

Hereditary causes without systemic manifestations

- Familial ectopia lentis (dominant).
- Ectopia lentis et pupillae (recessive) in which pupil and lens are displaced in opposite directions (**Fig. 9.15a and b**). It may be associated with microspherophakia.
- Isolated familial microspherophakia.

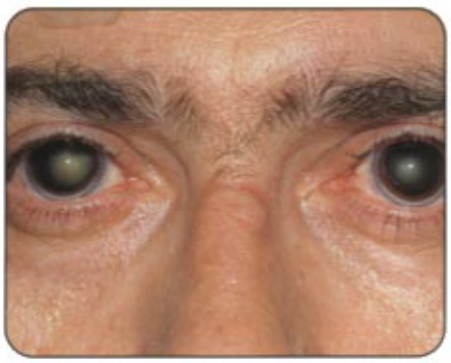


Fig. 9.10

Fig. 9.11

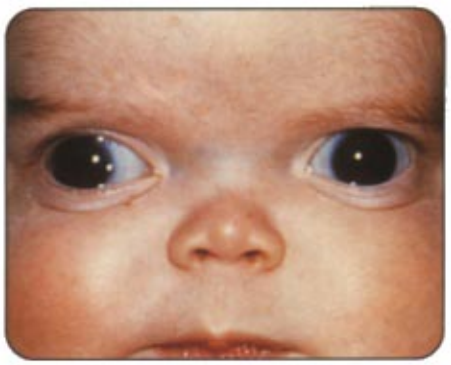


Fig. 9.12

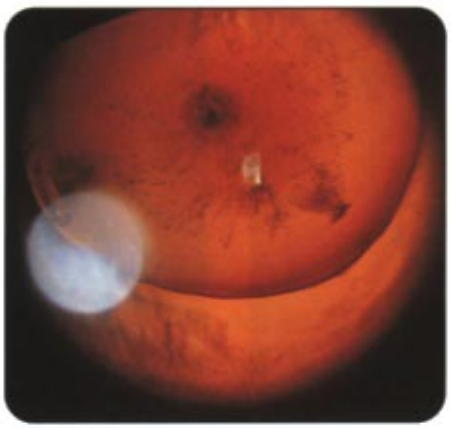


Fig. 9.13

Fig. 9.14

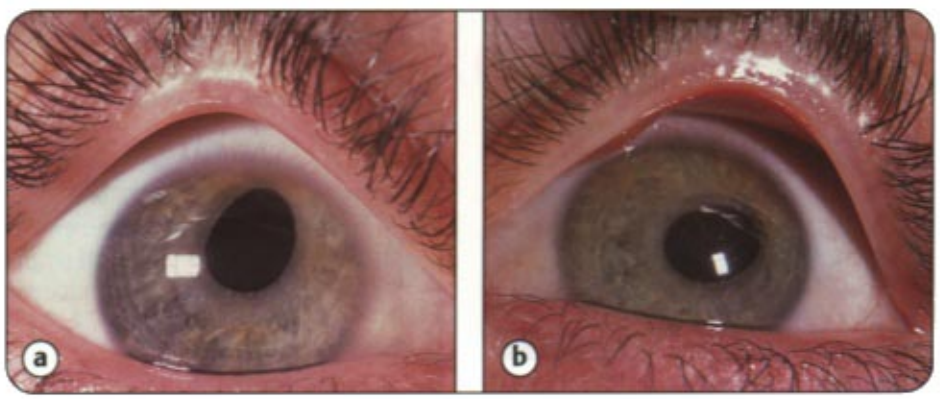


Fig. 9.15

Systemic syndromes

- Marfan – subluxation is usually upward and inward (**Figs 9.16a and b, 9.17a and b**) but may be in any direction. The lens may occasionally be microspherophakic.
- Weill–Marchesani (microspherophakia) – subluxation is usually inferior.
- Ehlers–Danlos.
- Stickler.
- Kneist.
- Mandibulofacial dysostosis.
- Osteogenesis imperfecta.

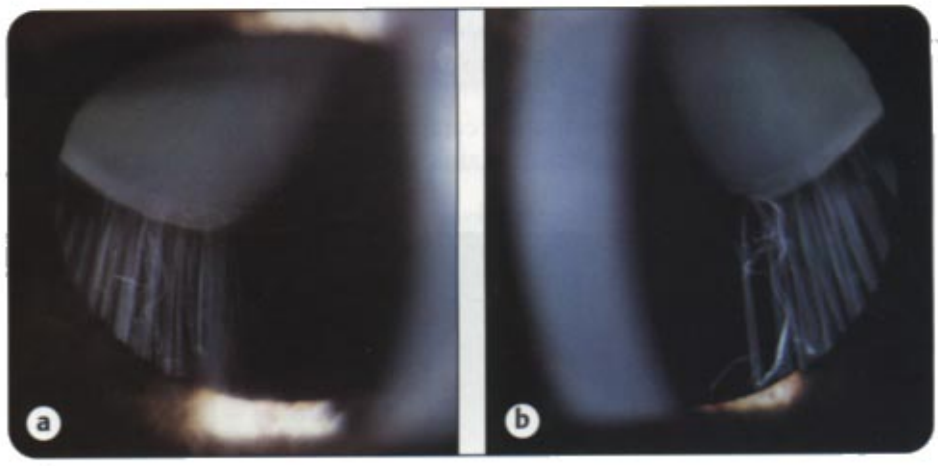


Fig. 9.16

Metabolic disorders

- Homocystinuria.
- Hyperlysinemia (microspherophakia),
- Sulfite oxidase deficiency.

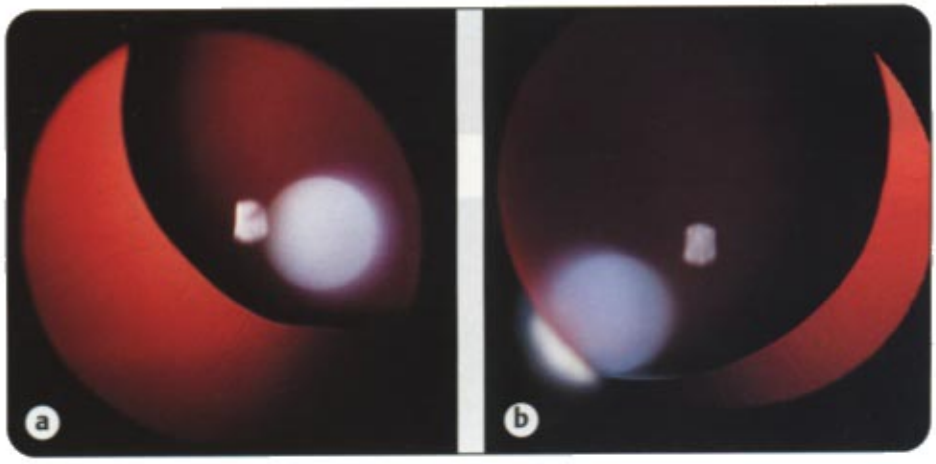


Fig. 9.17

Acquired

Causes

- Blunt ocular trauma – **Figure 9.18** shows nasal subluxation of a mature cataract; **Figure 9.19** shows a lens under the conjunctiva.

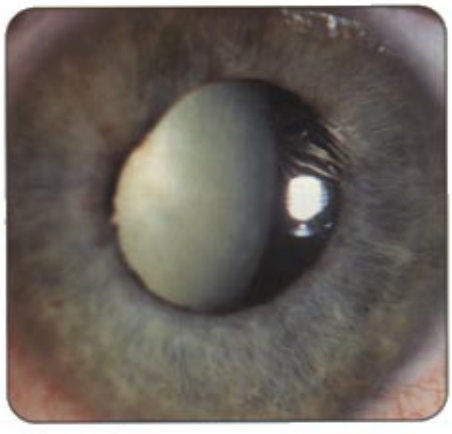


Fig. 9.18

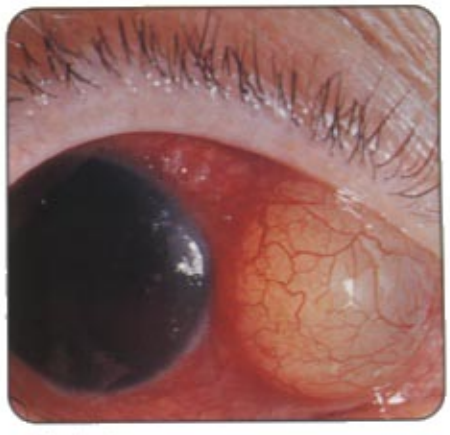


Fig. 9.19

- Anterior uveal tumors – **Figure 9.20** shows a ciliary body melanoma which may cause a slight shift of the lens as well as localized lens opacity.
- Hypermaturity – in a hypermature cataract the cortex liquefies and the hard nucleus may sink to the bottom (**Fig. 9.21**); less frequently, the entire lens may sublux-ate inferiorly (**Fig. 9.22**).
- Degenerate eye (**Fig. 9.23**).
- Syphilis.

Fig. 9.20

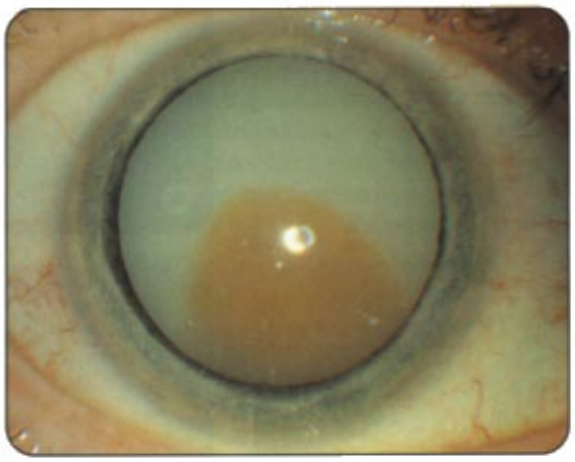


Fig. 9.21

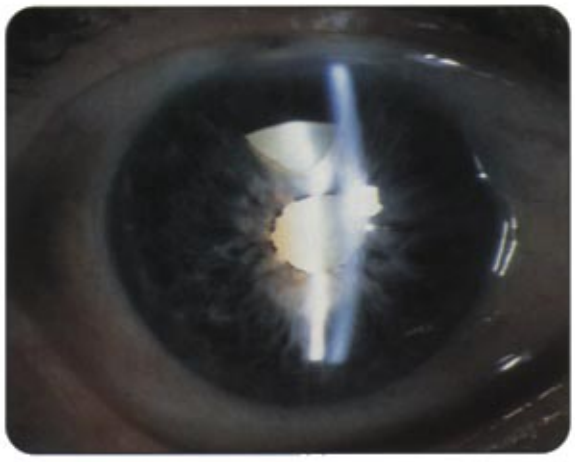


Fig. 9.22

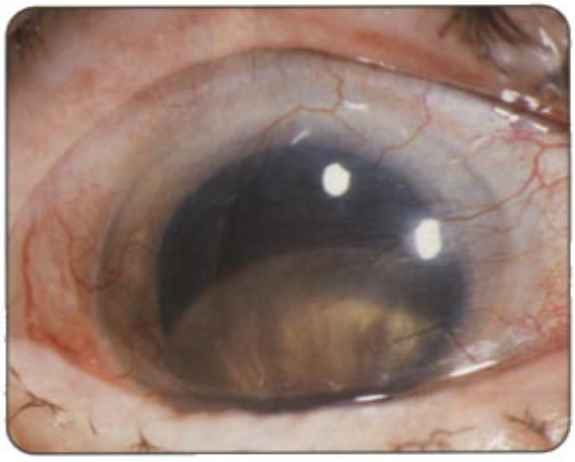


Fig. 9.23

CONGENITAL AND INFANTILE CATARACT

A congenital cataract is present at birth and an infantile cataract develops during the first year of life. Because some congenital lens opacities escape detection at birth and are noted later, these two terms are generally used interchange-ably. About a third are inherited, a third are associated with systemic diseases, and a third are idiopathic. Hereditary cataracts and those associated with systemic disorders are usually bilateral, whereas sporadic cases may be either bi-lateral or unilateral. About 50% of eyes with congenital cataracts have other ocular anomalies.

Cataract types

Anterior polar cataract

Anterior polar cataracts may be dominantly inherited.

Signs

- The opacity may involve only the capsule (**Fig. 9.24**).
- The opacity may be pyramidal and project into the anterior chamber (**Figs 9.25, 9.26**).

Ocular associations

- Persistent pupillary membrane (**Figs 9.27, 9.28**).
- Anterior lenticonus.
- Peters anomaly (**Figs 9.29, 9.30**).
- Microphthalmos.
- Aniridia (**Fig. 9.31**).
- Pigmented 'chicken tracks' on the central capsule (**Fig. 9.32**).

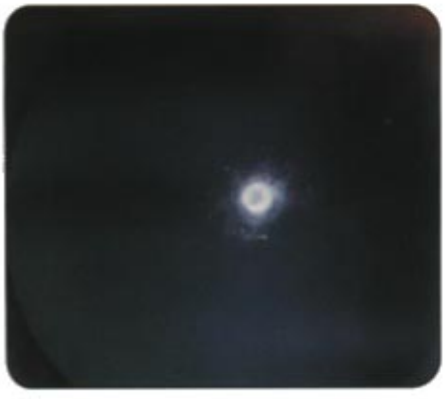


Fig. 9.24

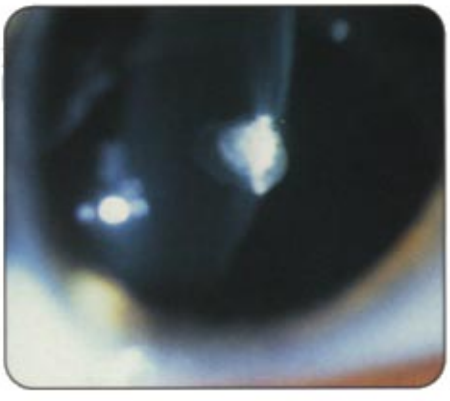


Fig. 9.25

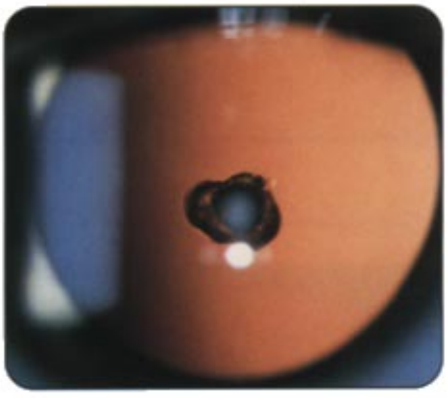


Fig. 9.26

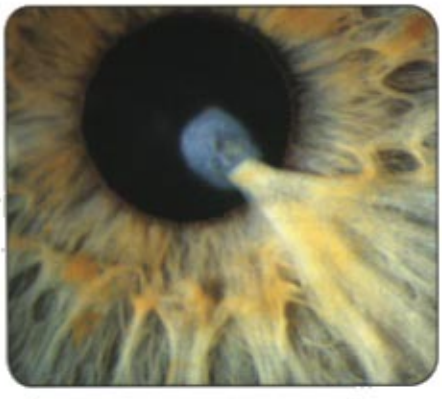


Fig. 9.27

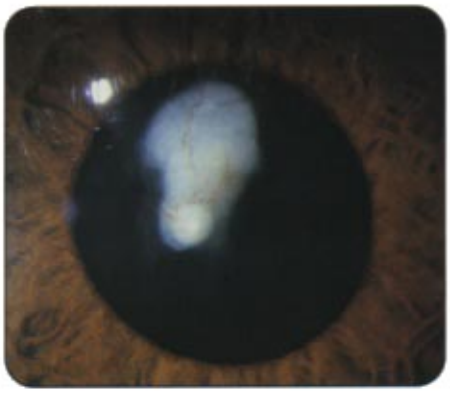


Fig. 9.28

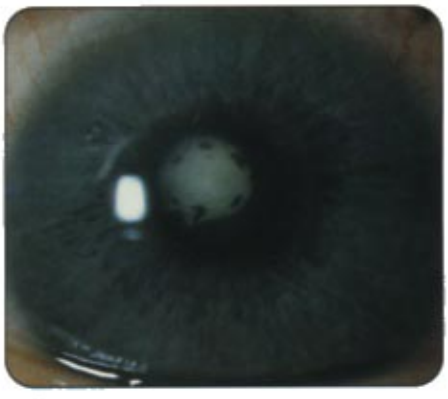


Fig. 9.29

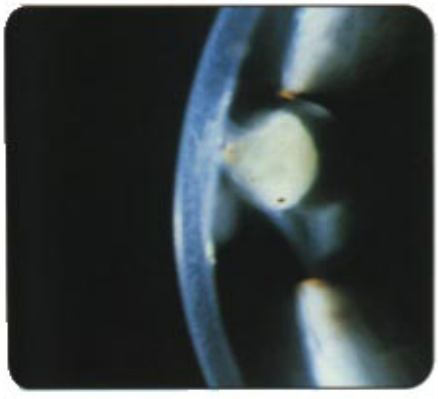


Fig. 9.30

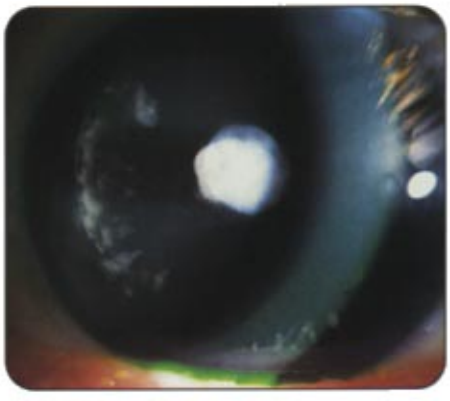


Fig. 9.31

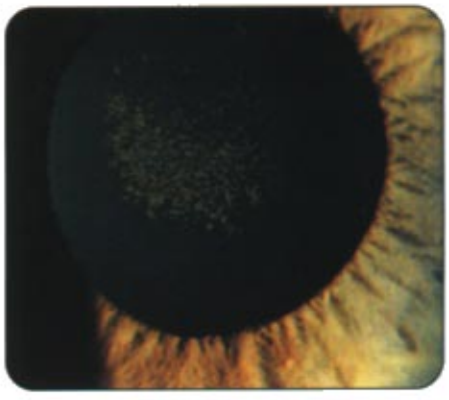


Fig. 9.32

Posterior polar cataract

Signs

- The opacity may involve only the capsule or it may form a plaque.

Ocular associations

- Persistent hyaloid remnants (Mittendorf dots).
- Posterior lenticonus (**Figs 9.33, 9.34**).
- Persistent hyperplastic primary vitreous (**Figs 9.35, 9.36**).

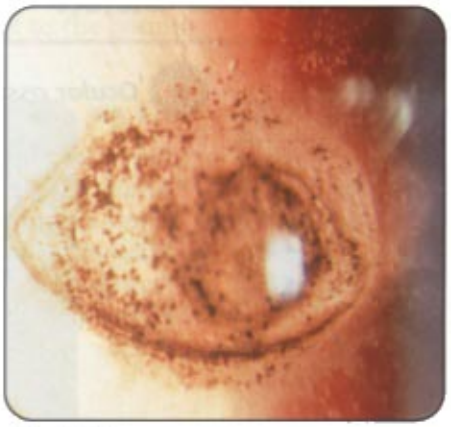


Fig. 9.33

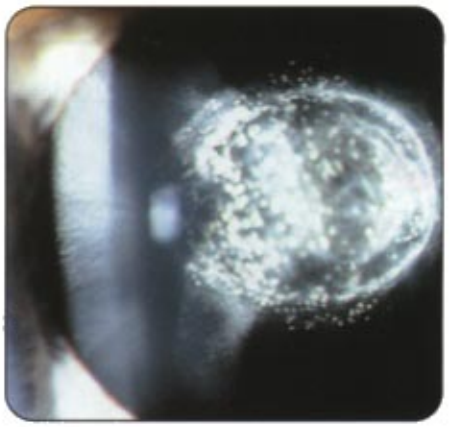


Fig. 9.34

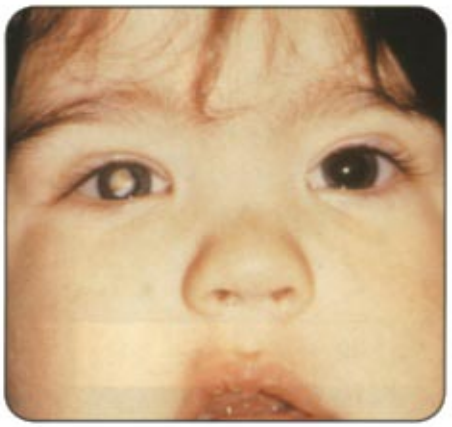


Fig. 9.35

Fig. 9.36

Coronary (supranuclear) cataract

A coronary cataract is usually sporadic and only occasionally hereditary.

Signs

- Round opacities are located in the deep cortex (**Figs 9.37, 9.38**) and surround the nucleus like a crown (**Fig. 9.39**).

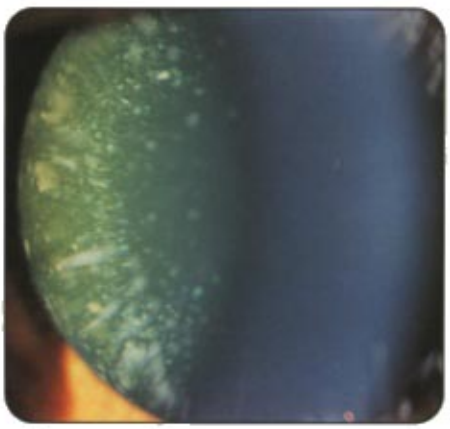


Fig. 9.37

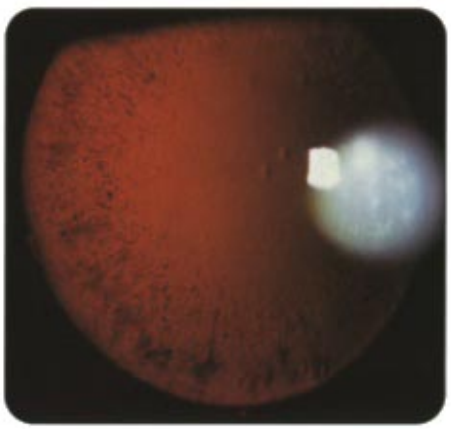


Fig. 9.38

Fig. 9.39

Cortical spoke-like cataract

Signs

- Wedge-shaped peripheral cortical opacities (**Fig. 9.40**).

Systemic associations

- Fabry disease.
- Mannosidosis.

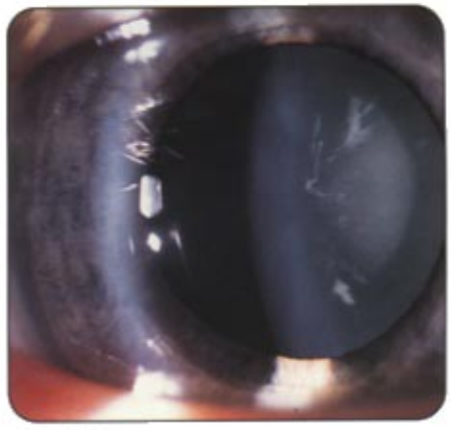


Fig. 9.40

Lamellar cataract

A lamellar cataract is one of the most common infantile forms. Inheritance is usually dominant but occasionally recessive. It is therefore important to examine family members as there is variable expressivity.

Signs

- Round central shell-like opacity surrounding a clear nucleus (**Figs 9.41, 9.42**) which may be associated with riders (**Fig. 9.43**).
- In some cases, an initially lamellar cataract may subsequently progress to involve the entire nucleus (**Fig. 9.44**).

Systemic associations

- Galactosemia.
- Hypoglycemia.
- Hypocalcemia.

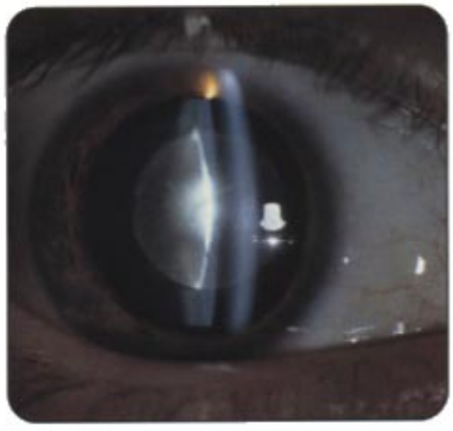


Fig. 9.41

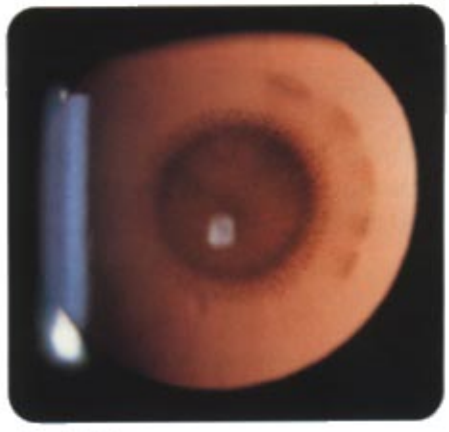


Fig. 9.42

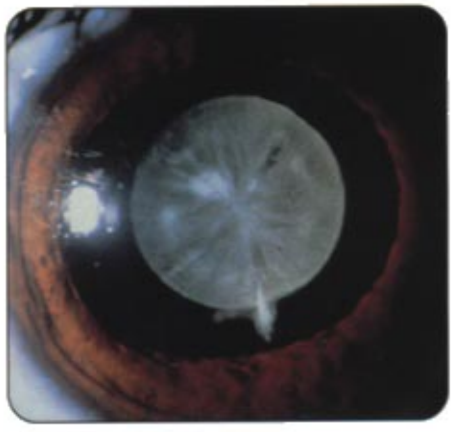


Fig. 9.43

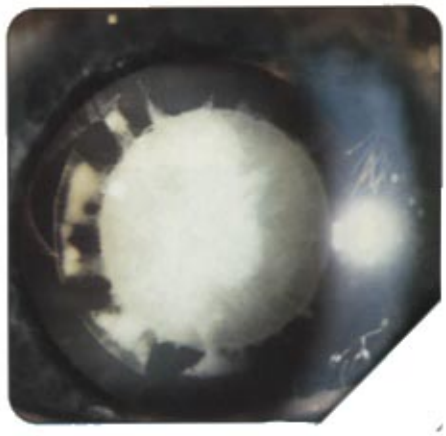


Fig. 9.44

Central pulverulent cataract

Central pulverulent cataracts are dominantly inherited and nonprogressive.

Signs

- Spheroidal opacity between 1 and 4 mm in diameter within the lens nucleus, with a clearer center (**Figs 9.45, 9.46**).

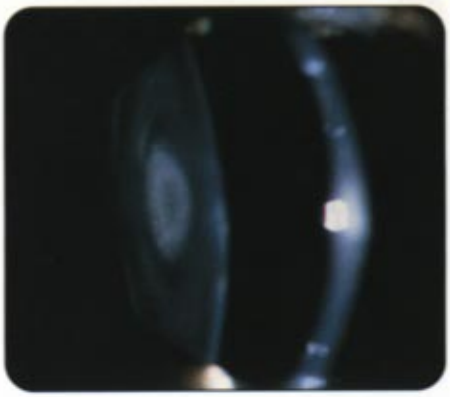


Fig. 9.45

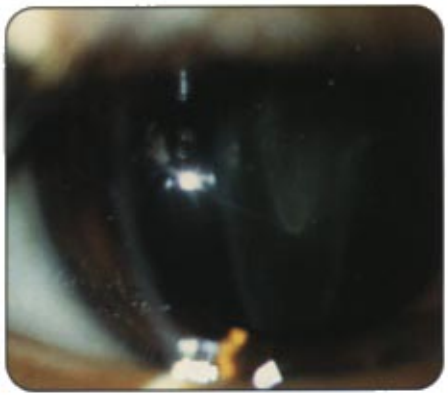


Fig. 9.46

Sutural cataract

Inheritance of sutural cataracts is usually X-linked, in which the males have a significant opacity and the females only a minor one.

Signs

- The opacity follows the shape of the anterior or posterior Y suture (**Figs 9.47, 9.48**).

Fig. 9.47

Fig. 9.48

Focal dot opacities

Signs

- Blue dot cortical opacities (**Figs 9.49, 9.50**) are extremely common and innocuous and they may coexist with other types of congenital cataract (**Fig. 9.51**).

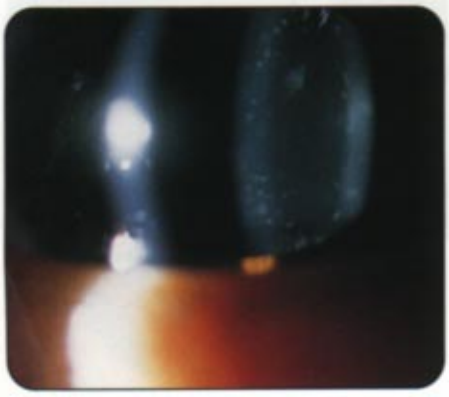


Fig. 9.49

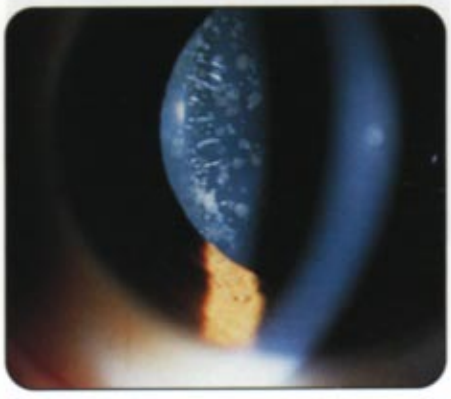


Fig. 9.50

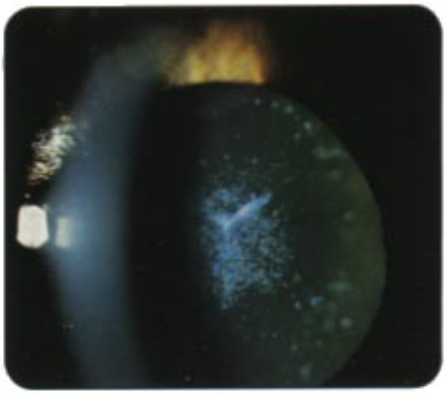


Fig. 9.51

Systemic associations

Table 9.2 Systemic associations of congenital and infantile cataract

Maternal intrauterine infections	**Craniofacial syndromes**
• rubella	• Crouzon
• toxoplasmosis	• Apert
• cytomegalovirus	• Pierre Robin
• varicella	• Hallermann–Streiff–François
Metabolic disorders	• Stickler
• galactosemia	**Chromosomal abnormalities**
• galactokinase deficiency	• Down syndrome (trisomy 21)
• hypoglycemia	• Patau syndrome (trisomy 13)
• hypocalcemia	• Edward syndrome (trisomy 18)
• Lowe syndrome	• cri-du-chat syndrome (deletion of short arm of chromosome 5)
• congenital hemolytic jaundice	**Skin abnormalities**
Central nervous system abnormalities	• incontinentia pigmenti (Bloch–Sulzberger syndrome)
• Zellweger syndrome	• Cockayne syndrome
• Sjögren–Larsson syndrome	• congenital ichthyosis
• Norrie disease	

DIFFERENTIAL DIAGNOSIS OF CHILDHOOD CATARACT

In a healthy neonate

In healthy neonates, the cataract may be unilateral (**Fig. 9.52**) or bilateral.

1. Hereditary (usually autosomal dominant)

Look for

- Similar cataract in family members.

2. Associated with other ocular anomalies

- Persistent hyperplastic primary vitreous.
- Aniridia.
- Colobomas.
- Microphthalmos.
- Buphthalmos (**Fig. 9.53**), which may be present in Lowe syndrome.

3. Maternal radiation

When this occurs during the first trimester of pregnancy.

4. Idiopathic

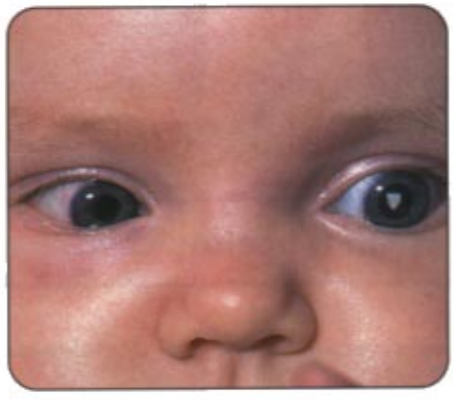


Fig. 9.52

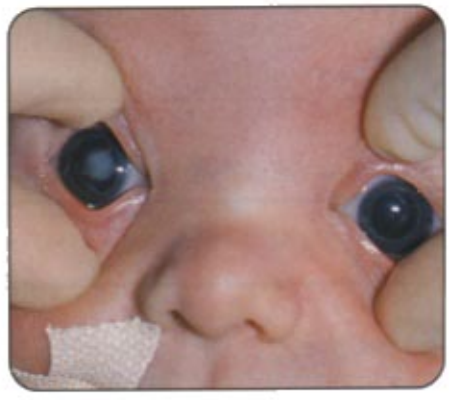


Fig. 9.53

In an unwell neonate

In systemically unwell neonates (**Fig. 9.54**), the cataract is usually bilateral.

1. Lowe syndrome

Look for

- Glaucoma (*see* **Fig. 9.53**).

2. Intrauterine infections

Signs

- Dense central cataracts.

3. Galactosemia

The cataract develops during the postnatal period.

Signs

- Early changes have a central oil-droplet appearance (**Fig. 9.55**).
- Unless treated with a galactose-restricted diet, lamellar opacities develop (*see* **Fig. 9.41**) and the cataract may eventually become mature.

4. Hallermann–Streiff–François syndrome

5. Neonatal hypoglycemia

6. Neonatal hypocalcemia

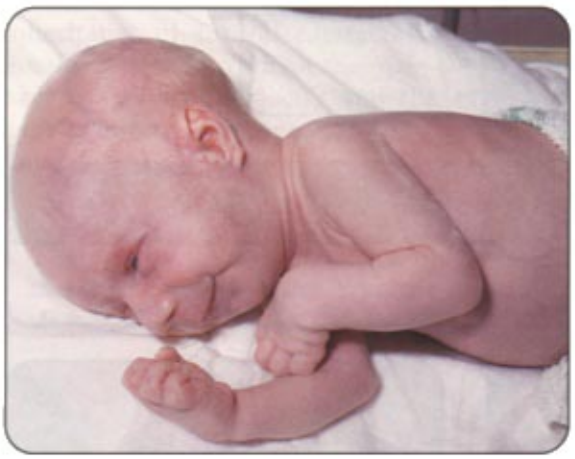


Fig. 9.54

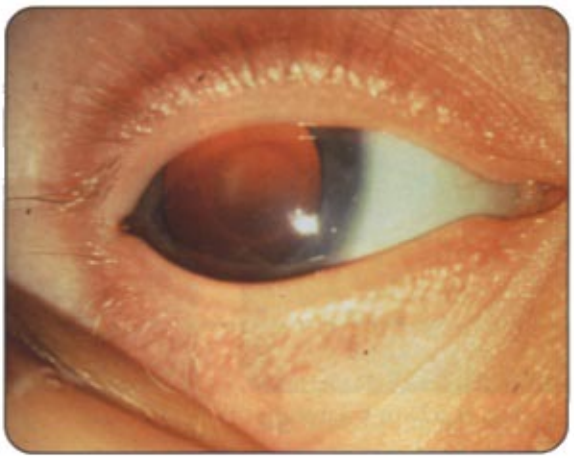


Fig. 9.55

In a healthy child

1. Hereditary

- Anterior polar (dominant).
- Lamellar (usually dominant or occasionally recessive).
- Sutural (X-linked).
- Coronary (occasionally inherited).

2. Galactokinase deficiency

3. Idiopathic

Fifty percent of bilateral infantile cataract cases are idiopathic and over 50% of unilateral cataracts are idiopathic.

In an unwell child

1. Fabry disease
This causes cortical spoke-like opacities.

2. Mannosidosis
This causes cortical spoke-like opacities.

Look for

- Corneal clouding.

3. Alport syndrome

4. Wilson disease
This causes a disc-shaped central polychromatic opacity with peripheral radiations (sunflower). Cataract is found in less than 10% of cases.

Look for

- Kayser–Fleischer ring.

5. Skin abnormalities

6. Hypoparathyroidism
This is associated with lamellar cataract.

7. Diabetes mellitus
May cause white snowflake cortical opacities which may progress quickly.

8. Chromosomal abnormalities

TYPES OF ACQUIRED CATARACT

Anterior capsular

1. True exfoliation
This is extremely rare and is caused by exposure to infrared radiation (glass-blowers, furnace workers, and desert dwellers).

2. Pseudoexfoliation syndrome
This is a common condition which affects elderly patients and is bilateral in 50% of cases.

Signs

- White flecks on the pupil margin (**Fig. 9.56**).
- White flecks on the anterior lens capsule (**Figs 9.57, 9.58**) in a bull's-eye configuration (**Fig. 9.59**).

3. Pigmentation on anterior capsule

Causes

- In pigment dispersion syndrome (**Fig. 9.60**).
- Following iritis – often at the site of previous posterior synechiae (**Figs 9.61–9.63**).
- Following blunt trauma – Vossius ring (**Figs 9.64, 9.65**).
- Drug-induced – chlorpromazine (**Fig. 9.66**), amiodarone, gold, and tamoxifen.
- Copper deposits – blue–green discoloration in a sunflower pattern seen in Wilson disease.
- Mercury exposure – both topical and systemic may give rise to a gray sheen.

Fig. 9.56

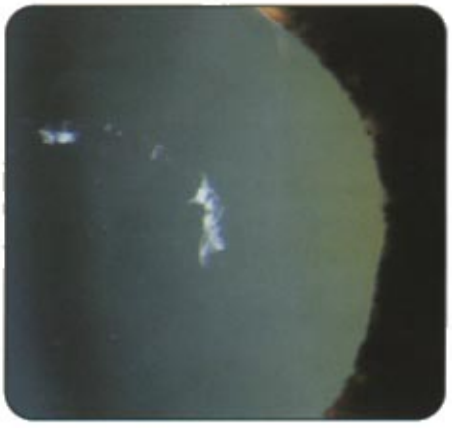


Fig. 9.57

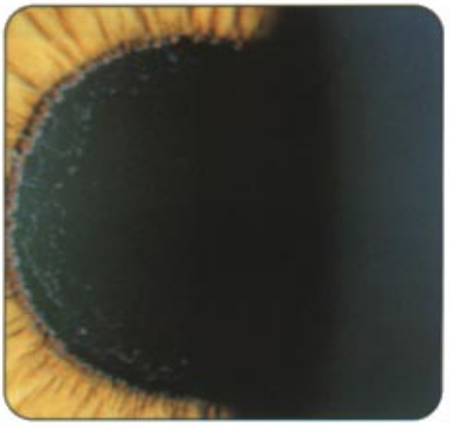


Fig. 9.58

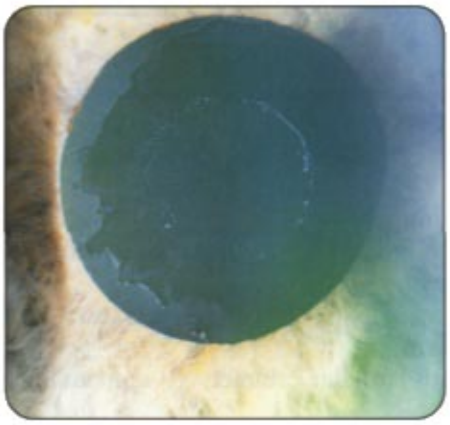


Fig. 9.59

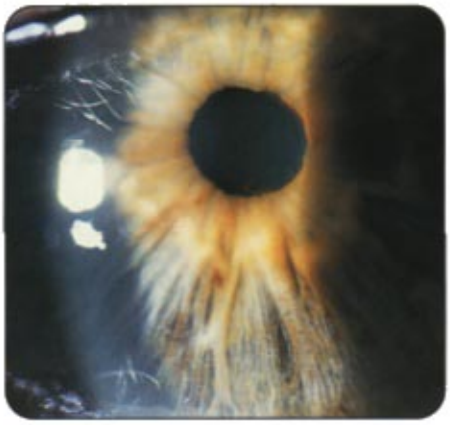


Fig. 9.60

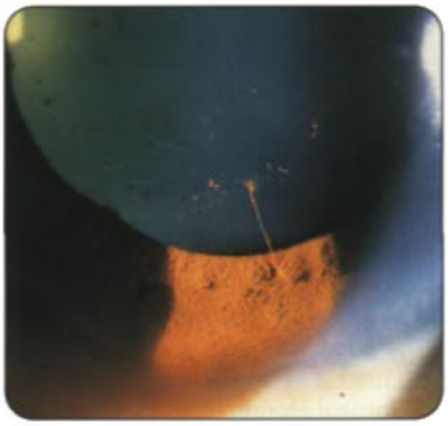
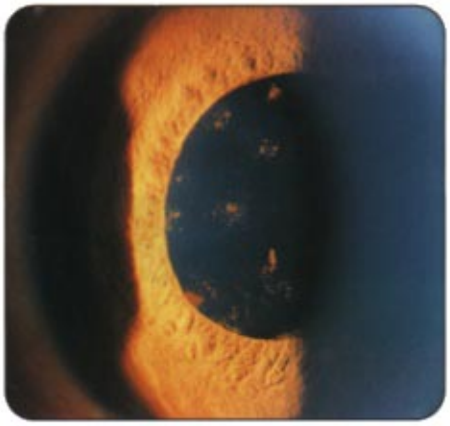


Fig. 9.61



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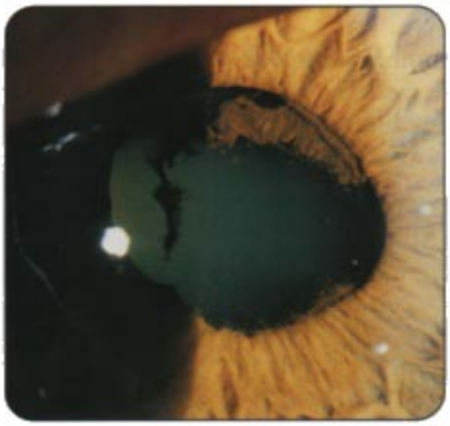

Fig. 9.63

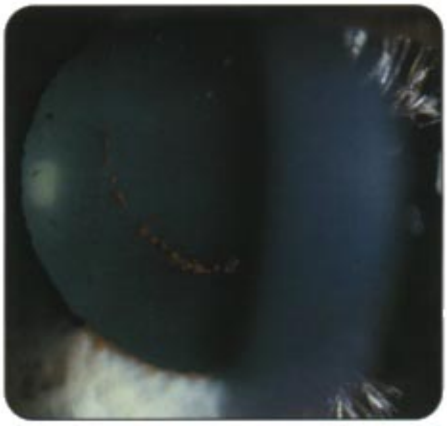

Fig. 9.64

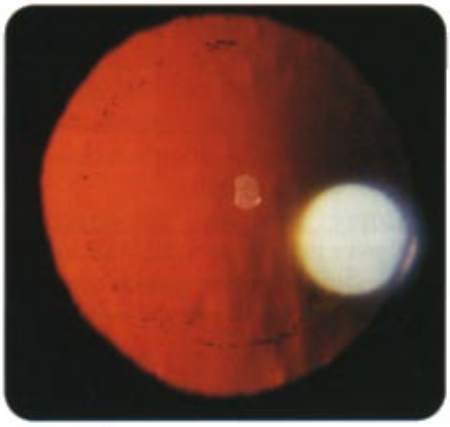

Fig. 9.65

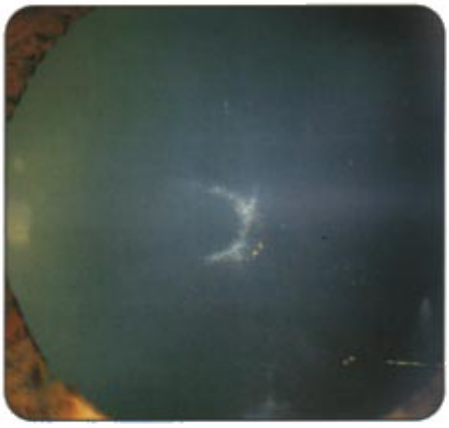

Fig. 9.66

Anterior subcapsular

1. Glaukomflecken

This is a common, usually unilateral condition which follows an acute congestive attack of angle-closure glaucoma.

Signs

- Gray–white opacities in the pupillary zone which resemble 'spilled milk'; associated with iris atrophy (**Fig. 9.67**).

2. Fibrous plaques

These are uncommon lens opacities caused by epithelial hyperplasia.

Causes

- Atopic dermatitis (**Fig. 9.68**).
- Chronic iritis.
- Trauma.

3. Feathery opacities

Causes

- Ionizing radiation.
- Intravitreal gas.
- Long-term use of strong miotics.

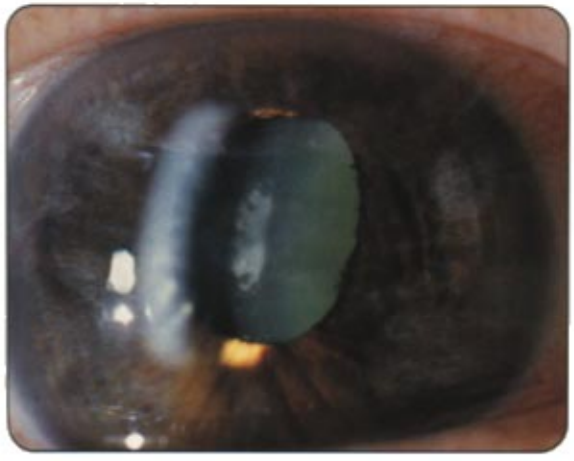

Fig. 9.67

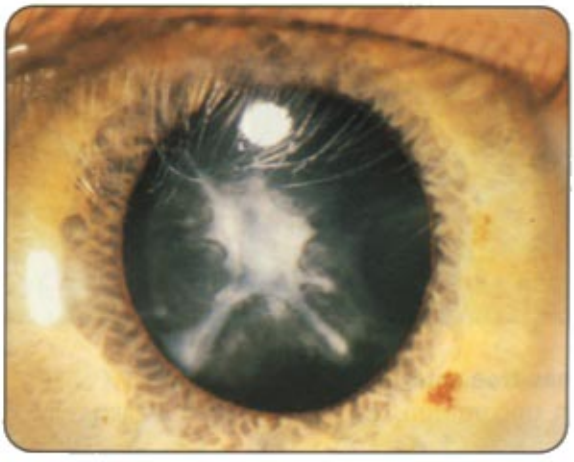

Fig. 9.68

Posterior subcapsular

Posterior subcapsular cataracts may be age-related or associated with the following conditions.

1. Chronic intraocular inflammation

Chronic intraocular inflammation, and Fuchs heterochromic cyclitis in particular, very frequently presents with unilateral presenile cataract.

Signs

- Initially, a polychromatic luster of the posterior capsule (**Fig. 9.69**).
- Later, subcapsular cataract (**Fig. 9.70**).
- In longstanding cases, the cataract may become mature (**Fig. 9.71**).

2. Posterior segment disease

- Degenerative myopia, which is also associated with nuclear cataract.
- Hereditary retinal dystrophies such as retinitis pigmentosa and gyrate atrophy.

3. Drug-induced

- Topical and systemic steroids.
- Busulfan.

4. Neurofibromatosis-II

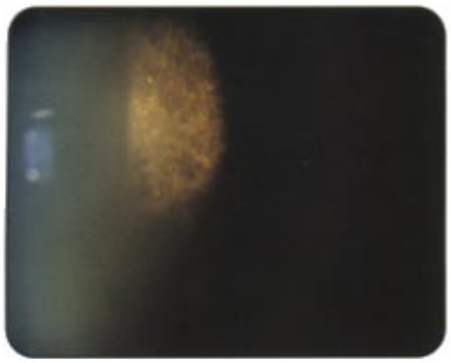

Fig. 9.69

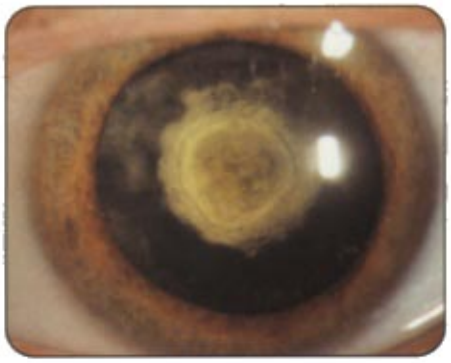

Fig. 9.70

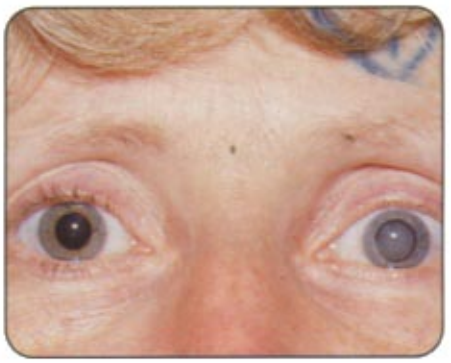

Fig. 9.71

Cortical

1. Cuneiform cataract
This is the most common type of age-related lens opacity.

Signs

- Starts as clefts between lens fibers (**Figs 9.72–9.74**).
- Progresses to spoke-like opacities (**Fig. 9.75**).

2. Snowflake cataract
This occurs in young diabetics. It has an abrupt onset and may become mature within a few days.

3. Christmas-tree cataract
This is an uncommon but striking type of age-related cataract.

Signs

- Polychromatic, needle-like deposits in the deep cortex and nucleus (**Figs 9.76, 9.77**).
- Other opacities (**Fig. 9.78**) may coexist.

4. Stellate posterior cortical cataract
This is associated with myotonic dystrophy.

Signs

- Initially, fine polychromatic posterior subcapsular granules (**Fig. 9.79**).
- Later, posterior cortical star-shaped opacity (**Fig. 9.80**).

5. Retro-dot opacities
These are common in the aging lens.

Signs

- Small dots which are best seen by retroillumination (**Fig. 9.81**).

6. Flower-shaped opacity
This (**Fig. 9.82**) may be caused by blunt trauma.

7. Localized
These may be caused by penetrating trauma (**Fig. 9.83**) and tumors of the anterior uvea.

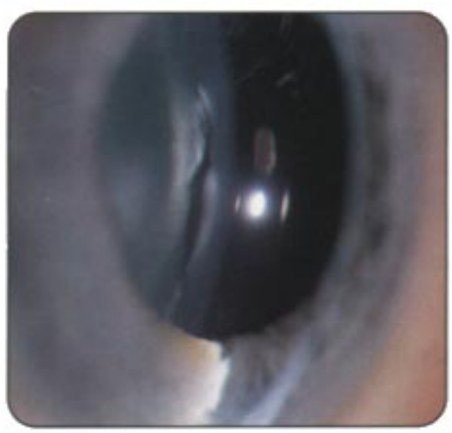

Fig. 9.72

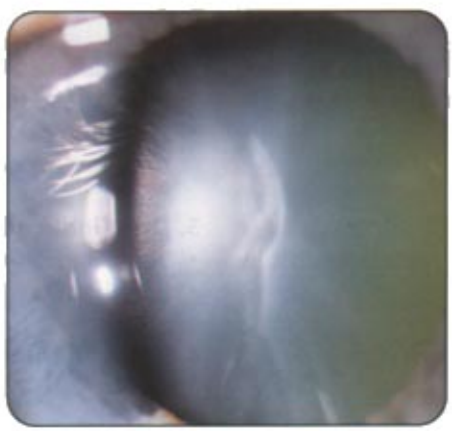

Fig. 9.73

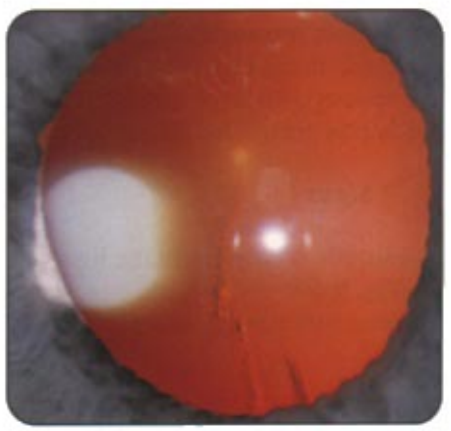

Fig. 9.74

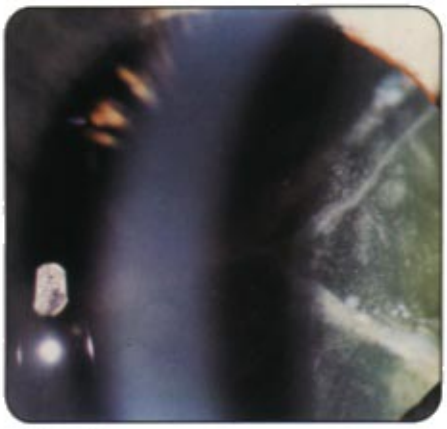

Fig. 9.75

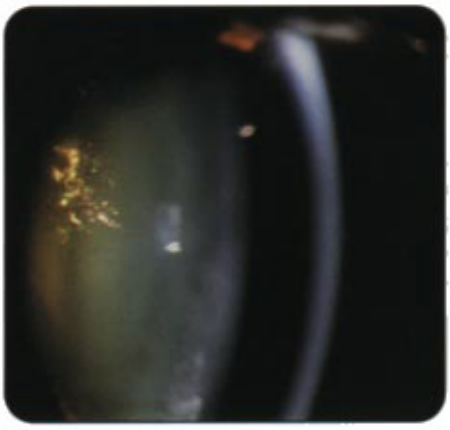

Fig. 9.76

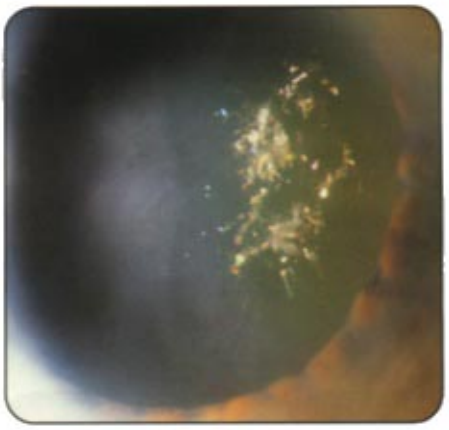

Fig. 9.77

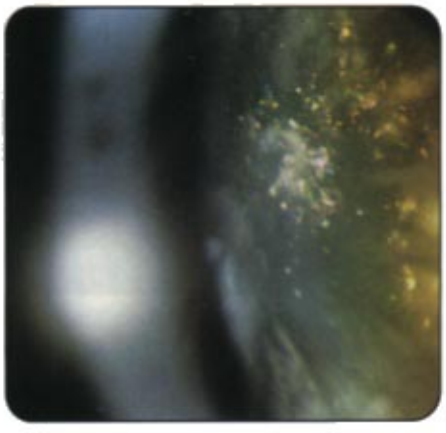

Fig. 9.78

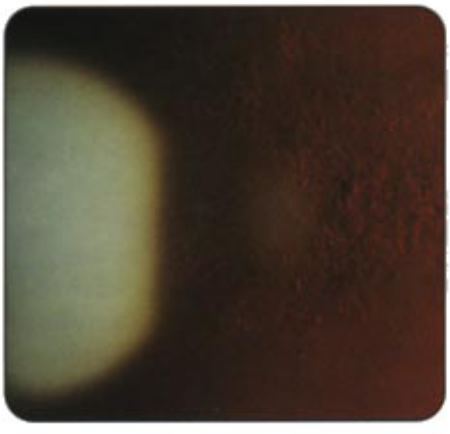

Fig. 9.79

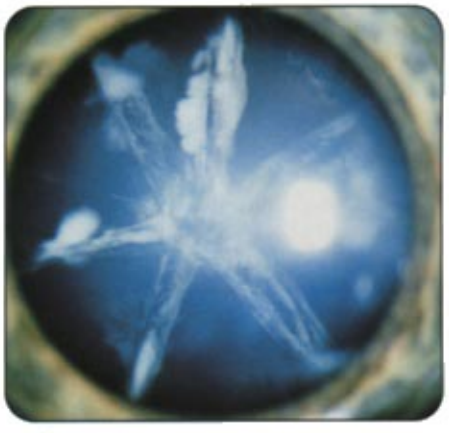

Fig. 9.80

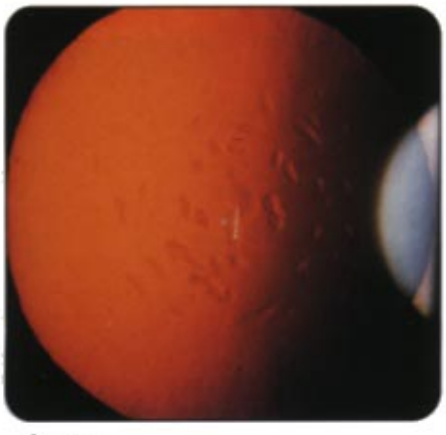

Fig. 9.81

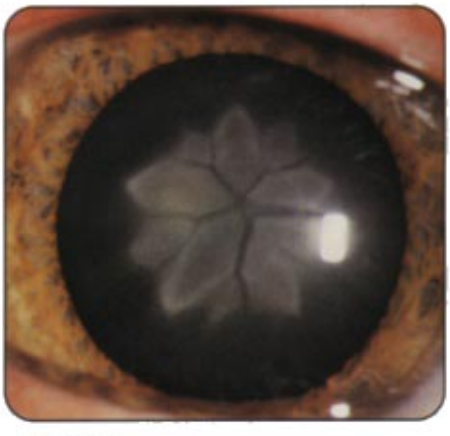

Fig. 9.82

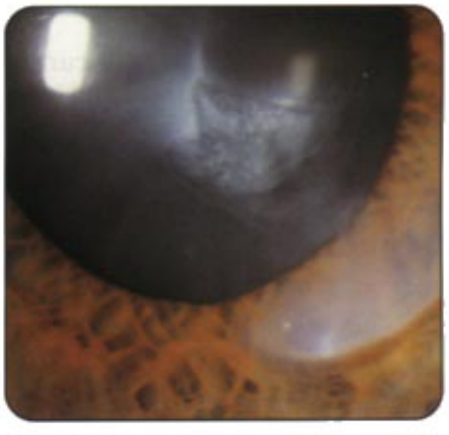

Fig. 9.83

Nuclear

Signs

- Nuclear cataracts (**Fig. 9.84**) vary in density and color, from yellow (**Fig. 9.85**), through amber (**Fig. 9.86**), brown, to virtually black (cataract nigrans).

Causes

- Age-related.
- Degenerative myopia.
- Postvitrectomy.
- Diabetes may cause early-onset nuclear cataract.

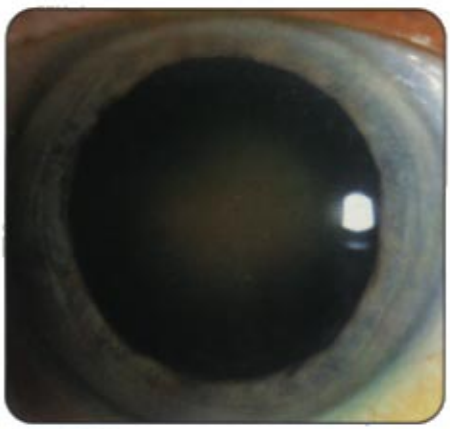

Fig. 9.84

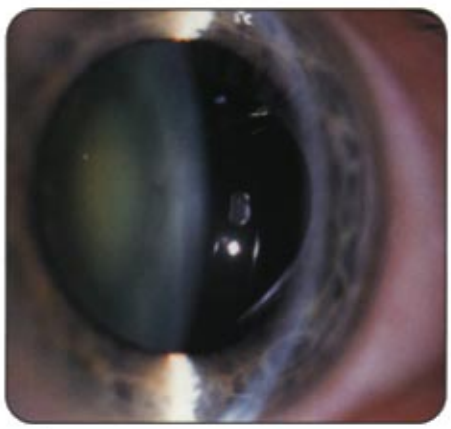

Fig. 9.85

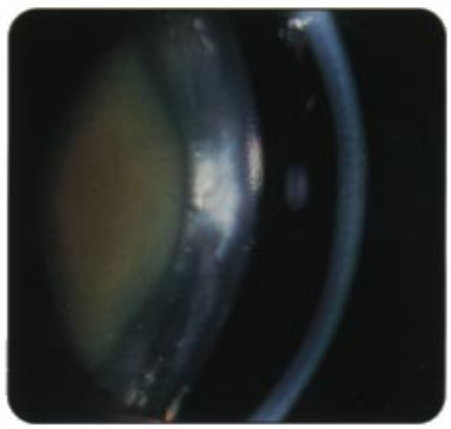

Fig. 9.86

Non-opaque lens defects

Nonopaque lens defects occur in association with cataract and may represent an aspect of the cataract-forming process.

1. Fiber folds
These are parallel, white ridges which occasionally branch and rejoin. They are most common in the lower half of the superficial cortex.

2. Water clefts
These are radial, wedge-like defects in the cortex which may cause ghosting of images. **Figure 9.87** shows both water clefts and vacuoles.

3. Vacuoles
These are cystoid spaces in the superficial cortex (**Figs 9.88–9.90**).

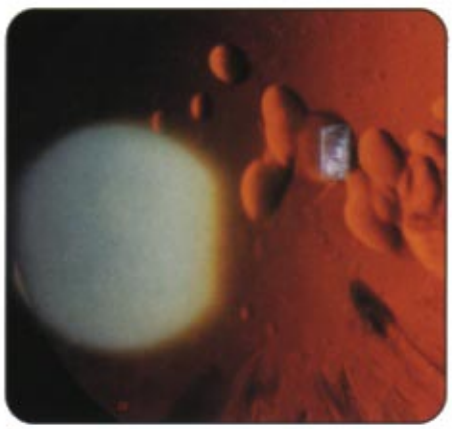

Fig. 9.87

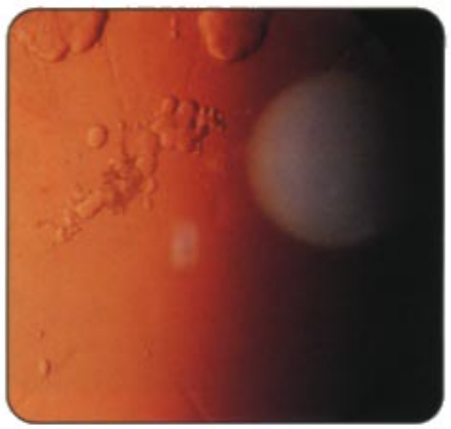

Fig. 9.88

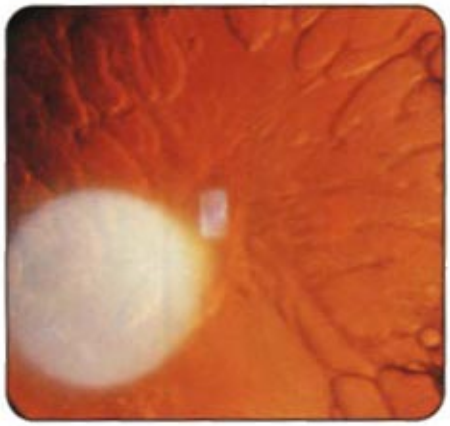

Fig. 9.89

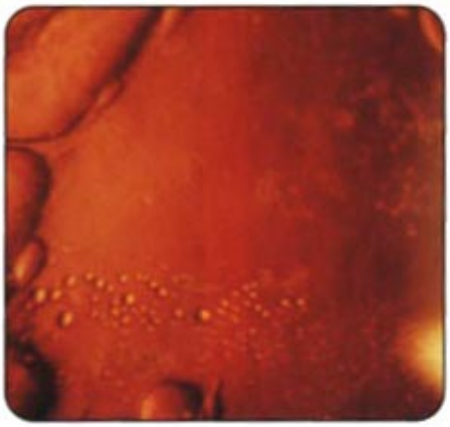

Fig. 9.90

Table 9.3 Systemic causes of acquired cataract

- diabetes
- atopic dermatitis
- myotonic dystrophy
- hypocalcemia
- Wilson disease
- neurofibromatosis–II

OPACITIES IN PSEUDOPHAKIA

Capsular

1. Capsular fibrosis

This usually appears within 2 to 6 months after surgery. Fibrosis of the anterior capsule, without phimosis (**Fig. 9.91**) or with (**Fig. 9.92**), usually occurs because the capsulorhexis was too small.

2. Elschnig pearls

These (**Fig. 9.93**) are due to proliferation of lens epithelium onto the posterior capsule. They are more common in young patients and usually appear after several months or years.

3. Soemmerring ring

This has a doughnut-shaped configuration (**Fig. 9.94**) and is caused by sequestration of proliferating lens fibers in the equatorial region of the capsule.

4. Inflammatory plaques

These are associated with chronic endophthalmitis and are characterized by a localized fluffy white thickening of the capsule (**Fig. 9.95**).

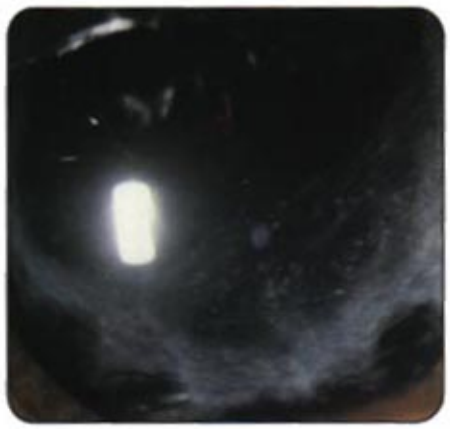

Fig. 9.91

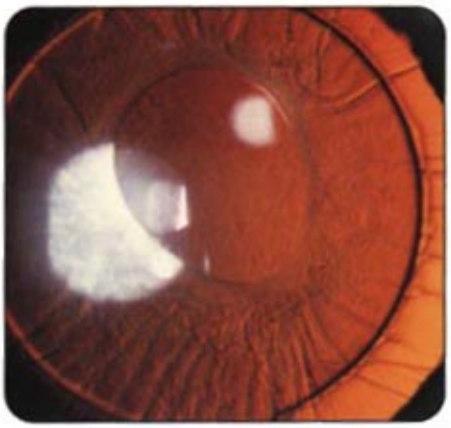

Fig. 9.92

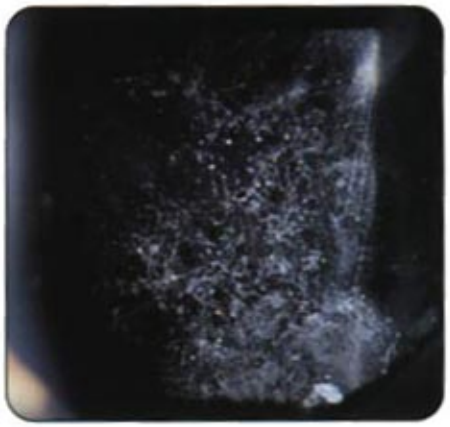

Fig. 9.93

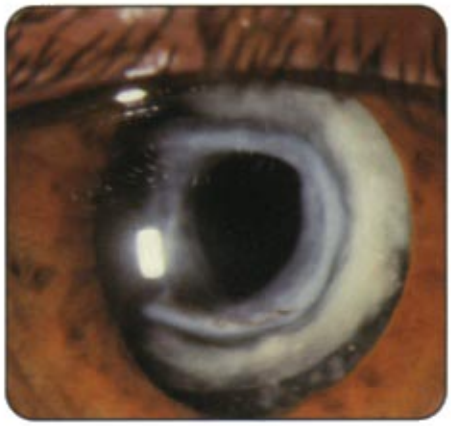

Fig. 9.94

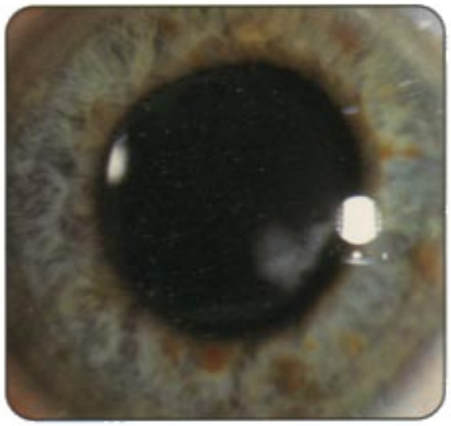

Fig. 9.95

Lenticular

1. Inflammatory precipitates
These (**Fig. 9.96**) are usually associated with postoperative uveitis or endophthalmitis.

2. Pitting
This is caused inadvertantly during YAG-laser posterior capsulotomy.

3. Glistening
This may occur on acrylic (Acrasoft) implant.

4. Pseudoexfoliative material
This may rarely be seen in eyes with pseudoexfoliation.

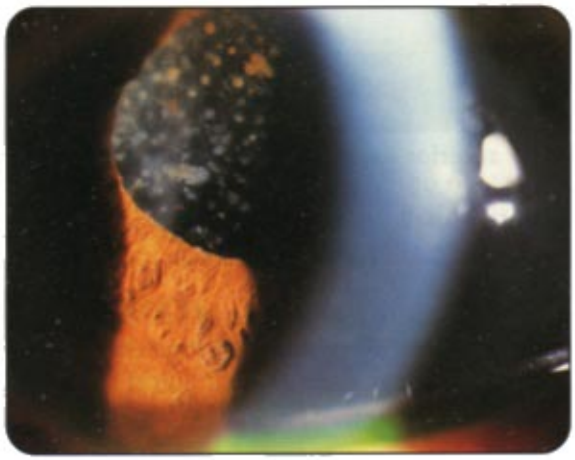

Fig. 9.96

10 The vitreous

VITREOUS CELLS 236

- Inflammatory white blood cells
- Red blood cells
- Miscellaneous cells

VITREOUS OPACITIES 239

- Common innocuous vitreous floaters
- Opacities associated with acute posterior vitreous detachment
- Inflammatory opacities
- Iridescent particles
- Hereditary vitreoretinopathies
- Miscellaneous opacities

'EMPTY' VITREOUS 245

VITREOUS CELLS

Inflammatory white blood cells

1. Iridocyclitis
Retrolental cells (**Fig. 10.1**) are seen in association with aqueous cells.

2. Intermediate uveitis
This is frequently bilateral.

Look for

- Vitreous cells (vitritis) (**Fig. 10.2**).
- Mild anterior uveitis.
- Inferior peripheral snowbanking (pars planitis).
- Vitreous cotton balls (*see* **Figs 10.19, 10.20**).
- Peripheral periphlebitis.
- Chronic cystoid macular edema.

3. Infectious endophthalmitis
This may be:
- Acute – postsurgical and post-traumatic.
- Chronic postoperative.
- Delayed-onset associated with a filtering bleb.
- Endogenous (**Fig. 10.3**).

4. Posterior uveitis
Vitreous cells are most dense at the site of the inflammatory focus (**Fig. 10.4**).

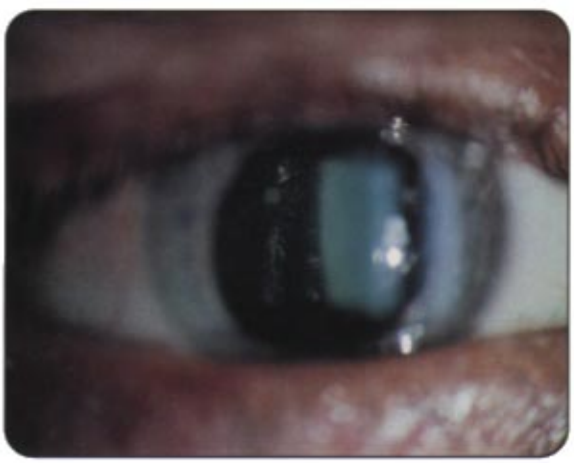

Fig. 10.1

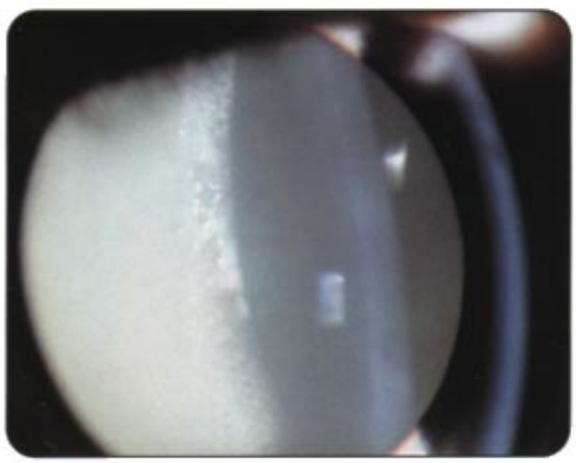

Fig. 10.2

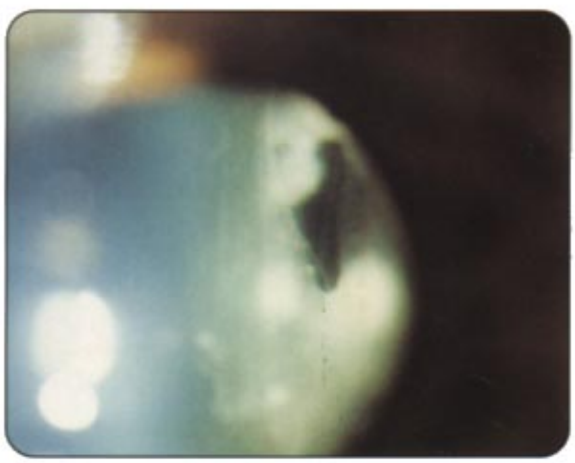

Fig. 10.3

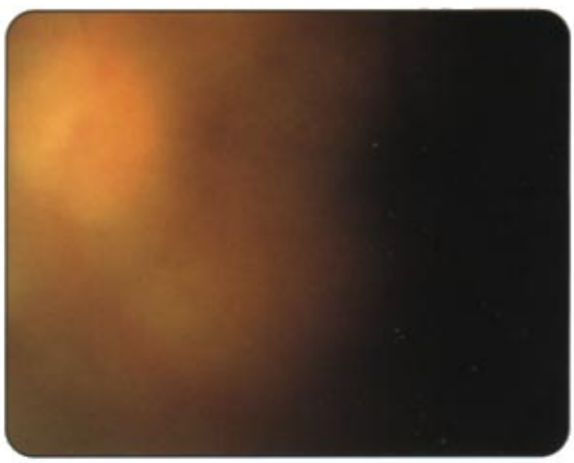

Fig. 10.4

Red blood cells

The severity of vitreous hemorrhage can vary from relatively trivial (**Fig. 10.5**) to extremely severe (**Fig. 10.6**).

1. Acute posterior vitreous detachment

This typically presents with a unilateral, sudden shower of minute red-colored or dark spots, frequently associated with photopsia.

Causes of bleeding

- Tractional retinal tear (**Fig. 10.7**) which is most frequently located in the upper temporal quadrant.
- Less frequently, avulsion of a peripheral blood vessel unassociated with a tear.

2. Proliferative retinopathies

- Diabetic (**Figs 10.8, 10.9**).
- Following retinal vein occlusion (**Fig. 10.10**).
- Sickle cell disease (**Fig. 10.11**).
- Eales disease (**Fig. 10.12**).
- Vasculitis (**Fig. 10.13**).

3. Miscellaneous retinal disorders

- Macroaneurysm.
- Telangiectasias.
- Capillary hemangioma.
- Congenital vitreoretinopathies.

4. Choroidal disease

- Choroidal neovascularization.
- Tumors (**Fig. 10.14**).

5. Trauma

- Blunt.
- Penetrating (**Fig. 10.15**).
- During surgery for retinal detachment.

6. Systemic causes

- Bleeding disorders.
- Terson syndrome (subarachnoid hemorrhage + vitreous hemorrhage).

Fig. 10.5

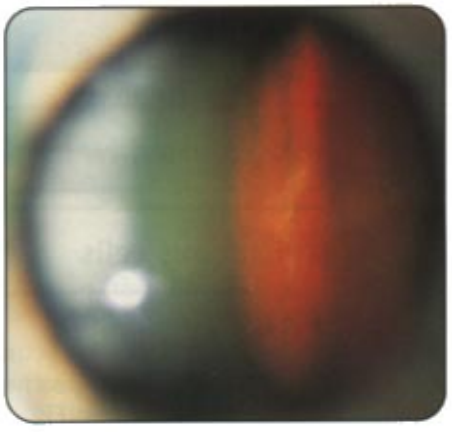

Fig. 10.6

Fig. 10.7

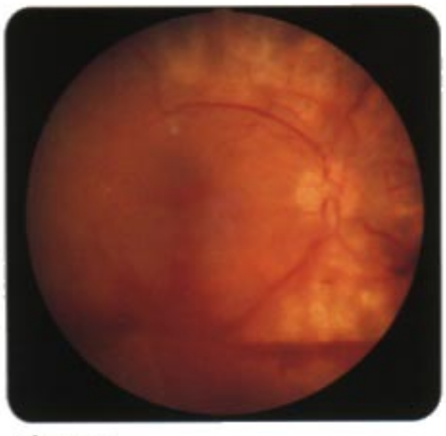

Fig. 10.8

Fig. 10.9

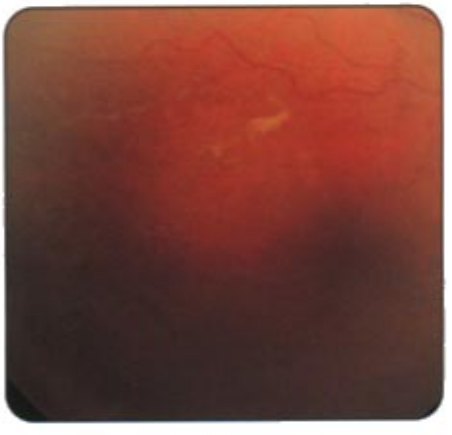

Fig. 10.10

Fig. 10.11

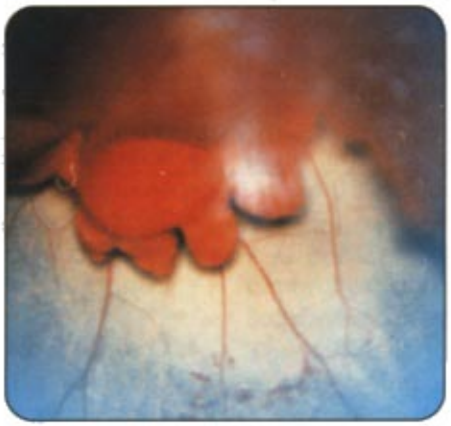

Fig. 10.12

Fig. 10.13

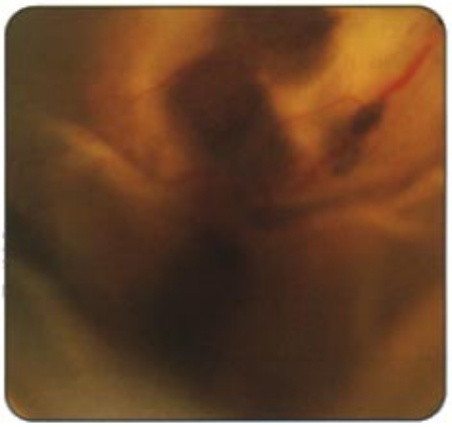

Fig. 10.14

Fig. 10.15

Miscellaneous cells

1. Pigment cells ('tobacco dust') in the anterior vitreous

These consist of macrophages containing retinal pigment epithelial cells.

Causes

- Acute retinal tear associated with posterior vitreous detachment (**Fig. 10.16**).
- Rhegmatogenous retinal detachment.
- Excessive retinal cryotherapy.
- Proliferative vitreoretinopathy.
- Trauma (**Fig. 10.17**).

2. Tumor cells

These may occur in:

- Retinoblastoma.
- Reticulum cell sarcoma.
- Leukemia.

Fig. 10.16

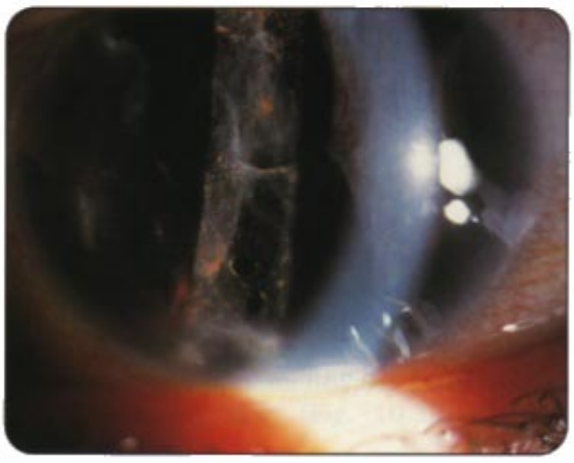

Fig. 10.17

VITREOUS OPACITIES

Common innocuous vitreous floaters

1. Muscae volitantes
These are bilateral, minute, multiple worm-like opacities best seen by the patient against a white or bright background.

2. Bilateral opacities of various sizes
These are characteristic of degeneration of the vitreous gel and are common in myopes.

Opacities associated with acute posterior vitreous detachment

1. Multiple cobwebs
These are condensations of collagen fibers within the collapsed vitreous gel.

2. Discrete ring-like opacity located on the posterior vitreous face
This represents a ring of glial tissue detached from the margin of the optic disc (Weiss ring) (**Fig. 10.18**).

Fig. 10.18

Inflammatory opacities

1. Cotton balls

Causes

- Intermediate uveitis (**Figs 10.19, 10.20**).
- Sarcoidosis – usually seen inferiorly (**Fig. 10.21**).
- Candidiasis – cotton balls are composed of micro-abscesses (**Figs 10.22, 10.23**).
- Lyme disease.
- Whipple disease.

2. Other opacities

- Stringy (**Fig. 10.24**) – caused by alterations in the vitreous gel itself.
- Coarse (**Fig. 10.25**) – caused by severe tissue destruction.

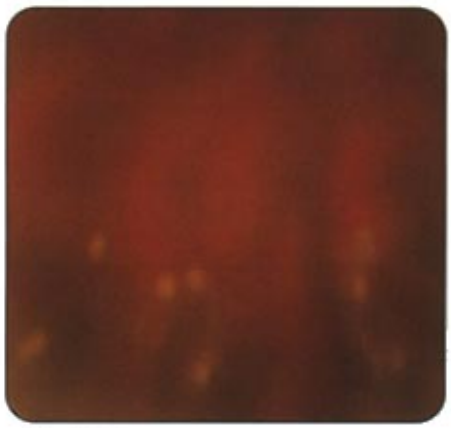

Fig. 10.19

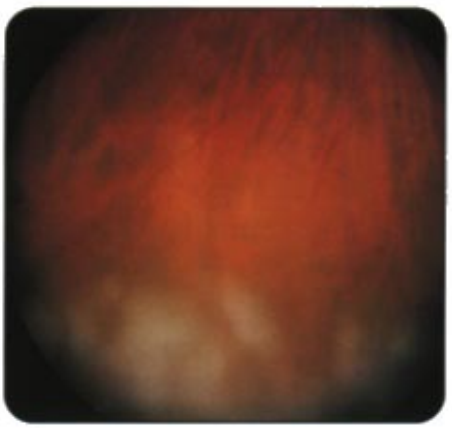

Fig. 10.20

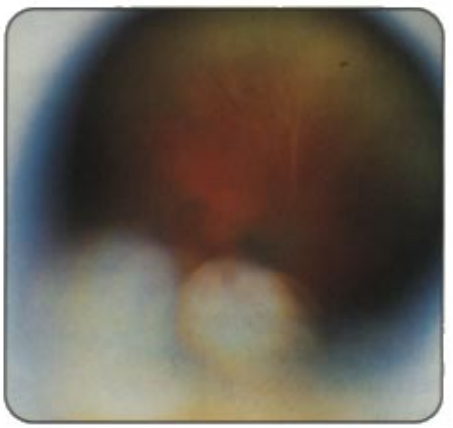

Fig. 10.21

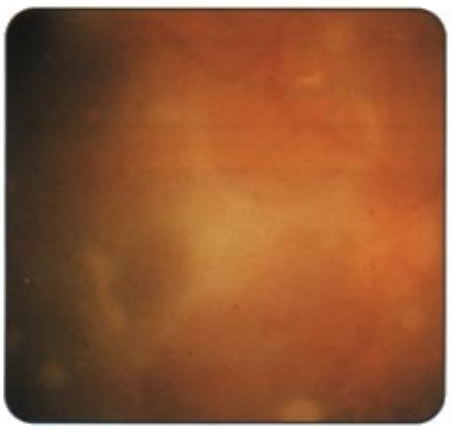

Fig. 10.22

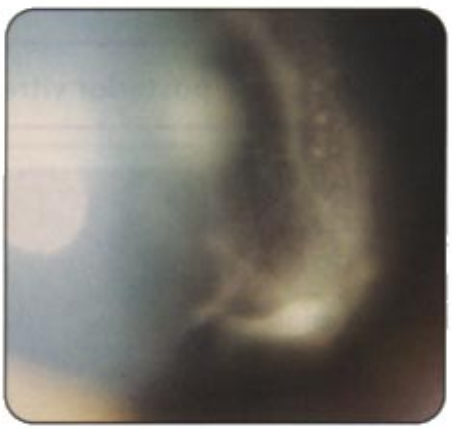

Fig. 10.23

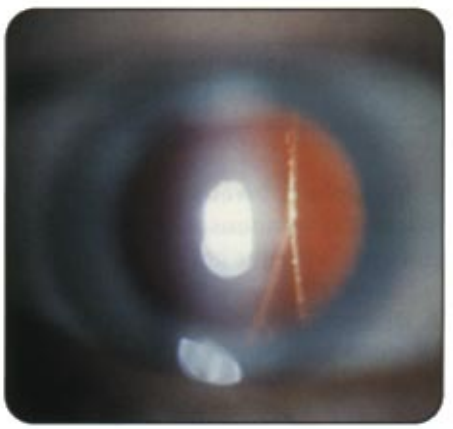

Fig. 10.24

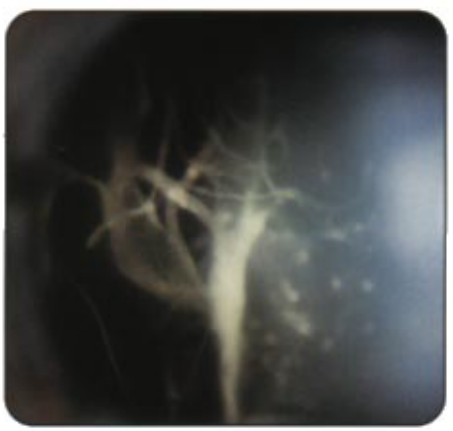

Fig. 10.25

Iridescent particles

1. Asteroid hyalosis (Benson disease)

This is a common, usually unilateral, innocuous condition which is thought to occur with increased frequency in diabetics.

Signs

- Numerous, small white opacities consisting of calcium soaps in an otherwise normal vitreous (**Figs 10.26–10.28**).

2. Synchysis scintillans

This is an uncommon, usually unilateral condition which occurs in eyes with previous large vitreous hemorrhages.

Signs

- Numerous, golden, cholesterol crystals (**Fig. 10.29**).
- In contrast to asteroid hyalosis, the opacities tend to sediment inferiorly (**Fig. 10.30**) when the eye is immobile.

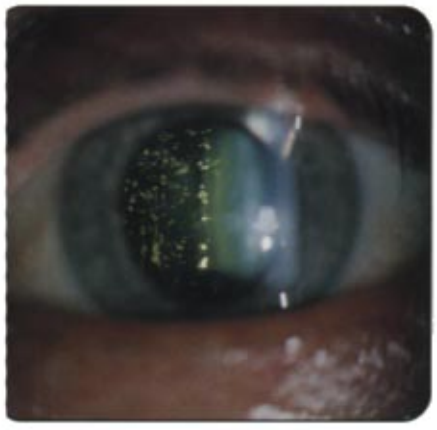

Fig. 10.26

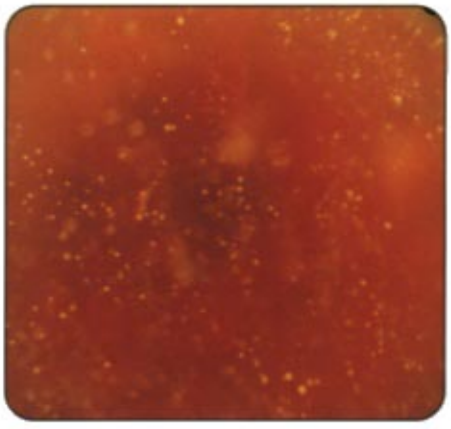

Fig. 10.27

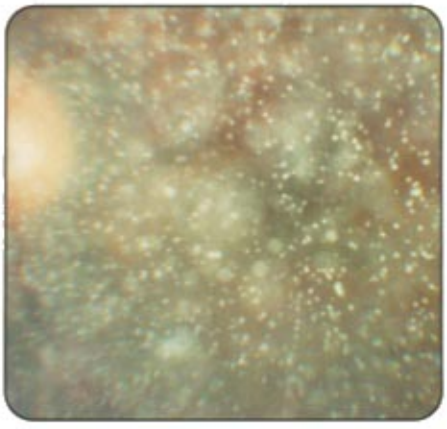

Fig. 10.28

Fig. 10.29

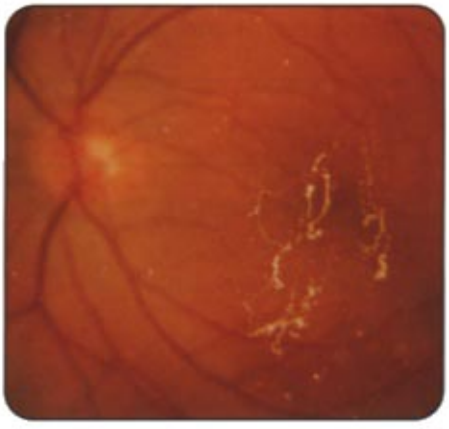

Fig. 10.30

Hereditary vitreoretinopathies

The following conditions are associated with vitreous condensations (**Fig. 10.31**).

1. Familial exudative vitreoretinopathy

This is a rare, dominantly inherited condition which is similar to retinopathy of prematurity but is not associated with prematurity.

Signs

- Dragged disc vessels (**Fig. 10.32**) and abrupt termination of retinal vessels along a scalloped edge at the equator (**Fig. 10.33**).

Complications

- Heterotopia of the macula.
- Retinal detachment.

2. Congenital (X-linked) retinoschisis

Signs

- Large defects in very thin inner leaf (**Fig. 10.34**).
- In extreme cases, the defects coalesce, leaving only retinal blood vessels floating in the vitreous, which have the appearance of vitreous veils.
- 'Bicycle-wheel-like' maculopathy is present in 50% of cases (**Fig. 10.35**).

Complications

- Vitreous hemorrhage.
- Retinal detachment.

3. Autosomal dominant vitreoretinochoroidopathy

Signs

- Vitritis.
- Peripheral retinal pigmentation and neovascularization, with a sharp demarcation between normal and abnormal retina.

Complications

- Cystoid macular edema.

4. Autosomal dominant neovascular inflammatory vitreoretinopathy

Signs

- Vitritis.
- Peripheral retinal neovascularization.
- Occasionally, disc neovascularization.

Complications

- Neovascular glaucoma.
- Vitreous hemorrhage.
- Chronic cystoid macular edema.
- Retinal detachment.

5. Erosive vitreoretinopathy

Signs

- Nyctalopia.
- Progressive retinal pigment epithelial atrophy.
- No myopia or systemic features.

Complications

- Retinal detachment.

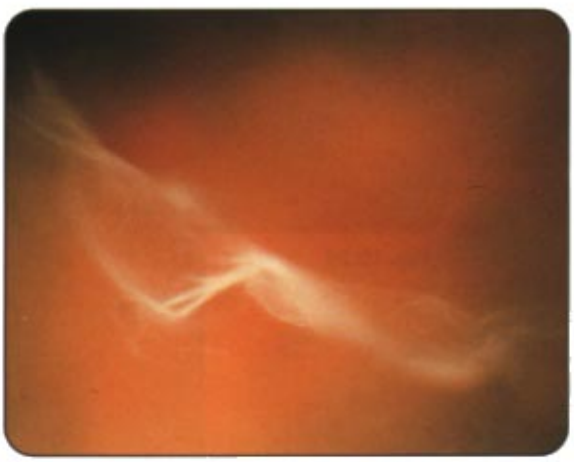

Fig. 10.31

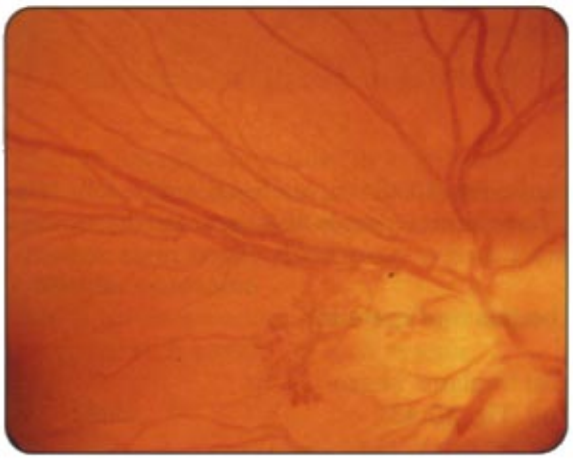

Fig. 10.32

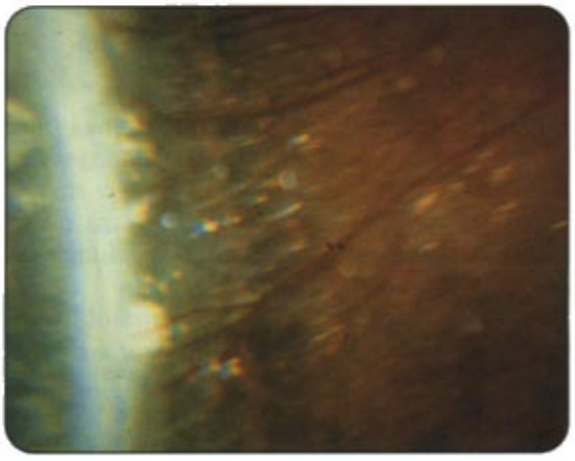

Fig. 10.33

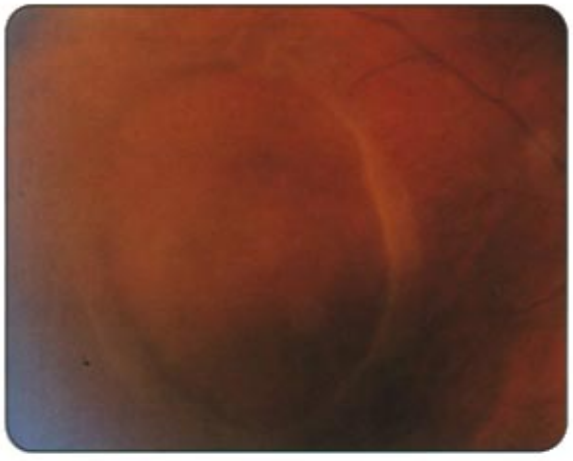

Fig. 10.34

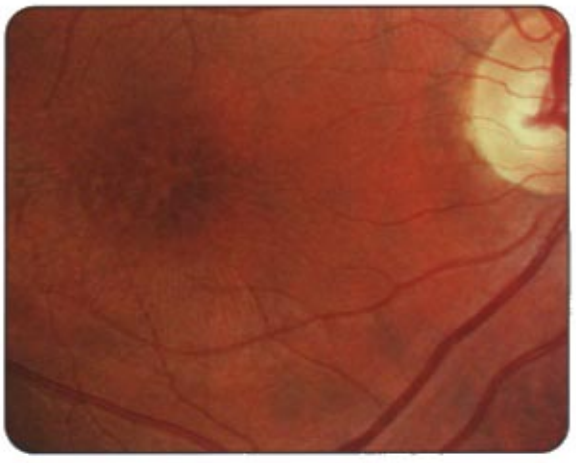

Fig. 10.35

Miscellaneous opacities

1. Retinal fragments (operculum)
This condition is shown in **Figure 10.36**.

2. Ochre membrane
This forms on the posterior hyaloid face following a vitreous hemorrhage. Biodegradation of the released hemoglobin produces pigments which stain the gel orange.

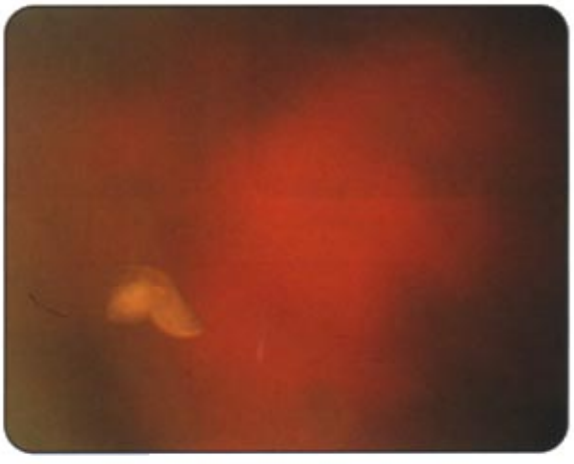

Fig. 10.36

3. Congenital remnants
- Cyst (**Fig. 10.37**).
- Persistent hyaloid artery (**Fig. 10.38**).
- Posterior persistent hyperplastic primary vitreous (**Fig. 10.39**).

4. Amyloid
Vitreous deposits occur in the dominant form of familial amyloidosis.

Signs

- Bilateral, sheet-like ('glass-wool') opacification (**Fig. 10.40**).

5. Acquired cysts
- Cysticercosis.
- Echinococcosis.

6. Lens
Lens fragments (**Fig. 10.41**) or, rarely, the entire lens (**Fig. 10.42**) may become accidentally dislodged into the vitreous during cataract surgery, particularly phacoemulsification.

7. Foreign bodies and material
- Metallic.
- Intravitreal implants of ganciclovir.
- Silicone oil (**Fig. 10.43**).
- Heavy liquids.
- Air and other gases (**Fig. 10.44**).

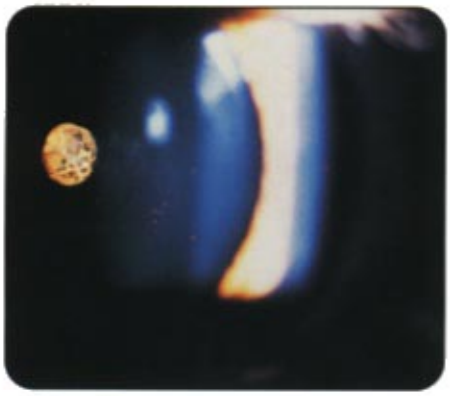

Fig. 10.37

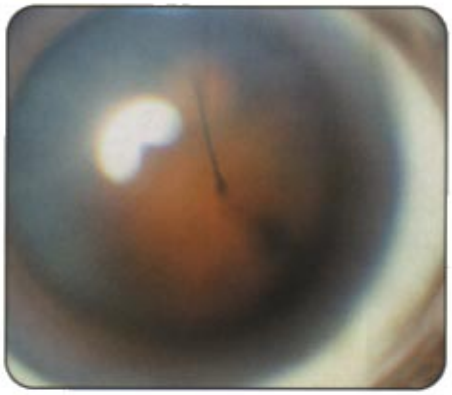

Fig. 10.38

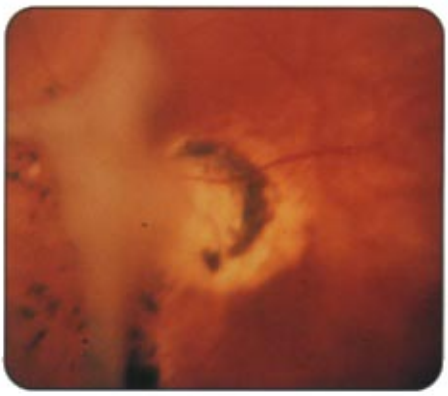

Fig. 10.39

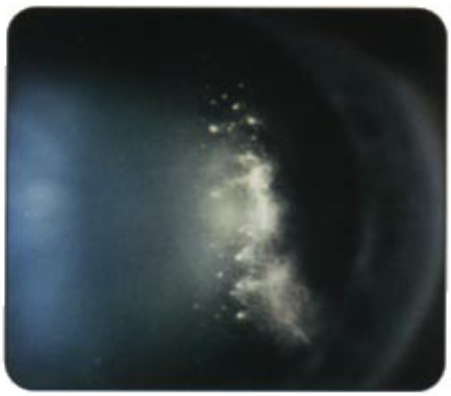

Fig. 10.40

Fig. 10.41

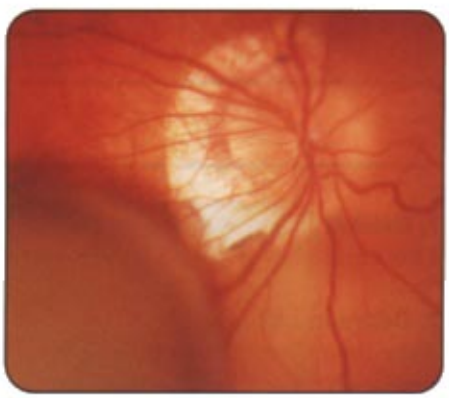

Fig. 10.42

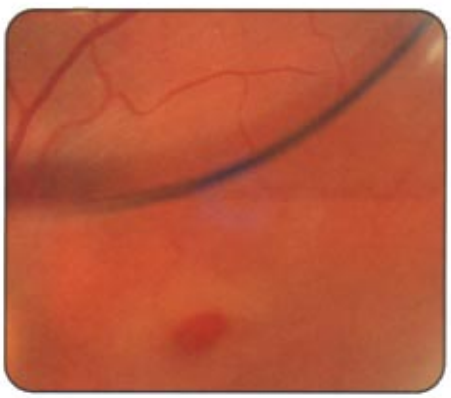

Fig. 10.43

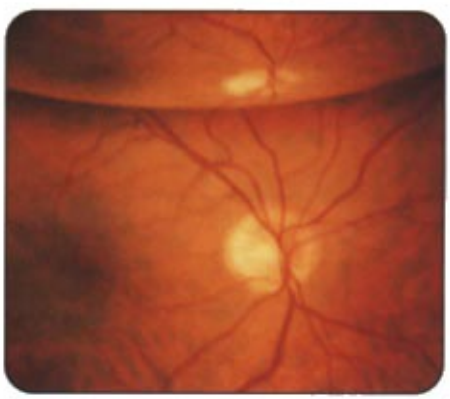

Fig. 10.44

'EMPTY' VITREOUS

Vitreous liquefaction results in an optically empty cavity except for a thin layer of cortex behind the lens and avascular white membranes adherent to the retina (**Fig. 10.45**).

1. Stickler syndrome

This is an uncommon, dominantly inherited condition associated with congenital high myopia.

Signs

- Lattice-like radial perivascular pigmentation and sclerosis (**Fig. 10.46**).

Complications

- Cataract.
- Lens subluxation.
- Glaucoma.
- Retinal detachment, which may be caused by giant retinal tears.

2. Wagner disease

In this very rare condition, retinal findings are similar to those in Stickler syndrome but without either retinal detachment or systemic features.

3. Goldmann–Favre disease

This is a very rare, recessively inherited condition associated with night blindness.

Signs

- Pigmentary chorioretinal degeneration.
- Vascular sheathing (**Fig. 10.47**).
- Bone-spicule pigmentation.

Complications

- Macular schisis causes visual loss during the first two decades of life.

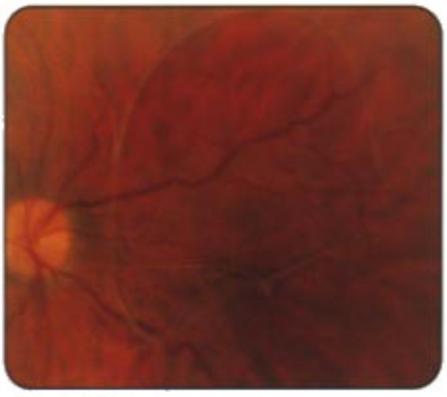

Fig. 10.45

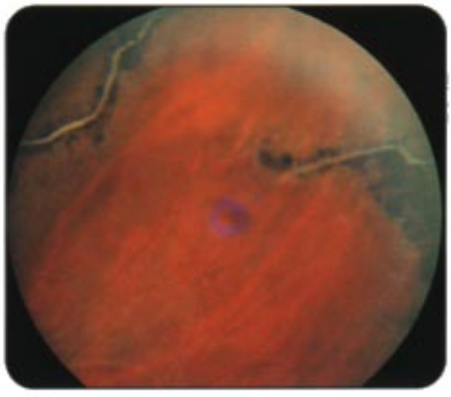

Fig. 10.46

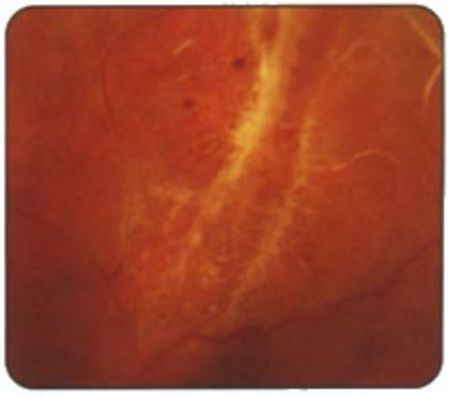

Fig. 10.47

11 The optic disc

ABNORMALITIES OF SIZE 248

Small disc
- Hypermetropia
- Tilted disc
- Optic disc hypoplasia

Large disc
- High myopia
- Congenital optic disc pit
- Optic disc coloboma
- Morning glory anomaly
- Megalopapilla

Large cup
- Physiological excavation
- Glaucomatous cupping

VASCULAR ABNORMALITIES 256

Congenital abnormalities
- Pre-papillary loop
- Bergmeister papilla
- Persistent hyaloid artery
- Posterior persistent hyperplastic primary vitreous
- Increased number of vessels crossing the disc margin

Acquired abnormalities
- Disc collaterals
- Disc new vessels
- Optico-ciliary shunts
- Disc hemorrhages
- Dragged vessels

UNILATERAL DISC SWELLING 261

Ocular causes
- Central retinal vein occlusion
- Papillophlebitis
- Longstanding ocular hypotony
- Intermediate uveitis
- Posterior scleritis
- Postcataract papillopathy

Systemic vascular causes
- Nonarteritic anterior ischemic optic neuropathy
- Arteritic anterior ischemic optic neuropathy

Optic neuritis
- Papillitis and retrobulbar neuritis
- Neuroretinitis
- Leber hereditary optic neuropathy

Compressive optic neuropathies

BILATERAL DISC ELEVATION 267

Buried optic disc drusen

Papilledema
- Early papilledema
- Established (acute) papilledema
- Chronic papilledema
- Atrophic papilledema

Malignant hypertension

Miscellaneous causes

OPTIC ATROPHY 274
- Primary optic atrophy
- Consecutive optic atrophy
- Secondary optic atrophy
- Optic atrophy with contralateral disc swelling (Foster Kennedy syndrome)
- Primary hereditary optic atrophies
- Hereditary neurological disorders with optic atrophy

LESIONS OBSCURING THE OPTIC DISC 278

Primary optic disc tumors
- Capillary angioma
- Cavernous angioma
- Melanocytoma
- Astrocytoma
- Combined hamartoma of the retina and RPE

Infiltrative lesions

JUXTAPAPILLARY LESIONS 281

Atrophy
- Chronic glaucoma
- High myopia
- Presumed ocular histoplasmosis syndrome
- Postinflammatory
- Juxtapapillary coloboma
- Serpiginous choroidopathy

Miscellaneous
- Normal variants
- Opaque nerve fibers
- Juxtapapillary choroidal neovascularization
- Juxtapapillary angioid streaks
- Choroidal osteoma

ABNORMALITIES OF SIZE

Small disc

Hypermetropia

In hypermetropia, the reduced axial length is associated with a smaller than normal scleral canal. Because the number of nerve fibers passing through the canal is not reduced, the fibers are 'crowded'. This crowding is particularly marked in nanophthalmos, in which the entire globe is small and there is a very high degree of hypermetropia (*see* Chapter 5).

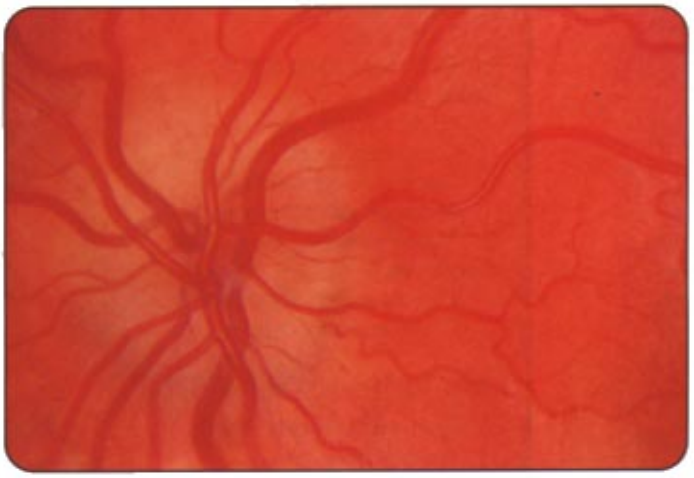

Fig. 11.1

Signs

- Slightly elevated pink disc with a small or absent cup (**Fig. 11.1**).
- Crowding of central retinal vessels.
- Absence of capillary dilatation on the disc.
- Chorioretinal folds (rare).
- Anterior chamber may be shallow.

Differential diagnosis

- Early papilledema (*see* **Fig. 11.86a and b**).
- Buried disc drusen (*see* **Fig. 11.81a and b**).

Tilted disc

Tilted disc is a common, usually bilateral, congenital condition that occurs with increased frequency in myopes. It is caused by an oblique entry of the optic nerve into the globe. Temporal visual field defects are often present, but visual acuity is usually normal.

Signs

- Small, oval or D-shaped disc in which the axis is most frequently directed obliquely (**Figs 11.2, 11.3**) but may be horizontal (**Fig. 11.4**) or nearly vertical.
- Hypopigmentation of the inferonasal fundus (**Figs 11.5, 11.6a and b**).
- Peripapillary chorioretinal atrophy (**Figs 11.3, 11.4**).
- Situs inversus, in which the temporal vessels deviate nasally before turning temporally (**Figs 11.3, 11.6a and b**).
- Variable myopic changes are common (**Fig. 11.7a and b**).

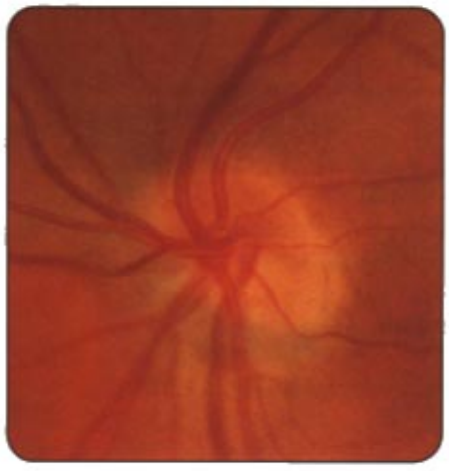

Fig. 11.2

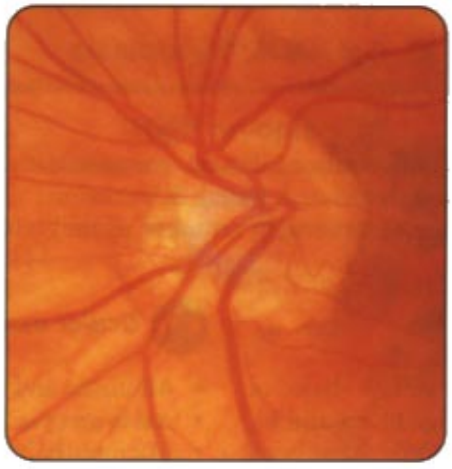

Fig. 11.3

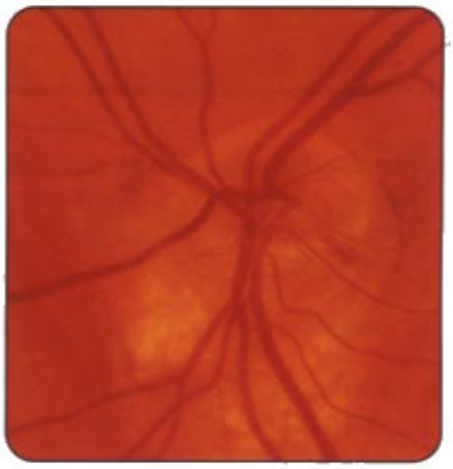

Fig. 11.4

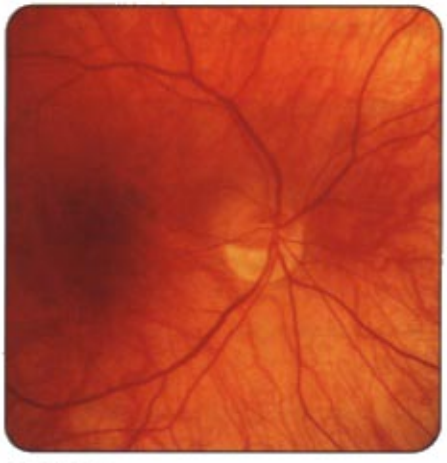

Fig. 11.5

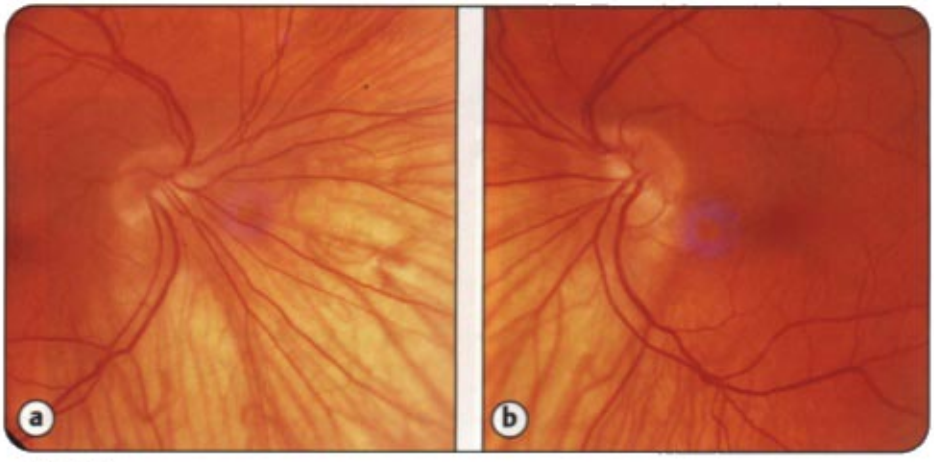

Fig. 11.6

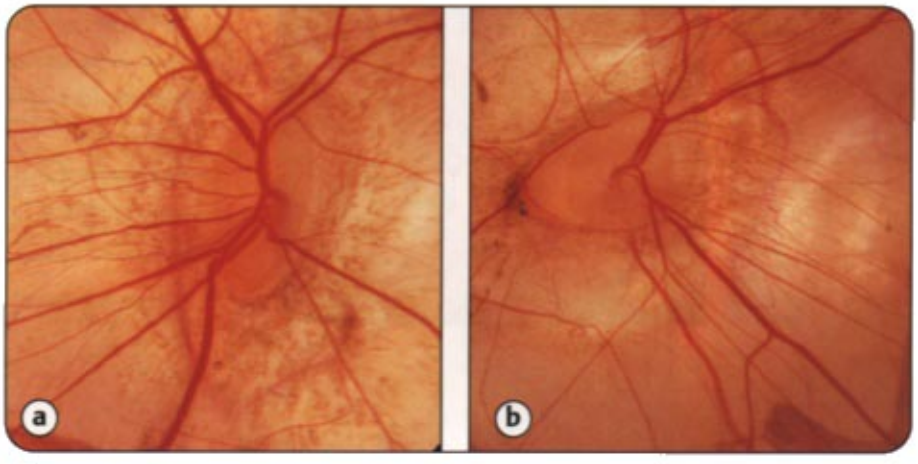

Fig. 11.7

Optic disc hypoplasia

Optic disc hypoplasia is an uncommon, unilateral or bilateral, congenital condition. The severity of hypoplasia and associated visual dysfunction shows a wide spectrum. Optic disc aplasia is extremely rare and usually unilateral.

Signs

- Small optic disc surrounded by a yellow halo of hypopigmentation (double-ring sign; **Fig. 11.8a and b**). The outer ring corresponds to what would have been the margin of a normal disc.
- Despite the small disc, the blood vessels are of normal caliber (**Fig. 11.9a and b**) although they may be tortuous.
- In some cases, only a part of the disc is hypoplastic.

Ocular associations

- Albinism and foveal hypoplasia (**Fig. 11.10**).
- Axial myopia.
- Microphthalmos.
- Aniridia.
- Persistent hyaloid artery.
- Strabismus.
- Nystagmus (in severe, bilateral cases).

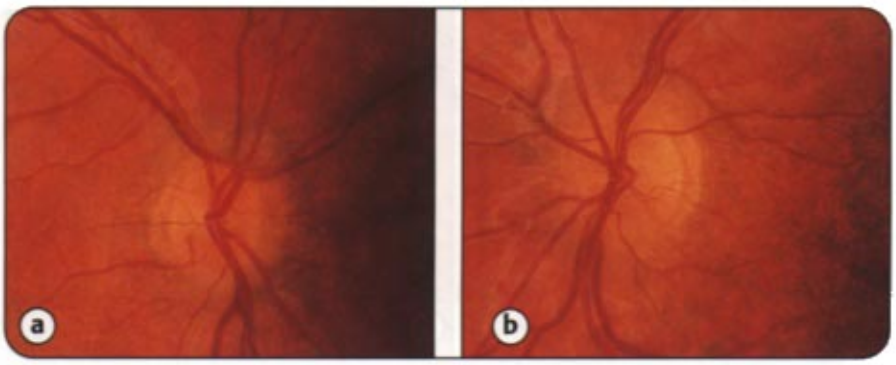

Fig. 11.8

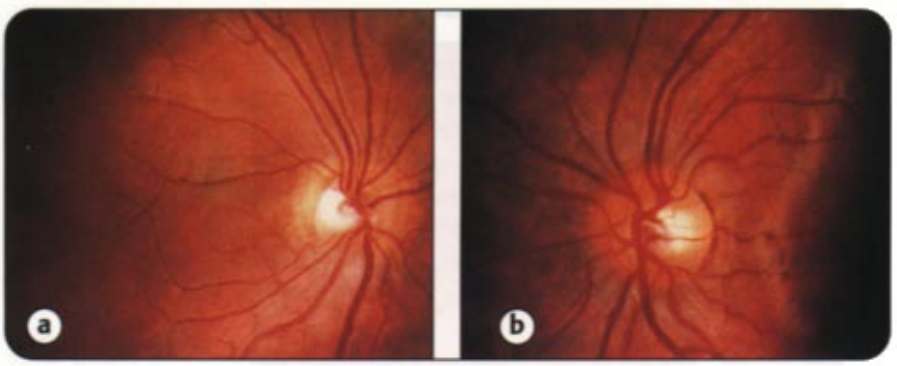

Fig. 11.9

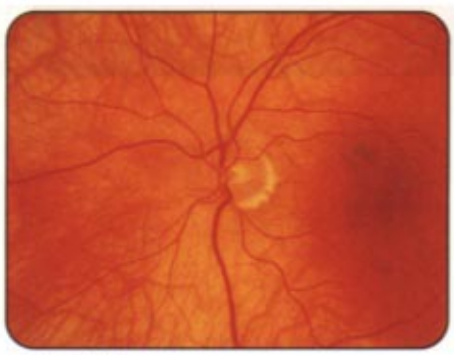

Fig. 11.10

Table 11.1 Systemic associations of optic disc hypoplasia

Maternal
• drug ingestion during pregnancy (e.g. phenytoin, quinine, LSD, steroids)
• intrauterine infections
• diabetes
Neurological
• isolated pituitary dysfunction
• partial or complete absence of septum pellucidum
• De Morsier syndrome
• basal encephalocele
• posterior fossa cyst
• anencephaly
• hydranencephaly
• Apert syndrome

Large disc

High myopia

High myopia is associated with a long axial length and a correspondingly larger than normal scleral canal (**Fig. 11.11**).

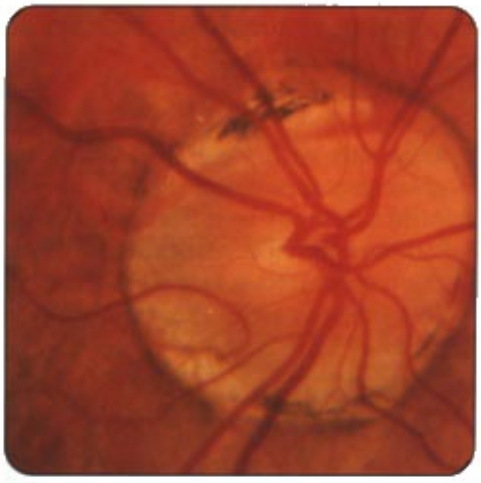

Fig. 11.11

Congenital optic disc pit

Congenital optic disc pit is an uncommon, isolated, usually unilateral condition that is associated with mild enlargement of the optic nerve head.

Signs

- Round or oval depression of variable size, most frequently involving the temporal margin of the disc (**Figs 11.12, 11.13**). A central pit is much less common.

Ocular associations

- Between 30% and 50% of eyes with noncentrally located optic disc pits eventually develop intraretinal fluid (retinoschisis), followed by detachment of the macula (**Fig. 11.14**). Eyes with centrally located pits are not at risk.

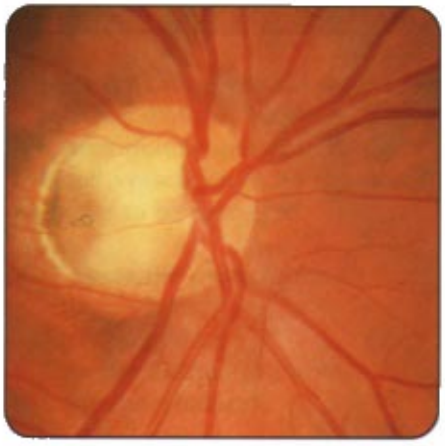

Fig. 11.12

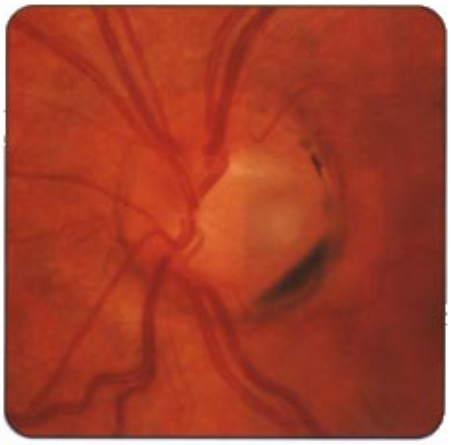

Fig. 11.13

Fig. 11.14

Optic disc coloboma

Optic disc coloboma is an uncommon, unilateral or bilateral, congenital condition caused by failure of fusion of the fetal fissure. Bilateral cases may be inherited in an autosomal dominant manner.

Signs

- Large disc with a partial inferior excavation that has a glistening appearance (**Fig. 11.15**).
- Mild cases may be mistaken for glaucomatous cupping, but the splayed appearance of the central blood through the colobomatous area gives the clue.
- In some cases the excavation can be very large (**Figs 11.16, 11.17**).

Ocular associations

- Other colobomas that may involve the lens, ciliary body, and choroid (**Figs 11.18, 11.19**).
- Serous macular detachment may occur in bilateral cases.
- Microphthalmos with cyst.
- Persistent hyaloid artery.
- Retinal dysplasia.

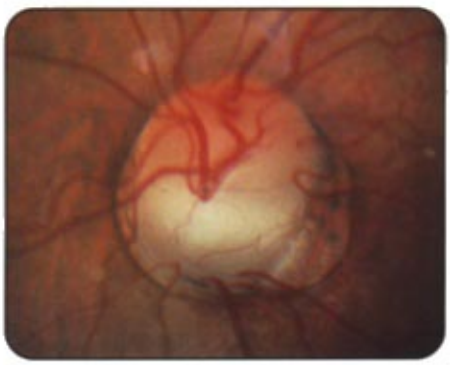

Fig. 11.15

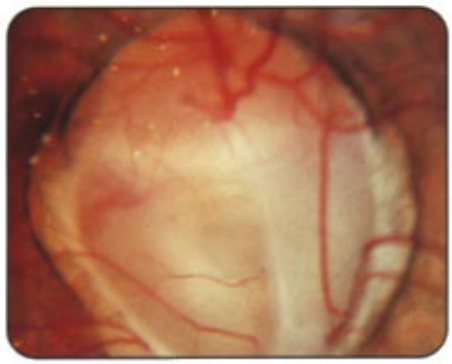

Fig. 11.16

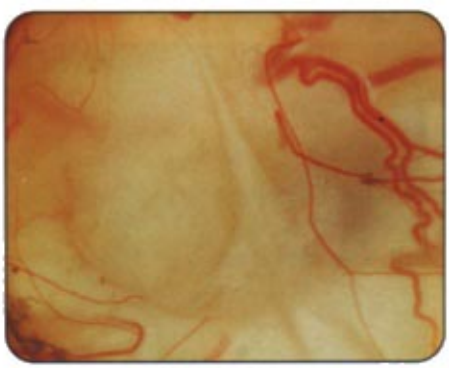

Fig. 11.17

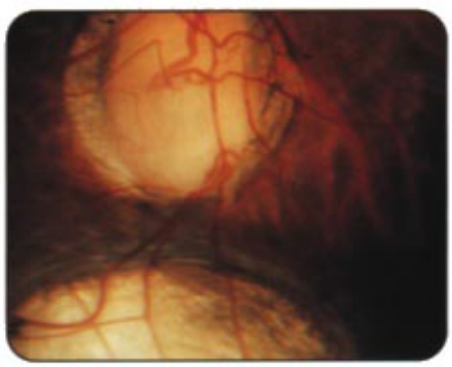

Fig. 11.18

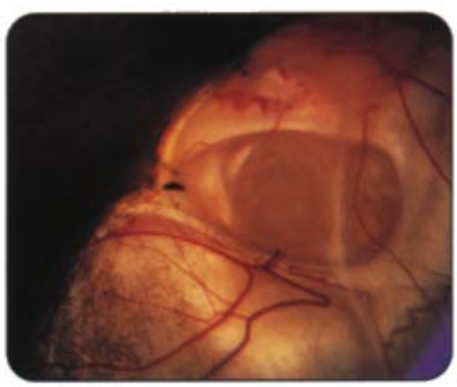

Fig. 11.19

Table 11.2 Systemic associations of optic disc coloboma

Chromosomal disorders
• Patau syndrome (trisomy 13)
• Edward syndrome (trisomy 18)
• Cat-eye syndrome (trisomy 22)
Brain malformations
• anencephaly
• agenesis of corpus callosum
• basal encephalocele
Other syndromes
• CHARGE
• Meckel–Gruber
• Golz
• Lenz microphthalmos
• Aicardi
• Hallerman–Streiff–François
• clefting
• Goldenhar

Morning glory anomaly

Morning glory anomaly is a rare, usually unilateral, congenital condition that is typically associated with poor visual acuity.

Signs

- Large disc with a core of white tissue occupying the central area (**Figs 11.20, 11.21**).
- Blood vessels emerge from the edge of the disc in a spoke-like manner (**Fig. 11.22**).
- The lesion itself is surrounded by an elevated annulus of chorioretinal pigmentary disturbance.

Ocular associations

- Aniridia.
- Cataract.
- Lens coloboma.
- Persistent hyperplastic primary vitreous.
- Foveal hypoplasia.
- Serous macular detachment.

Uncommon systemic associations

- Harelip and cleft palate.
- Cranial vault anomalies.
- Basal encephalocele.
- Absent corpus callosum.

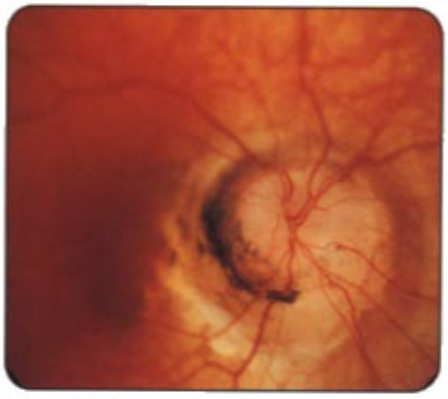

Fig. 11.20

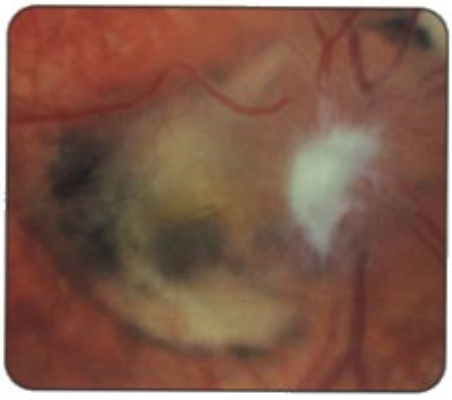

Fig. 11.21

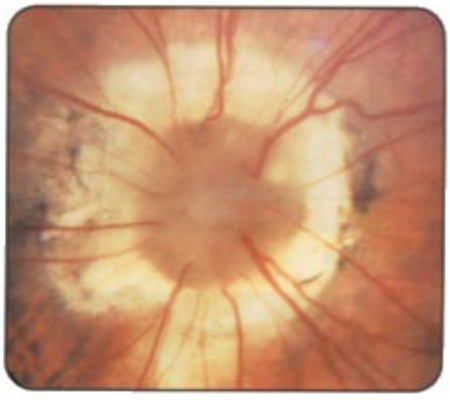

Fig. 11.22

Megalopapilla

Megalopapilla is a very rare, usually unilateral, congenital condition.

Signs

- Horizontal and vertical disc diameters of 2.1 mm or more (**Fig. 11.23**).
- Reduced distance between the temporal edge of the disc and the foveola, by approximately one disc diameter.
- Blood vessels crossing the disc have normal caliber although may falsely appear to be narrowed.

Systemic associations

- Cleft palate.
- Treacher Collins syndrome.
- Basal encephalocele.

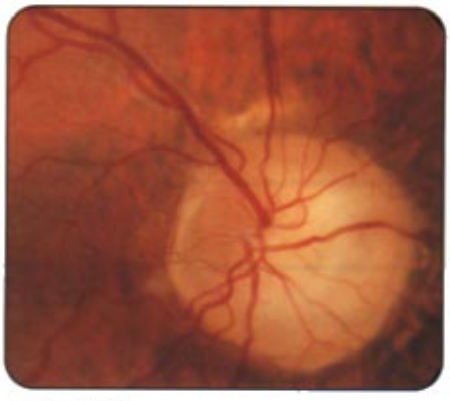

Fig. 11.23

Large cup

Physiological excavation

The three appearances of a normal cup are:
- Small (dimple) cup (**Fig. 11.24**).
- Punched-out cup (**Fig. 11.25**).
- Cup with a sloping temporal wall (**Fig. 11.26**).

Most normal cups have a cup:disc ratio of 0.3 or less. A cup:disc ratio greater than 0.7 is present in about 2% of the population. Blacks tend to have a higher ratio than whites because they have larger scleral canals.

Signs

- Bilateral and symmetrically large cups with a pink rim and absence of notching (**Fig. 11.27a and b**).
- The striations of the peripapillary nerve fiber layer can be seen up to the disc margin.

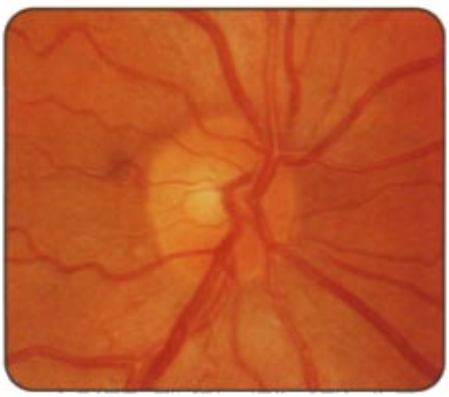

Fig. 11.24

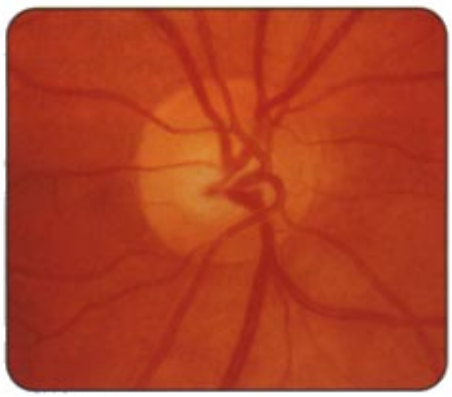

Fig. 11.25

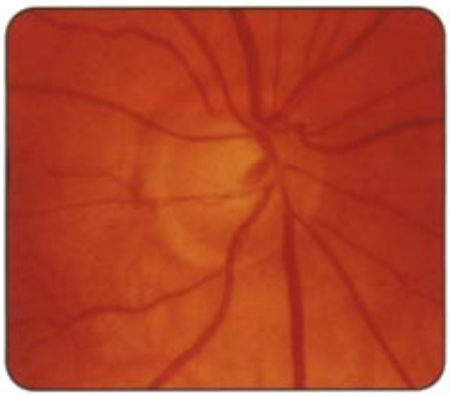

Fig. 11.26

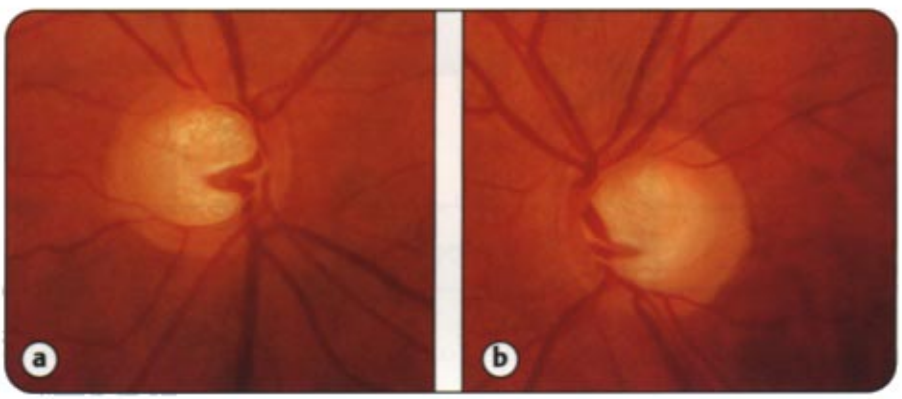

Fig. 11.27

Glaucomatous cupping

Signs

- Asymmetrically large cups in which the difference in cup:disc ratios between the eyes is 0.2 or more (**Fig. 11.28a and b**) is suggestive of glaucoma.
- Bilateral and symmetric glaucomatous cupping (**Fig. 11.29a and b**), which may be difficult to distinguish from physiological excavation, although in the former there is loss of the peripapilllary nerve fiber layer striations.
- Vertical expansion of the cup (**Fig. 11.30**).
- Splinter-shaped disc hemorrhages (**Fig. 11.31**).
- Notching of the inferior (**Fig. 11.32**) or superior neuroretinal rim.

- Notching of both the superior and inferior rims and bayonetting of the blood vessels as they emerge from the cupped disc onto the retina (**Fig. 11.33**).
- Increasing disc pallor, with nasal displacement of the blood vessels emerging from the disc (**Fig. 11.34**).
- A white and deeply excavated disc (**Fig. 11.35**).
- Peripapillary atrophy (**Figs 11.30, 11.33**).

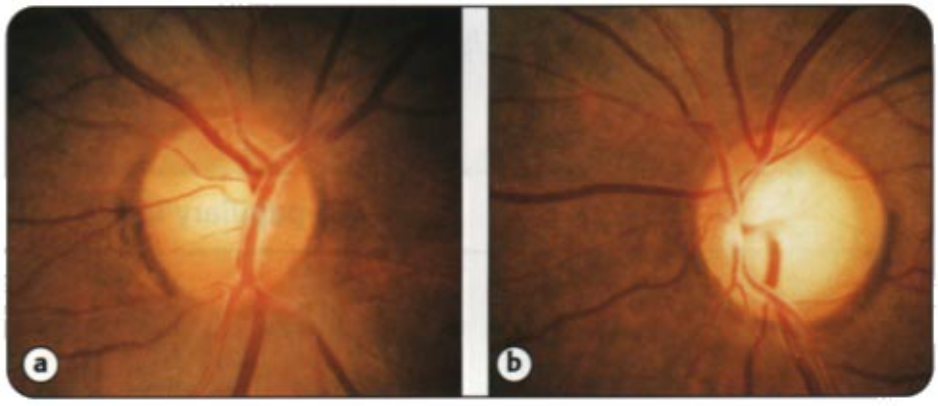

Fig. 11.28

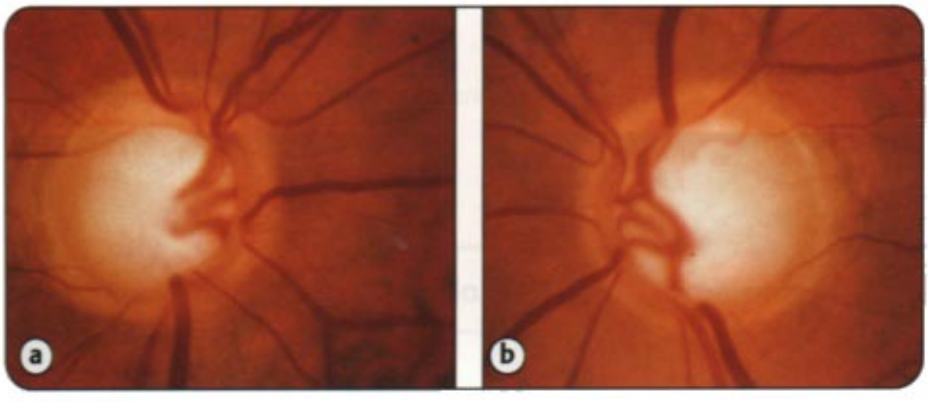

Fig. 11.29

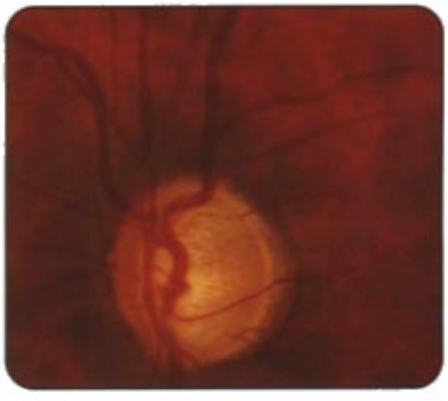
Fig. 11.30

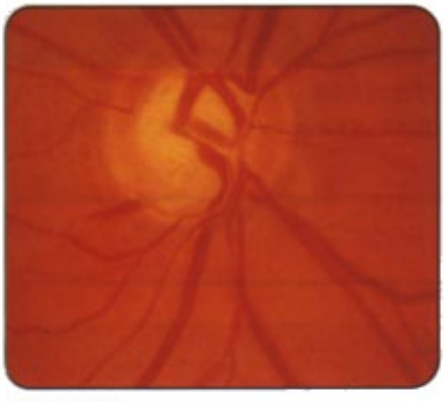
Fig. 11.31

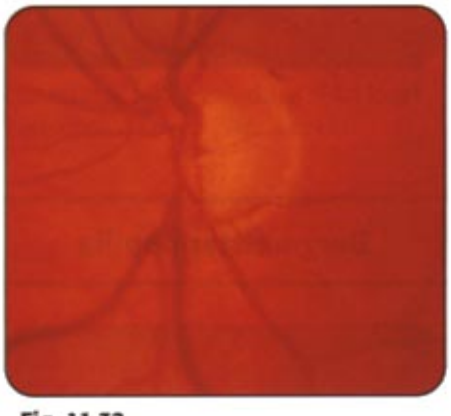
Fig. 11.32

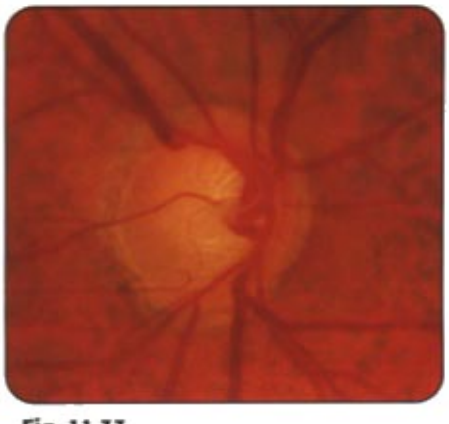
Fig. 11.33

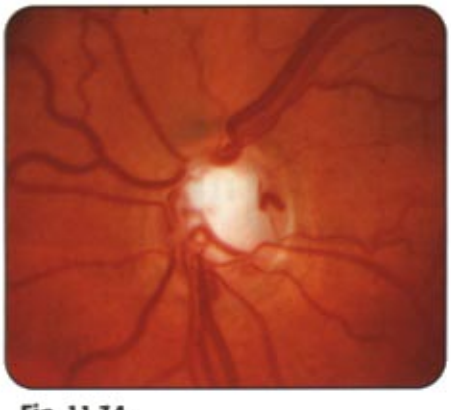
Fig. 11.34

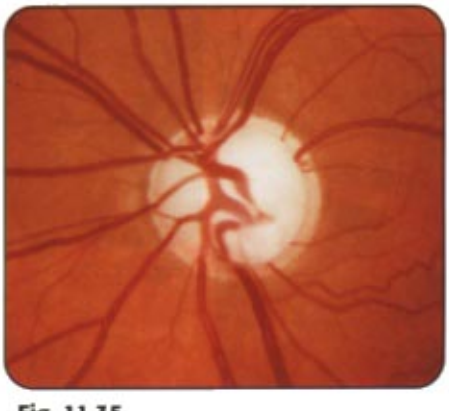
Fig. 11.35

VASCULAR ABNORMALITIES

Congenital abnormalities

Pre-papillary loop

Pre-papillary loop is an uncommon, usually unilateral condition.

Signs

- A vascular loop, which is arterial in 95% of cases, extends from the disc, into the vitreous cavity, and then back (**Figs 11.36–11.38**).

Complications

- Obstruction in the distribution of the retinal artery supplying the loop occurs in about 10% of eyes.
- Occasional vitreous hemorrhage.

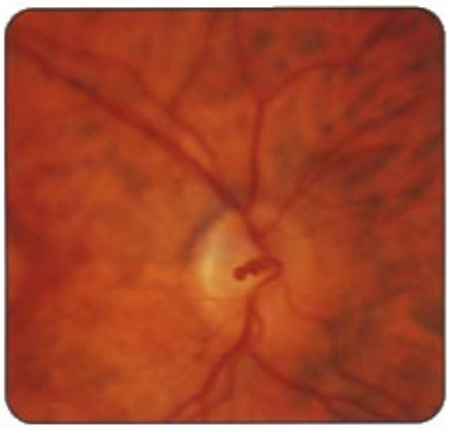

Fig. 11.36

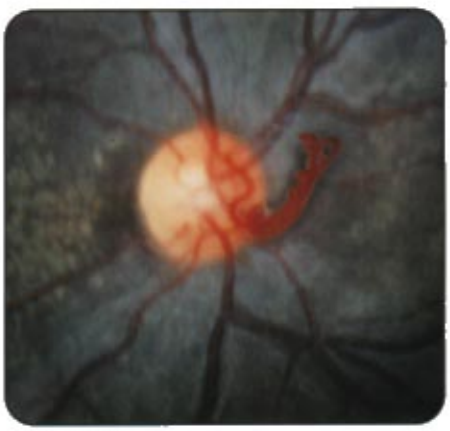

Fig. 11.37

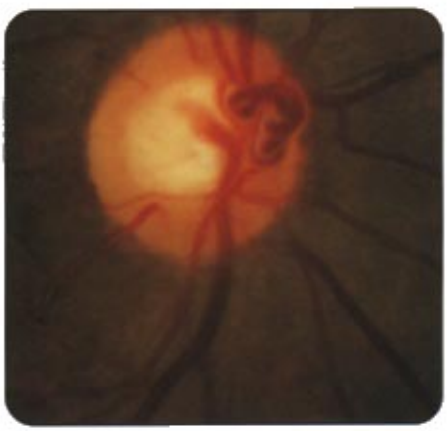

Fig. 11.38

Bergmeister papilla

Bergmeister papilla is an uncommon, unilateral condition that is derived from avascular remnants of the hyaloid system.

Signs

- Raised glial tissue on the disc (**Figs 11.39, 11.40**).

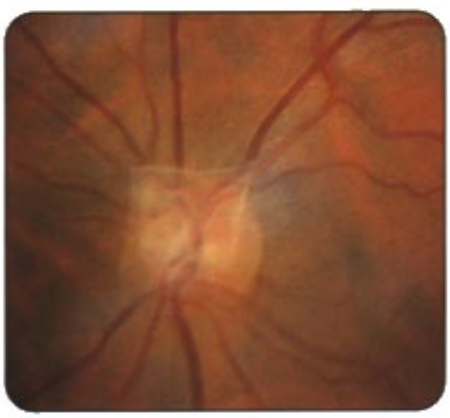

Fig. 11.39

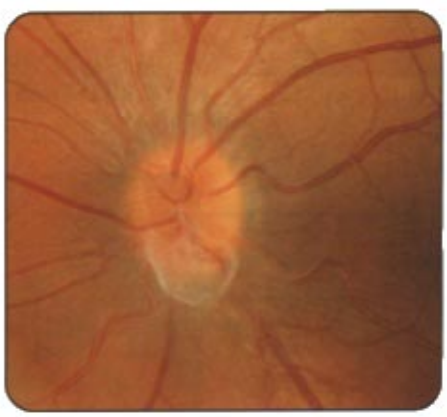

Fig. 11.40

Persistent hyaloid artery

Persistent hyaloid artery is a unilateral condition that is seen in 95% of premature infants but rarely in the adult population.

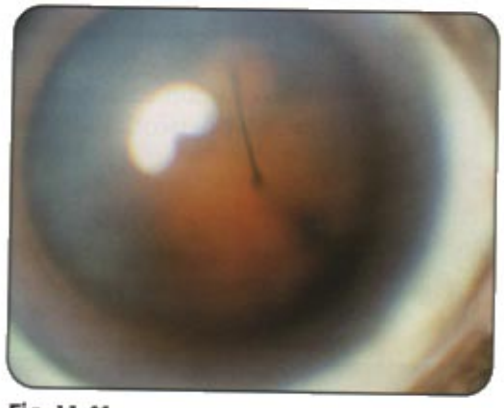

Fig. 11.41

Signs

- Glial remnants extending from the optic disc to the lens (Cloquet canal).
- The artery may contain blood at its point of attachment to the posterior capsule (Mittendorf spot; **Fig. 11.41**).
- Vitreous hemorrhage is an occasional complication.

Ocular associations

- Persistent hyperplastic primary vitreous.
- Posterior vitreous cyst.
- Optic disc coloboma.
- Optic disc hypoplasia.

Posterior persistent hyperplastic primary vitreous

Posterior persistent hyperplastic primary vitreous is a very rare, unilateral abnormality.

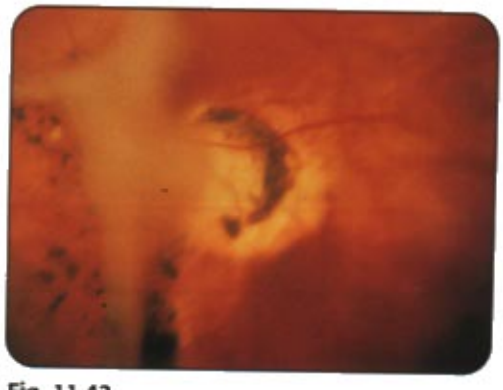

Fig. 11.42

Signs

- Dense white membrane or a prominent retinal fold extending from the optic disc to the peripheral retina or retrolental space (**Fig. 11.42**).

Increased number of vessels crossing the disc margin

Associations

- Optic disc drusen in which the vessels may also show abnormal branching patterns (**Fig. 11.43**).
- Morning glory anomaly (*see* **Fig. 11.22**).
- Down syndrome (trisomy 21).

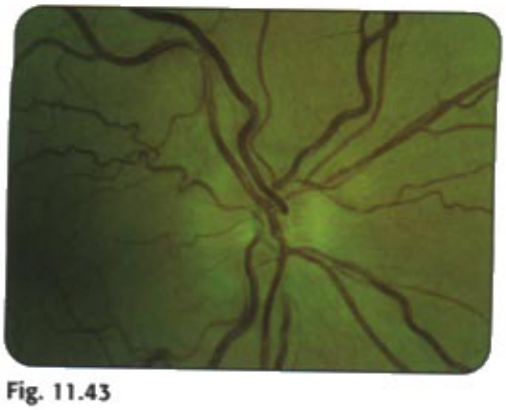

Fig. 11.43

Acquired abnormalities

Disc collaterals

Disc collaterals are common veno–venular shunts that develop within the framework of the existing vascular network to bypass an area of closure.

Signs

- Distended flat vessels that start and end on the disc (**Figs 11.44, 11.45**).

Causes

- Central retinal branch vein occlusion is the most common cause.

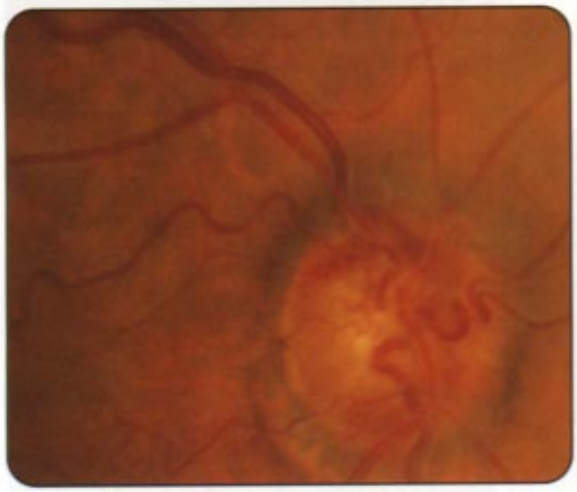

Fig. 11.44

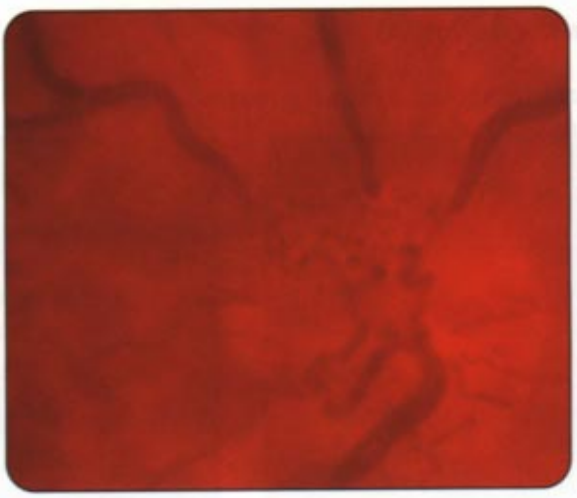

Fig. 11.45

Disc new vessels

New disc vessels are very common and may be unilateral or bilateral depending on the cause. They are usually associated with areas of retinal capillary closure.

Signs

- Fine, lace-like vessels that may be flat (**Fig. 11.46**) or elevated.
- Vessels may extend onto the adjacent retina (**Fig. 11.47**).
- Variable amounts of gliosis may be present (**Fig. 11.48**).

Causes

- Proliferative diabetic retinopathy.
- Retinal vein occlusion.
- Central retinal vasculitis.
- Old retinal artery occlusion (uncommon).
- Ocular ischemic syndrome – occlusive carotid artery disease and rarely in Takayasu disease.
- Carotid–cavernous fistula.
- Radiation retinopathy.

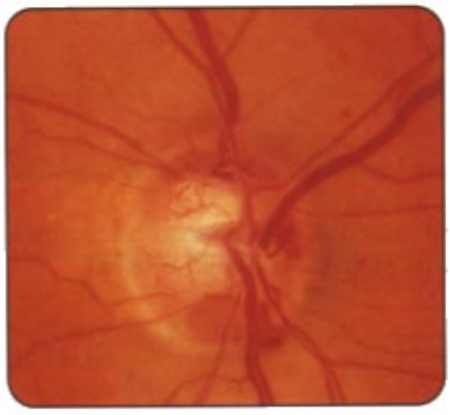

Fig. 11.46

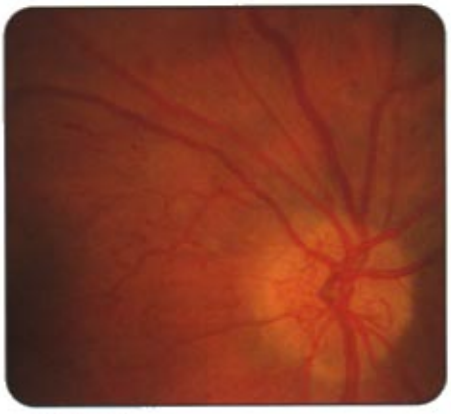

Fig. 11.47

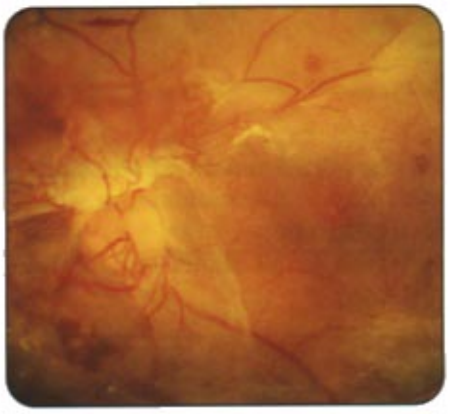

Fig. 11.48

Optico-ciliary shunts

Optico-ciliary shunts are rare, usually unilateral anastamoses between the retinal and choroidal circulations.

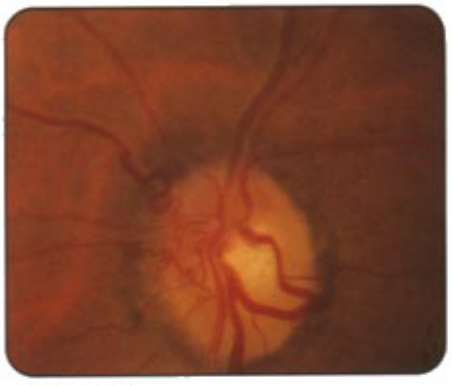

Fig. 11.49

Signs

- Vessels hook from the center of the cup to the retina–disc junction (**Fig. 11.49**).

Causes

- Optic nerve sheath meningioma (in 20–30% of cases).
- Optic nerve glioma (uncommon).
- Chronic papilledema.

Disc hemorrhages

Causes

- Acute papilledema.
- Anterior ischemic optic neuropathy.
- Papillitis.
- Diabetic papillopathy.
- Chronic glaucoma (*see* **Fig. 11.31**).
- Infiltrative optic neuropathy.
- After optic nerve sheath decompression.
- In association with acute posterior vitreous detachment.
- Optic disc drusen.

Dragged vessels

Figures 11.50 and 11.51 show dragged vessels associated with toxocaral granuloma, in which the dragging may be in any direction.

Figure 11.52 shows temporal dragging, which occurs in cicatricial retinopathy of prematurity and familial exudative vitreoretinopathy.

Figure 11.53 shows dragging in combined hamartoma of the retina and retinal pigment epithelium (RPE).

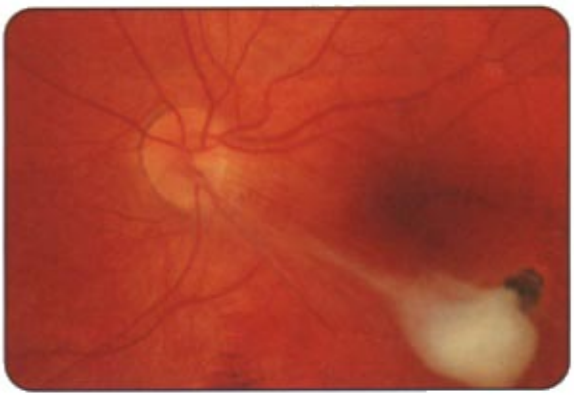

Fig. 11.50

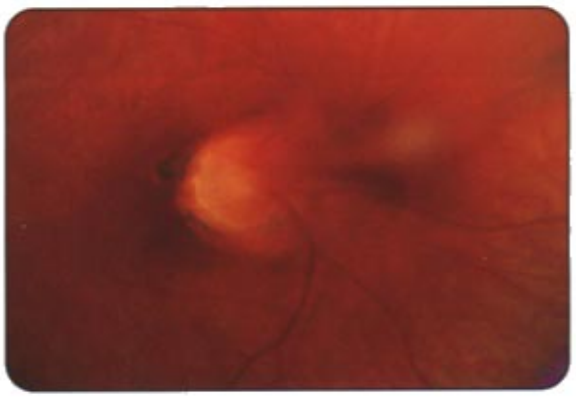

Fig. 11.51

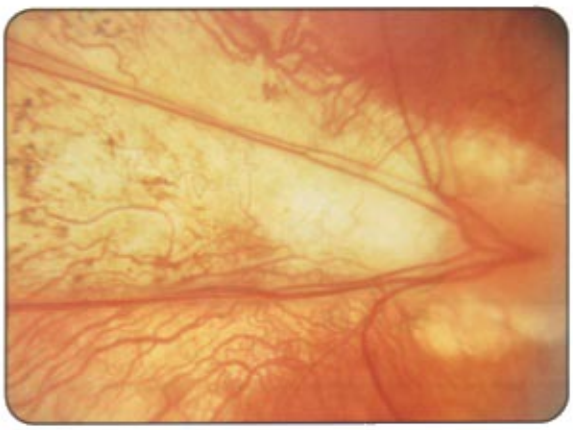

Fig. 11.52

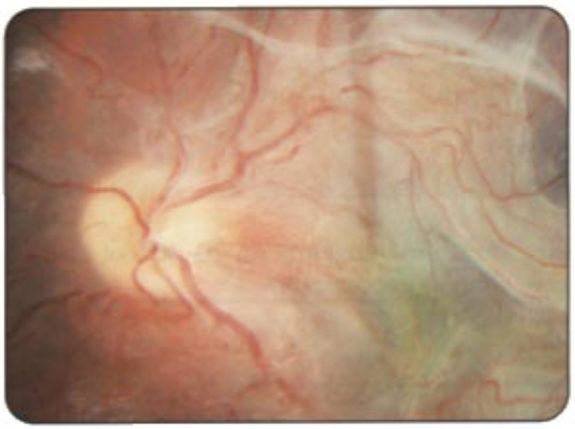

Fig. 11.53

Table 11.3 Causes of dragged vessels

- proliferative diabetic retinopathy
- proliferative sickle-cell retinopathy
- retinopathy of prematurity
- familial exudative vitreoretinopathy
- peripheral toxocaral granuloma
- incontinentia pigmenti (Bloch–Sulzberger syndrome)
- combined hamartoma of the retina and RPE

UNILATERAL DISC SWELLING

Ocular causes

Central retinal vein occlusion

Central retinal vein occlusion is a common, usually unilateral condition that typically causes moderate-to-severe visual loss in an elderly individual.

Signs

- Disc swelling, with hemorrhages on the disc and the surrounding retina (**Figs 11.54–11.56**).
  Venous engorgement and widespread retinal hemorrhages (**Figs 11.57, 11.58**).
  Cotton-wool spots may be present (**Fig. 11.59**).

Fig. 11.54

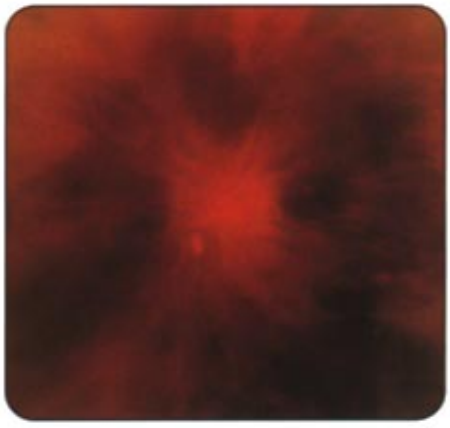

Fig. 11.55

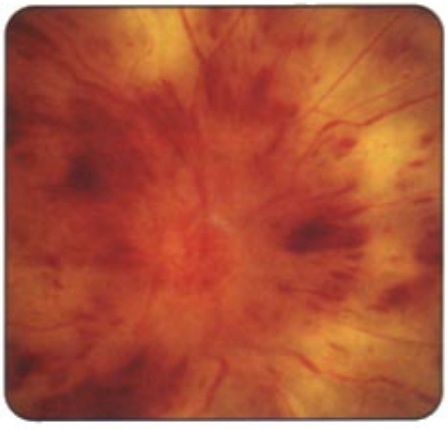

Fig. 11.56

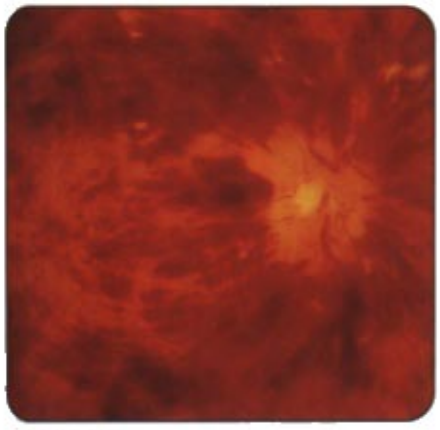

Fig. 11.57

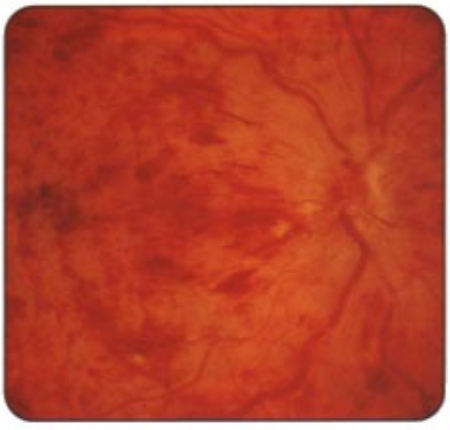

Fig. 11.58

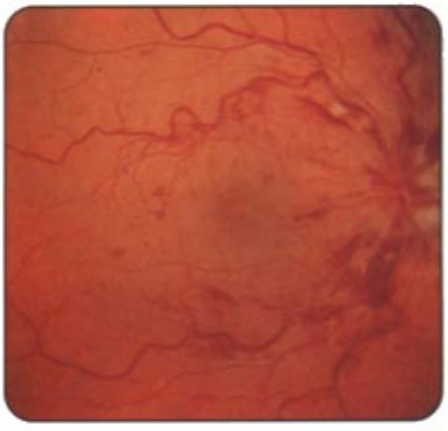

Fig. 11.59

Papillophlebitis

Papillophlebitis is an uncommon, unilateral condition that is also referred to as optic disc vasculitis. Although it may be a variant of central retinal vein occlusion, it affects younger, otherwise healthy individuals and has a better prognosis.

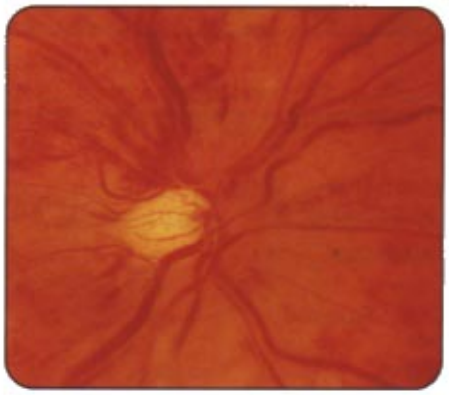

Fig. 11.60

Signs

- Similar to, but usually less severe than, central retinal vein occlusion, with variable disc edema and hyperemia, venous engorgement, and retinal hemorrhages (**Fig. 11.60**).

Longstanding ocular hypotony

Causes

- Glaucoma filtration surgery (**Fig. 11.61**).
- Chronic anterior uveitis (**Fig. 11.62**).
- Ciliochoroidal detachment (**Fig. 11.63**).
- Ocular ischemic syndrome.
- Penetrating trauma (**Fig. 11.64**).
- Cyclodialysis (**Fig. 11.65**).

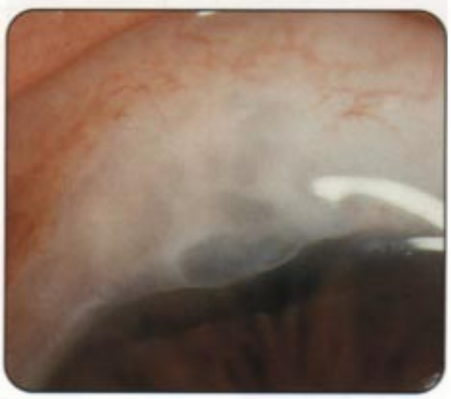

Fig. 11.61

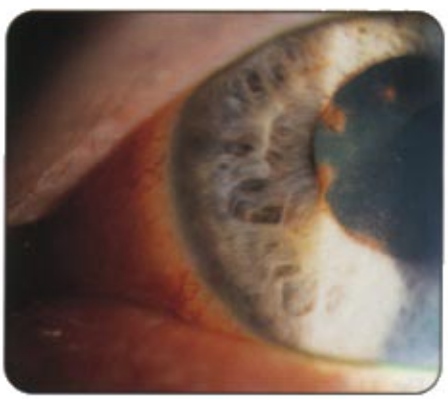

Fig. 11.62

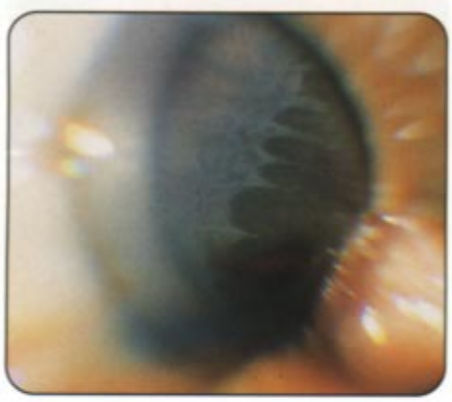

Fig. 11.63

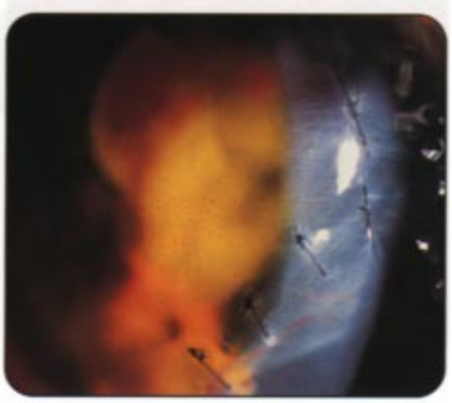

Fig. 11.64

Fig. 11.65

Intermediate uveitis

Intermediate uveitis is a common, usually bilateral but asymmetrical condition that typically affects young individuals.

Signs

- Unilateral or bilateral disc edema without hemorrhages.

Look for

- Mild anterior uveitis.
- Peripheral retinal periphlebitis.
- Vitritis (**Fig. 11.66**).
- Vitreous cotton-balls (**Figs 11.67, 11.68**).
- Inferior peripheral fundus snowbanking (pars planitis).
- Cystoid macular edema.

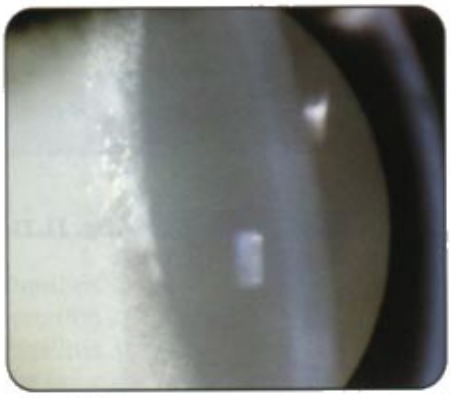

Fig. 11.66

Fig. 11.67

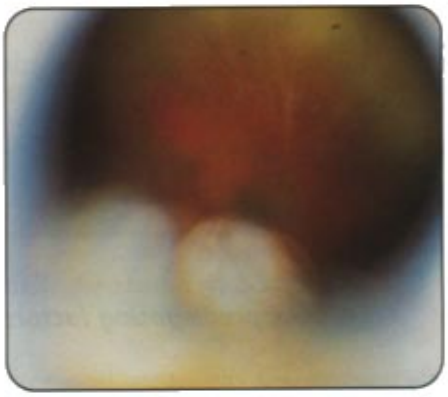

Fig. 11.68

Posterior scleritis

Posterior scleritis is an uncommon, unilateral or bilateral condition that may be associated with systemic disease.

Signs

- Usually unilateral disc edema without hemorrhages (**Fig. 11.69**).
- Other clinical features of posterior scleritis are described in Chapter 4.

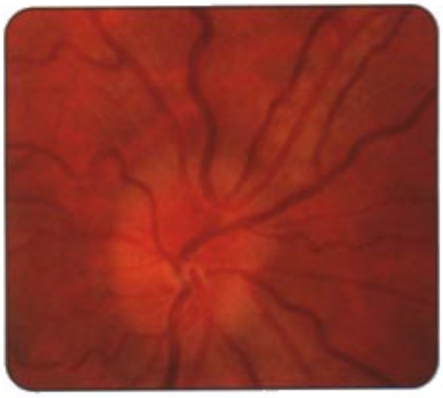

Fig. 11.69

Postcataract papillopathy

Postcataract papillopathy is a rare ischemic condition that occurs between 4 weeks and 15 months after uncomplicated cataract surgery. There is a 50% risk of a similar occurrence in the fellow eye.

Systemic vascular causes

Nonarteritic anterior ischemic optic neuropathy

Nonarteritic anterior ischemic optic neuropathy is an uncommon, initially unilateral condition that typically causes a sudden altitudinal visual field defect. Affected patients are most commonly between 60 and 65 years of age.

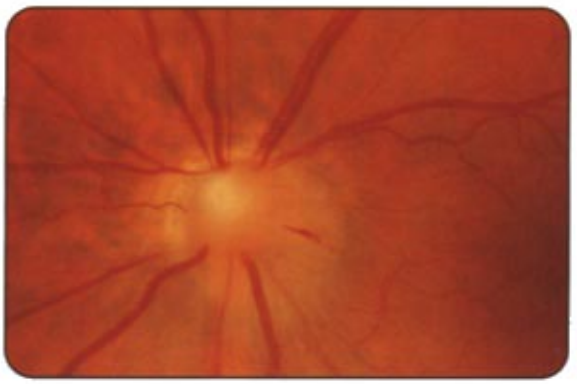

Fig. 11.70

Signs

- Pale, frequently sectorial, disc edema and small peripapillary hemorrhages (**Figs 11.70, 11.71**).

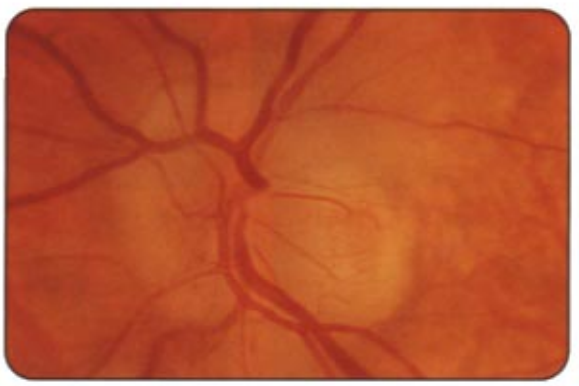

Fig. 11.71

Ocular predisposing factors

- Small optic cup.
- Optic disc drusen.

Systemic predisposing factors

- Hypertension.
- Hyperlipidemia.
- Migraine.
- Acute blood loss.

Arteritic anterior ischemic optic neuropathy

Arteritic anterior ischemic optic neuropathy is caused by giant-cell arteritis and typically occurs between 70 and 80 years of age. It causes an initially unilateral, severe loss of vision, and without treatment, the fellow eye is also frequently affected.

Signs

- Pale, usually diffuse, disc edema and small peripapillary hemorrhages (**Figs 11.72, 11.73**).

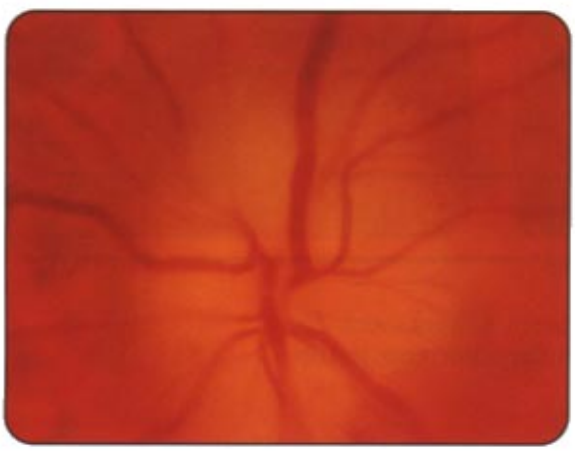

Fig. 11.72

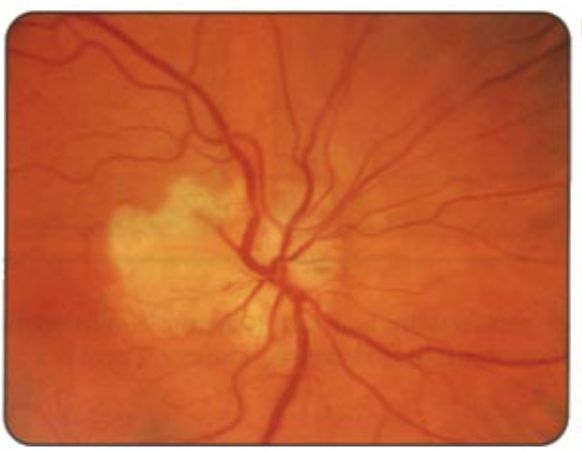

Fig. 11.73

Look for

- Polymyalgia rheumatica.
- Headache.
- Jaw claudication.
- Fever.
- Tender, nodular temporal arteries.
- Scalp necrosis, in very severe cases (**Fig. 11.74**).

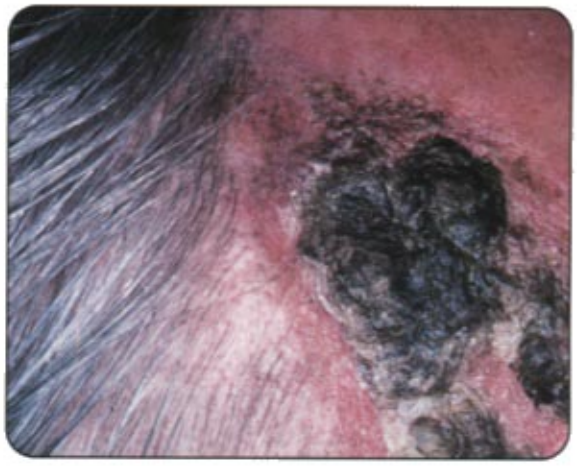

Fig. 11.74

Optic neuritis

Papillitis and retrobulbar neuritis

Papillitis and retrobulbar neuritis are two different manifestations of optic nerve inflammation (optic neuritis). Papillitis is the intraocular form. In retrobulbar neuritis the optic disc is initially normal, but both conditions may give rise to optic atrophy.

Causes of retrobulbar neuritis

- Idiopathic.
- Demyelination – multiple sclerosis, Devic disease, Schilder disease.
- Childhood viral infections.
- Contiguous spread from sinuses.

Causes of papillitis

- Childhood viral infections that typically cause bilateral papillitis.
- Autoimmune – usually associated with systemic lupus erythematosus.
- Sarcoidosis.
- HIV-associated – cytomegalovirus, syphilis, and herpes zoster.
- Demyelination.
- Idiopathic.

Signs of papillitis

- Disc edema, hyperemia, and indistinct disc margins (**Figs 11.75, 11.76**).
- Slit defects in the retinal nerve fiber layer.
- A few white cells may be seen just anterior to the disc.

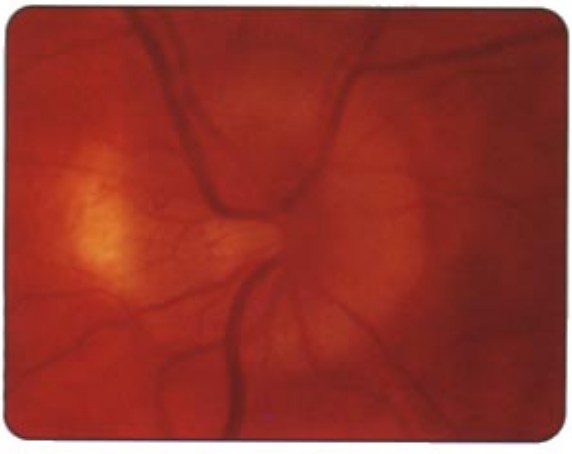

Fig. 11.75

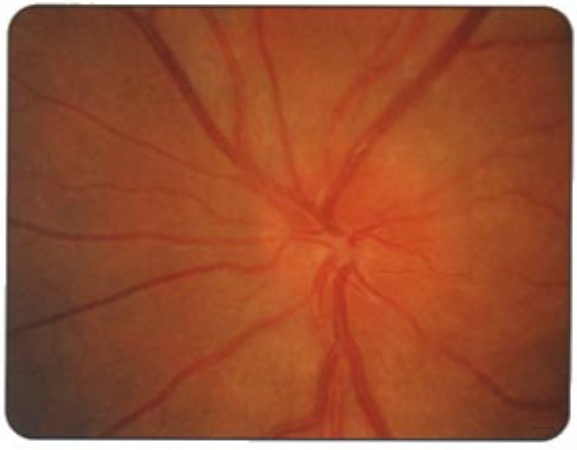

Fig. 11.76

Neuroretinitis

Neuroretinitis is an uncommon, self-limiting condition that causes acute unilateral visual loss. It may be associated with viral infections and cat-scratch fever, but is never associated with demyelination.

Signs

- Papillitis associated with a macular star (**Fig. 11.77**) and, occasionally, juxtapapillary exudates (**Fig. 11.78**).
- The macular star usually becomes more prominent as the papillitis subsides.

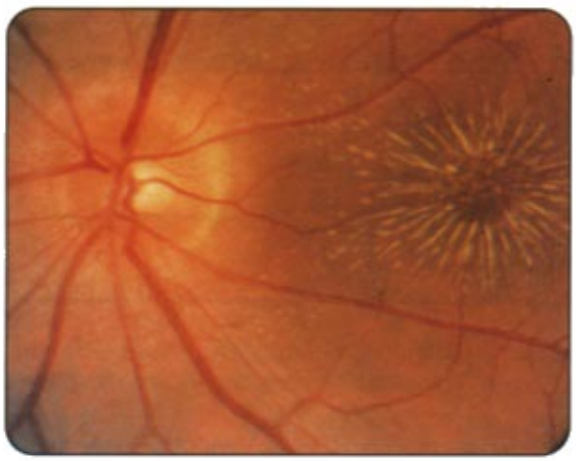

Fig. 11.77

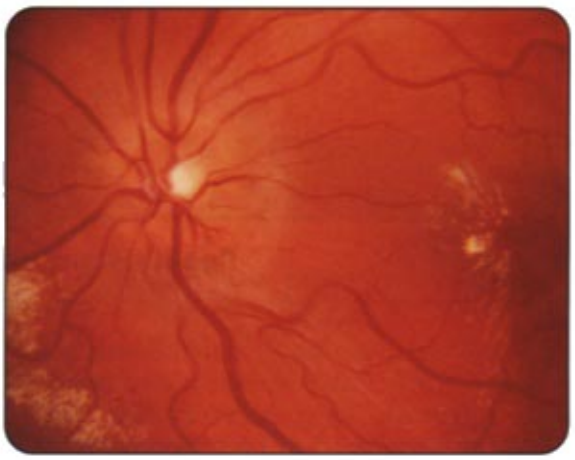

Fig. 11.78

Leber hereditary optic neuropathy

Leber hereditary optic neuropathy is a rare condition with mitochondrial inheritance that typically causes initially unilateral, acute visual loss in young males. The fellow eye is subsequently affected and the eventual outcome is bilateral optic atrophy.

Differential diagnosis

- Optic neuritis, which also occurs in the same age-group.
- Pseudo-Foster Kennedy syndrome.

Acute signs

- Disc hyperemia, vascular tortuosity, and swelling of the peripapillary nerve fiber layer (**Fig. 11.79**).
- Circumpapillary telangiectatic microangiopathy characterized by dilated capillaries usually located on the temporal side of the disc and extending onto the adjacent retina. This does not leak fluorescein and may be seen in asymptomatic family members.
- Pupillary reactions are usually normal.

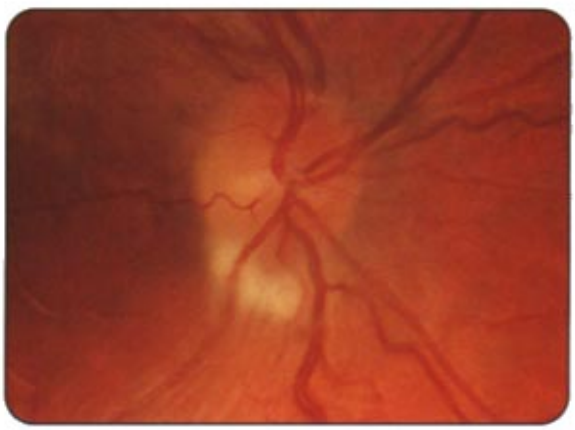

Fig. 11.79

Compressive optic neuropathies

Compressive lesions may initially cause disc edema (**Fig. 11.80**) and later cause optic atrophy. In some cases optic atrophy is the initial feature of compression.

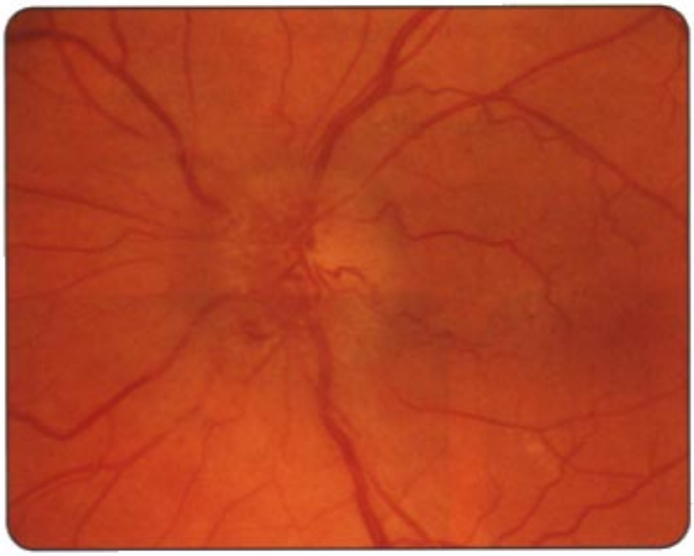

Fig. 11.80

Table 11.4 Causes of optic nerve compression

Intraorbital
• optic nerve glioma
• meningioma
• thyroid ophthalmopathy
• orbital tumors
• orbital cellulitis
Intracanalicular
• Paget disease
• fibrous dysplasia
• osteopetrosis
Intracranial
• meningioma
• pituitary adenoma
Choroidal osteoma

BILATERAL DISC ELEVATION

Buried optic disc drusen

Buried optic disc drusen is a common condition that is frequently bilateral and familial. It is the most common cause of pseudopapilledema in children.

Signs

- Elevated, 'lumpy'-looking disc that is not hyperemic (**Fig. 11.81a and b**).

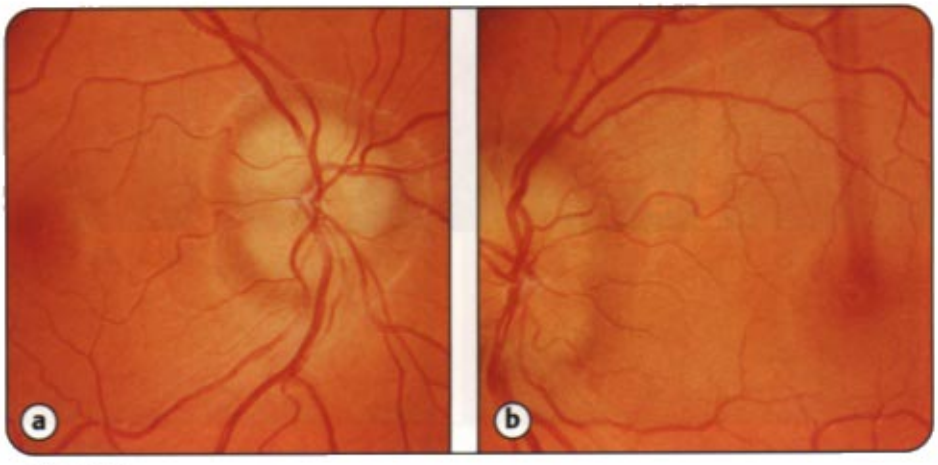

Fig. 11.81

- No physiological cup.
- Careful slit-lamp examination may show buried drusen.
- Hemorrhages are usually absent.
- Disc vessels are not obscured as they pass over the disc.
- Emerging vessels are more numerous than normal and show anomalous branching patterns.
- Peripapillary retinal nerve fiber layer is usually normal.
- Disc autofluorescence.
- With age, the drusen become more easy to detect because they enlarge and become exposed above the disc surface (**Fig. 11.82a and b**).

Occasional ocular associations

- Disc hemorrhages.
- Retinal nerve fiber bundle visual field defects.
- Peripapillary choroidal neovascularization.
- Increased incidence of anterior ischemic optic neuropathy – **Figure 11.83a** shows the involved eye; **Figure 11.83b** shows the normal eye.
- Angioid streaks (**Fig. 11.84**).
- Retinitis pigmentosa (**Fig. 11.85**).

Systemic association

- Alagille syndrome.

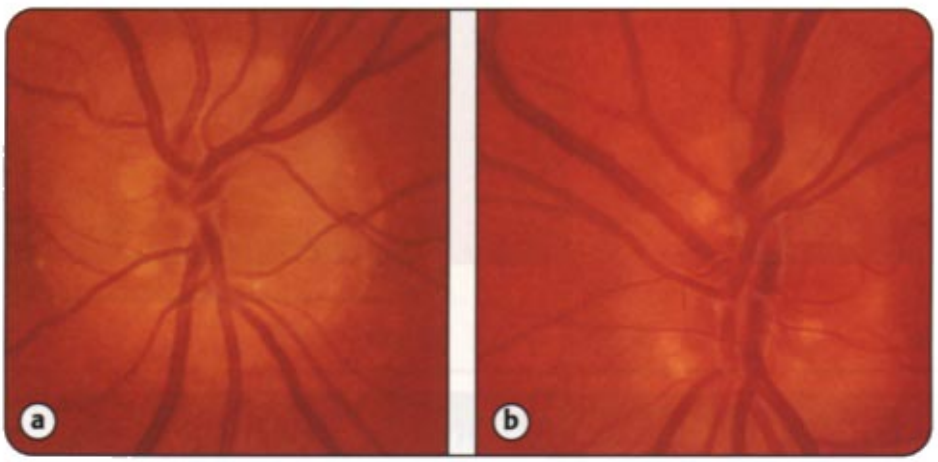

Fig. 11.82

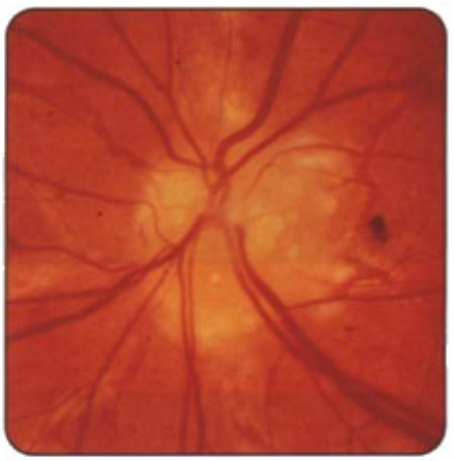

Fig. 11.84

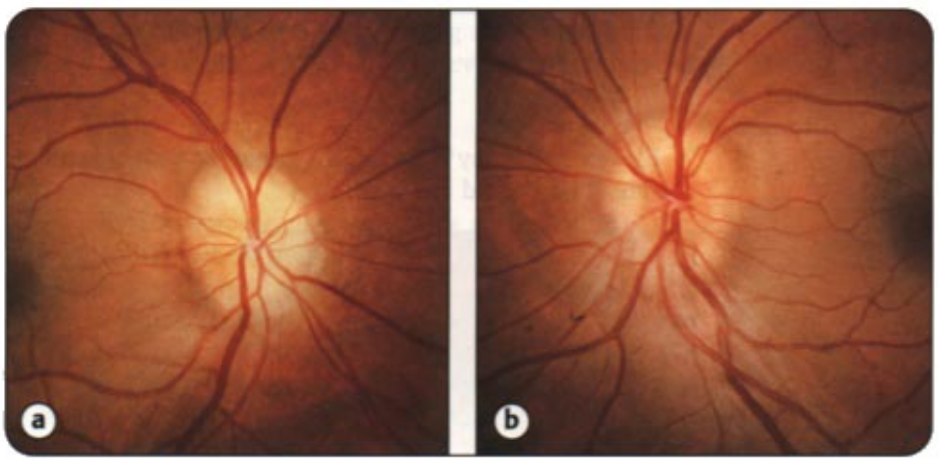

Fig. 11.83

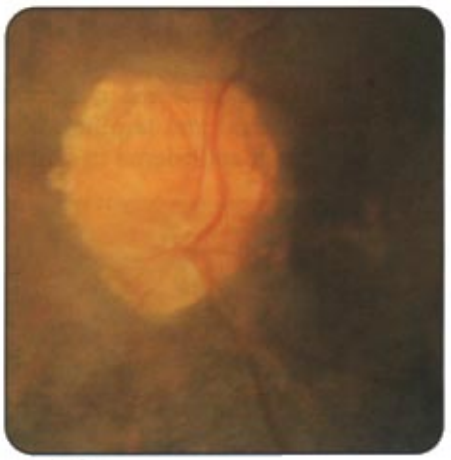

Fig. 11.85

Papilledema

Papilledema is disc edema secondary to raised intracranial pressure. It is invariably bilateral but may be asymmetric. Papilledema is the only condition to cause disc swelling without evidence of optic nerve dysfunction, at least in the early stages. The signs of papilledema depend on its severity and duration. The four stages are: early, established, chronic, and atrophic.

Table 11.5 Causes of raised intracranial pressure

Space-occupying lesions	**Metabolic**
• tumors • hemorrhage	• chronic renal failure • hypercapnia
Idiopathic intracranial hypertension	**Developmental**
(pseudotumor cerebri)	• aqueduct stenosis • craniodysostoses • arteriovenous malformations
Infections	
• meningitis • encephalitis	**Iatrogenic**
	• radical neck surgery • tetracycline
Toxic	
• lead poisoning • chronic hypervitaminosis A	

Early papilledema

Signs

- Mild disc hyperemia and elevation, with preservation of the physiological cup (**Fig. 11.86a and b**).
- Indistinct disc margins and peripapillary retinal nerve fiber striations (**Fig. 11.87a and b**).
- The nasal sector is usually involved first, followed by the superior and inferior sectors.
- Spontaneous venous pulsation is absent – this may not be significant because it is also absent in about 20% of the general population.

Differential diagnosis

- Buried optic disc drusen (*see* **Fig. 11.81a and b**).
- Hypermetropia (*see* **Fig. 11.1**).

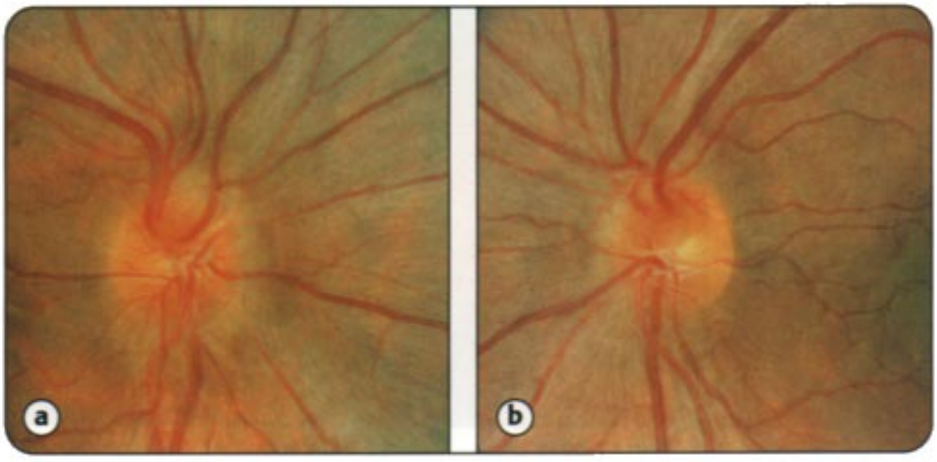

Fig. 11.86

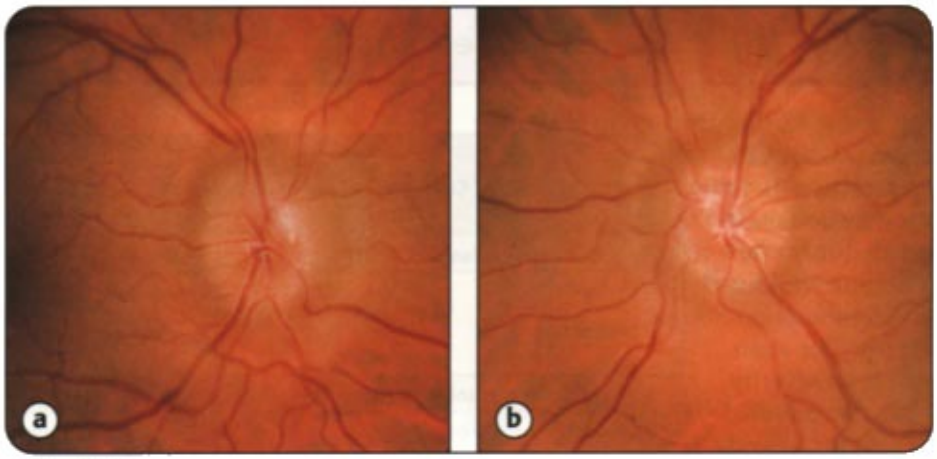

Fig. 11.87

Established (acute) papilledema

Signs

- Disc elevation, blurring of the entire disc margin, absence of the physiological cup, venous engorgement, and flame-shaped hemorrhages (**Figs 11.88a and b, 11.89a and b, 11.90a and b**).
- Cotton-wool spots on the disc (**Fig. 11.88a**).
- Circumferential peripapillary retinal folds (Paton lines) due to fluid accumulation (**Fig. 11.90b**).

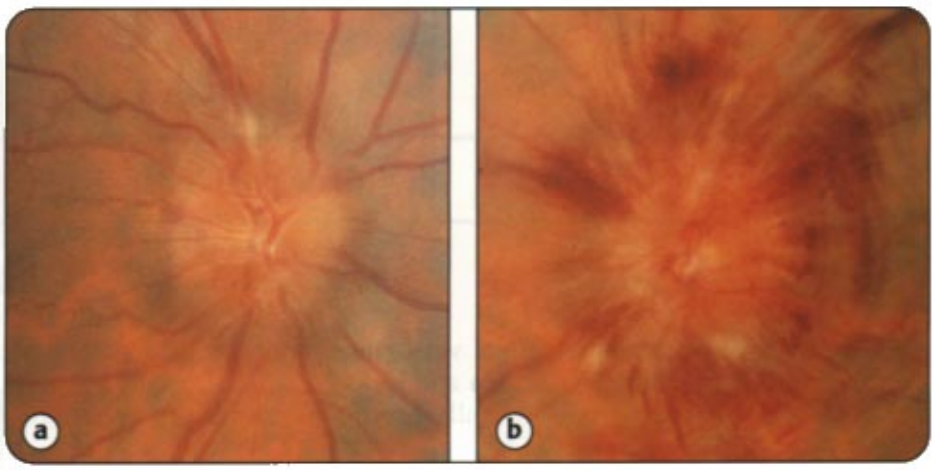

Fig. 11.88

Dx Differential diagnosis

- Malignant hypertension (*see* **Fig. 11.95a and b**).

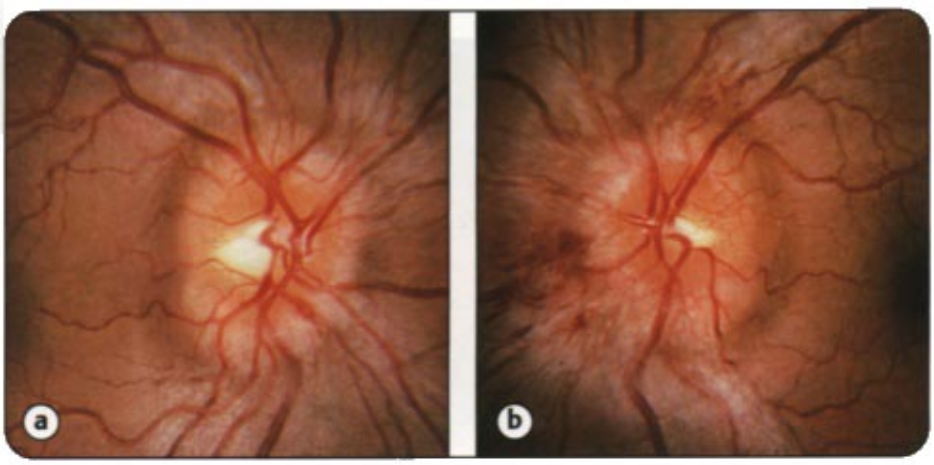

Fig. 11.89

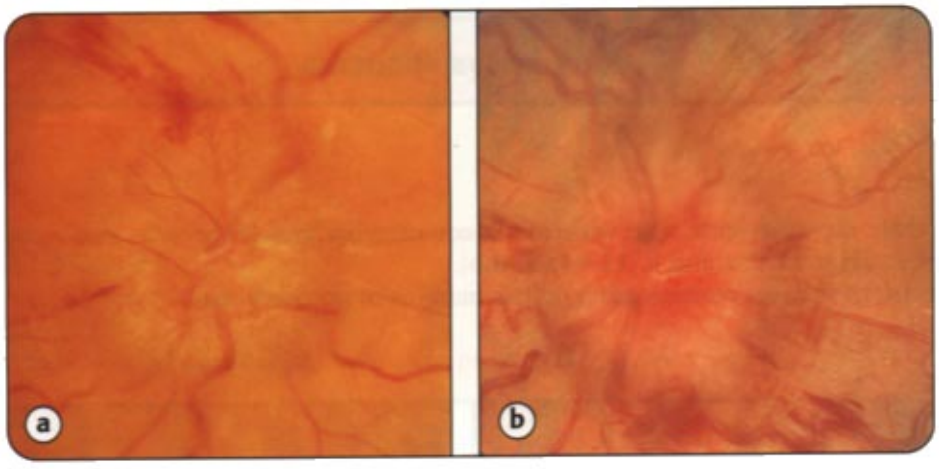

Fig. 11.90

Chronic papilledema

Signs

- Marked 'champagne-cork-like' elevation of the disc, without hemorrhages (**Figs 11.91a and b, 11.92a and b**).
- Optico-ciliary shunts and drusen-like crystalline deposits may be present on the disc.
- A macular star may also be present.

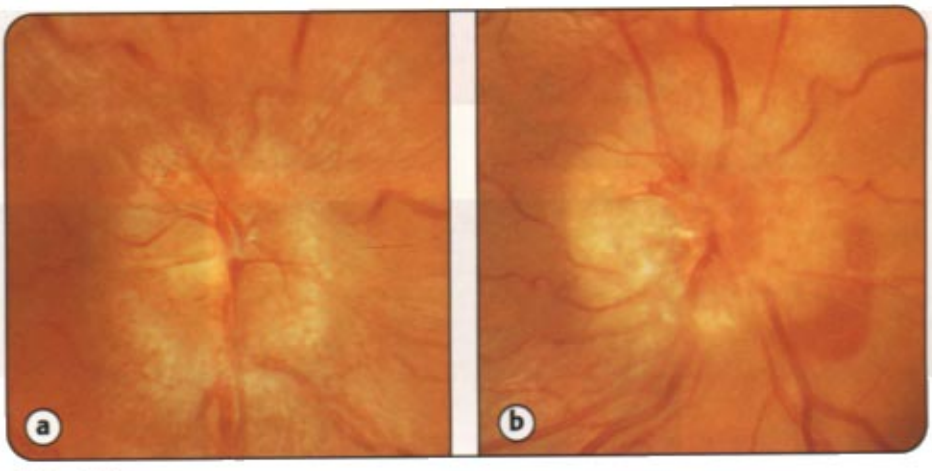

Fig. 11.91

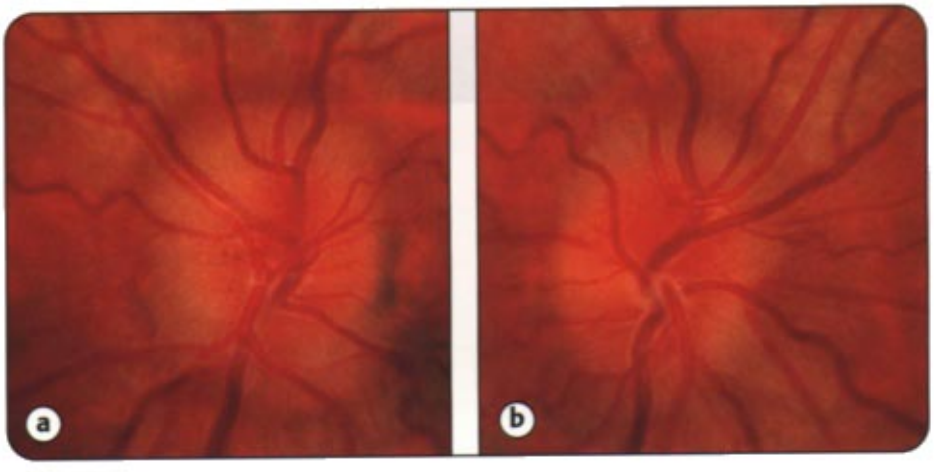

Fig. 11.92

Atrophic papilledema

Signs

- Grayish, pale, slightly elevated optic disc with indistinct margins and fewer crossing blood vessels (**Figs 11.93a and b, 11.94a and b**).
- Peripapillary pigmentary changes corresponding to the margins of previous disc edema may be seen.

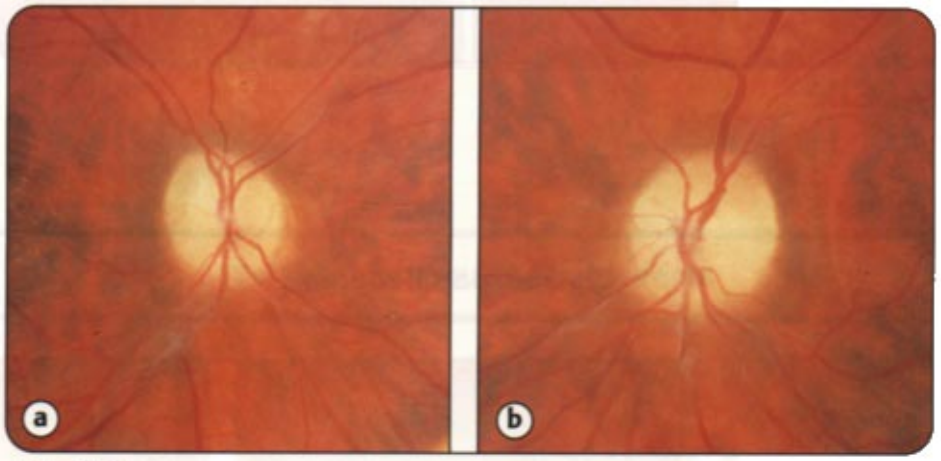

Fig. 11.93

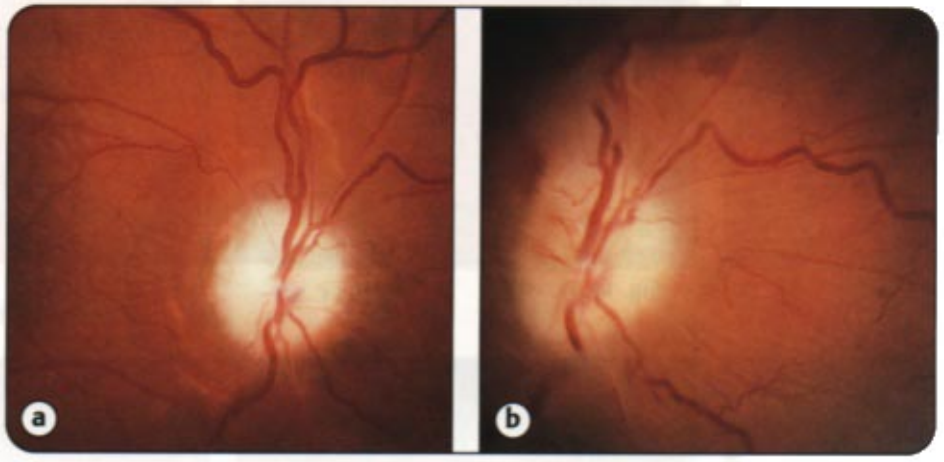

Fig. 11.94

Malignant hypertension

Signs

- Bilateral disc swelling, flame-shaped hemorrhages, cotton-wool spots, and a macular star (**Fig. 11.95a and b**).
- According to the age of the patient there are also variable hypertensive vascular changes.
- Pale peripheral areas indicative of choroidal infarcts may be present (**Fig. 11.96**).

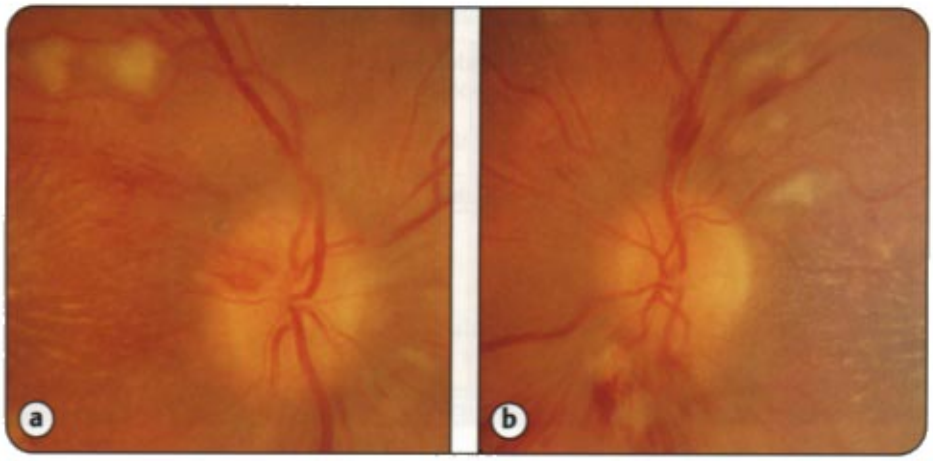

Fig. 11.95

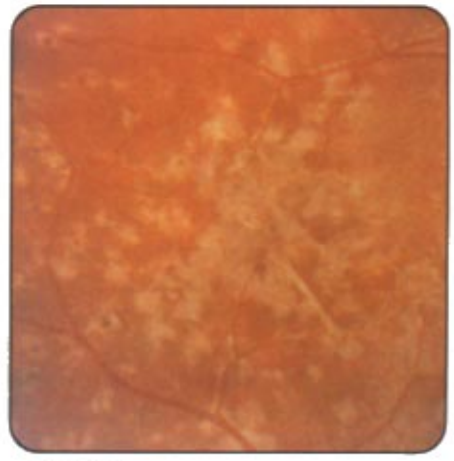

Fig. 11.96

Miscellaneous causes

- Compressive thyroid optic neuropathy is an uncommon condition that may give rise to bilateral disc swelling in the absence of significant proptosis.
- Diabetic papillopathy is an uncommon condition that is bilateral in 75% of cases. It typically causes mild-to-moderate visual loss in young patients with juvenile-onset diabetes, which usually recovers spontaneously within 6 months.
- Acute methanol toxicity is a very rare condition that is characterized by acute bilateral visual loss and dilated pupils.
- Bilateral, simultaneous, arteritic anterior ischemic optic neuropathy.
- Bilateral papillitis.
- Compromised venous drainage may result in bilateral disc swelling in patients with cavernous sinus thrombosis or direct carotid–cavernous fistula.

OPTIC ATROPHY

Primary optic atrophy

Primary optic atrophy is caused by any process affecting the visual pathways from the retrolaminar portion of the optic nerve to the lateral geniculate nucleus. Lesions affecting the anterior visual pathways from the globe to the chiasm will cause unilateral optic atrophy, whilst those affecting the optic chiasm or optic tracts will cause bilateral optic atrophy.

Signs

- White, flat optic disc with a clearly delineated outline, without significant gliosis (**Fig. 11.97**).
- Reduction in the number of small blood vessels crossing the disc.
- Attenuation of the peripapillary vessels.
- Thinning of the retinal nerve fiber layer.

The atrophy may be diffuse or sectorial depending on the cause and level of the lesion. For example, optic atrophy caused by a chiasmal lesion may involve the nasal and temporal portions but spare the superior and inferior (band atrophy).

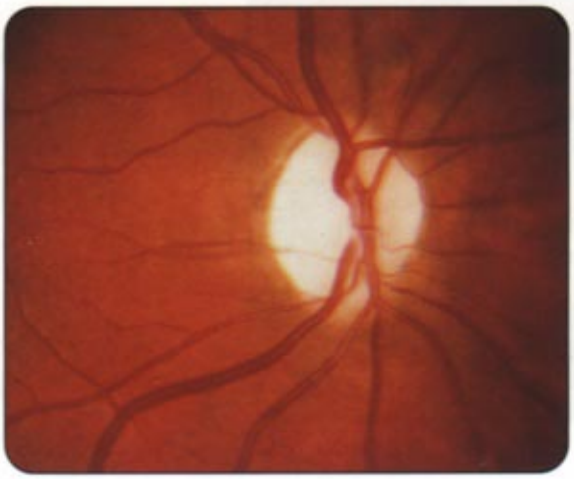

Fig. 11.97

Table 11.6 Causes of primary optic atrophy

Following retrobulbar neuritis
Compressive lesions *Of optic nerve (see Table 11.4)* *Of chiasm and optic tract (see below)*
• tumors – pituitary adenoma, craniopharyngioma, and suprasellar meningioma • aneurysm • chiasmal arachnoiditis
Toxic optic neuropathies
• methanol • ethambutol • isoniazid • rare causes – amiodarone, streptomycin, and chlorpropamide
Nutritional (deficiency) optic neuropathies
• thiamine deficiency – tobacco–alcohol amblyopia and beriberi • vitamin B_{12} deficiency – pernicious anemia • niacin – pellagra
Traumatic optic neuropathies
• with or without transection of the optic nerve • retrobulbar hemorrhage
Hereditary optic atrophies

Consecutive optic atrophy

Consecutive optic atrophy is caused by diseases of the inner retina or its blood supply. The cause is usually obvious from examination of the fundus (**Table 11.7**).

Fig. 11.98

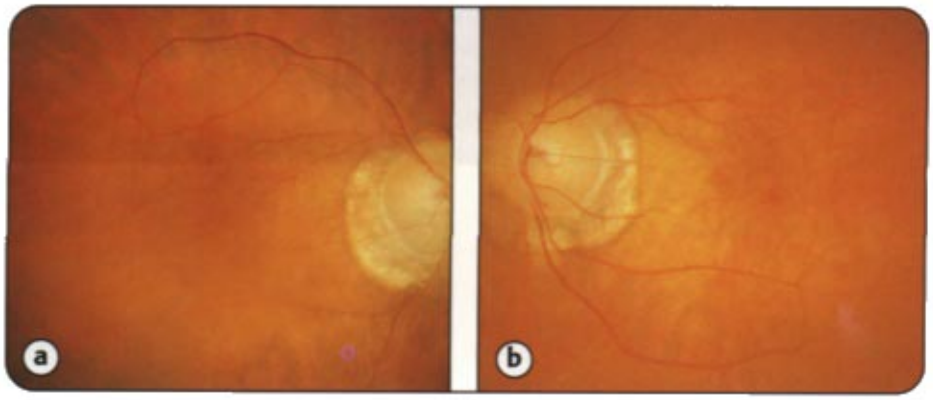

Fig. 11.99

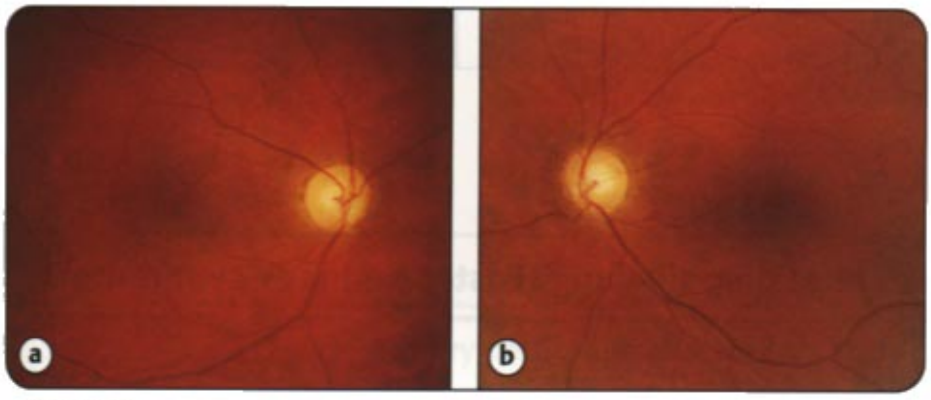

Fig. 11.100

Fig. 11.101

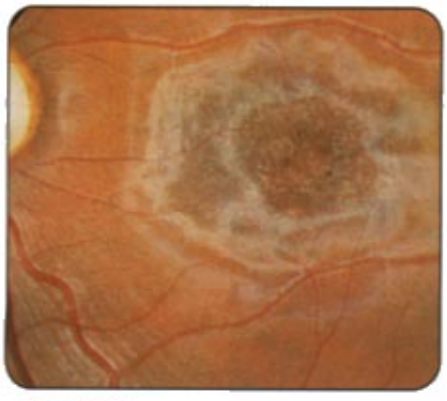

Fig. 11.102

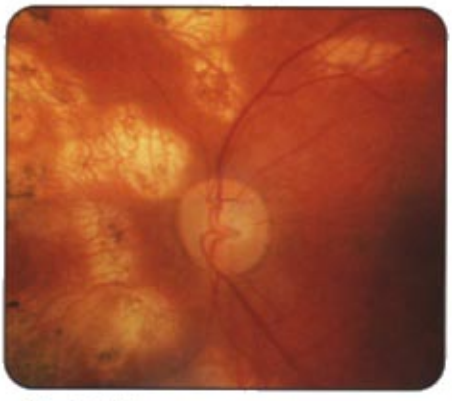

Fig. 11.103

Table 11.7 Fundus changes and consecutive optic atrophy
Vascular attenuation
• old central retinal artery occlusion (Fig. 11.98) • retinitis pigmentosa (Fig. 11.99a and b) • toxic optic neuropathy (Fig. 11.100a and b) • old vasculitis (Fig. 11.101)
Macular lesions giving rise to temporal disc pallor
• toxoplasmosis • cone dystrophy (Fig. 11.102)
Chorioretinal atrophy
• degenerative myopia • extensive panretinal photocoagulation (Fig. 11.103)
Diffuse retinal necrosis
• cytomegalovirus retinitis • acute retinal necrosis • Behçet disease
Cherry red spot at macula syndromes (*see* Chapter 12)

Secondary optic atrophy

Secondary optic atrophy is preceded by swelling of the optic nerve head as a result of edema, ischemia, or inflammation.

Causes

- Chronic papilledema (*see* **Figs 11.93 and 11.94**).
- Anterior ischemic optic neuropathy (**Fig. 11.104a** shows the acute stage and **Fig. 11.104b** shows optic atrophy).
- Papillitis.

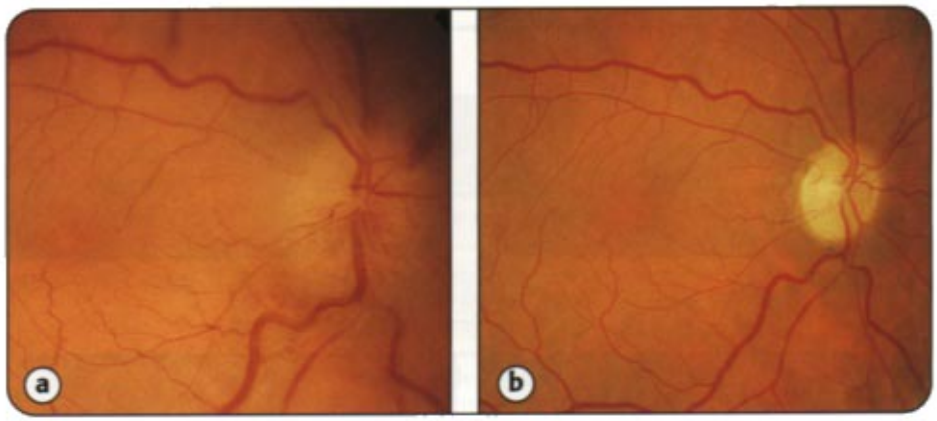

Fig. 11.104

Optic atrophy with contralateral disc swelling (Foster Kennedy syndrome)

True Foster Kennedy syndrome is a very rare condition that is classically caused by a frontal-space-occupying lesion.

Pseudo-Foster Kennedy syndrome is most frequently caused by anterior ischemic optic neuropathy in which there is optic atrophy in one eye from a previous episode and contralateral disc edema due to active disease.

Signs

- Contralateral papilledema due to raised intracranial pressure (**Fig. 11.105a**).
- Ipsilateral compressive optic atrophy by the tumor (**Fig. 11.105b**).

Onset at 4 years: simple (recessive)

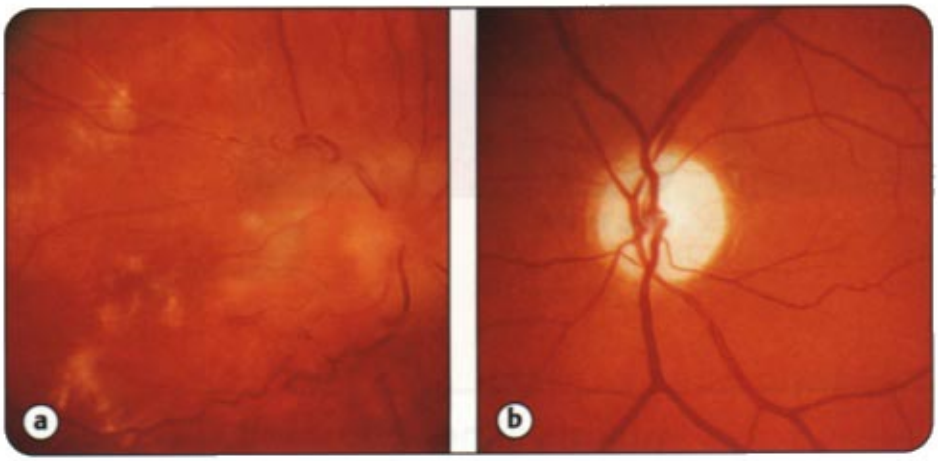

Fig. 11.105

Primary hereditary optic atrophies

- Diffuse optic atrophy with severe visual loss.

Onset at about 10 years: Kjer juvenile (dominant)

- Temporal disc pallor (**Fig. 11.106a and b**) with mild-to-moderate insidious visual loss.
- Tritan (blue–yellow) color deficit.

Onset between 5 and 14 years (recessive)

- Associated with diabetes insipidus, diabetes mellitus, optic atrophy, and deafness (DIDMOAD).
- Temporal disc pallor.

Onset at 16–30 years: Leber's hereditary optic neuropathy (mitochondrial); (*see* **Fig. 11.79**).

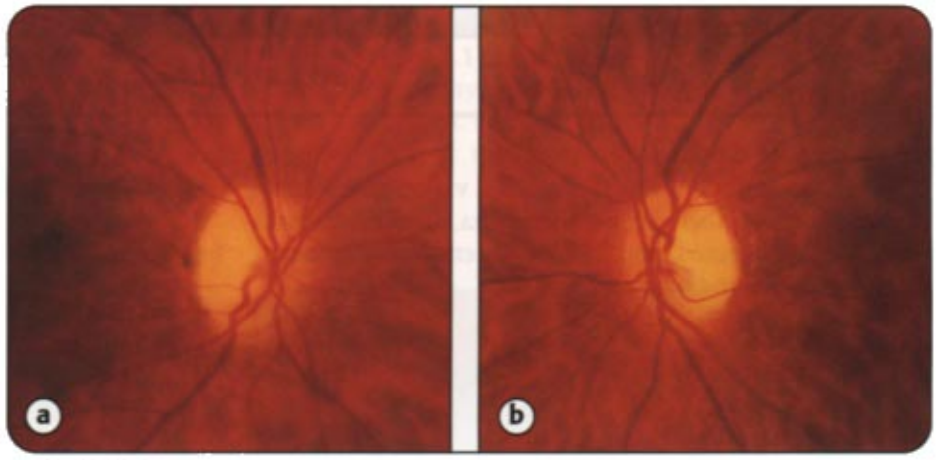

Fig. 11.106

Hereditary neurological disorders with optic atrophy

Hereditary neurological disorders with optic atrophy present during the first decade of life.

- Behr (recessive).
- Friedreich ataxia (recessive).
- Charcot–Marie–Tooth disease (dominant, X-linked).
- Adrenoleukodystrophies (two types – X-linked recessive and autosomal recessive).
- Cerebellar ataxia type I (dominant).

LESIONS OBSCURING THE OPTIC DISC

Primary optic disc tumors

Capillary angioma

Capillary angioma is a rare, benign but vision-threatening tumor that may also involve the retina. In 50% of cases both eyes are affected, and 25% of patients have associated von Hippel–Lindau syndrome.

Signs

- Orange-red lesion on the optic disc (**Fig. 11.107**) which may be associated with macular hard exudates (**Fig. 11.108**).
- Peripheral angiomas are associated with grossly dilated and tortuous blood vessels.

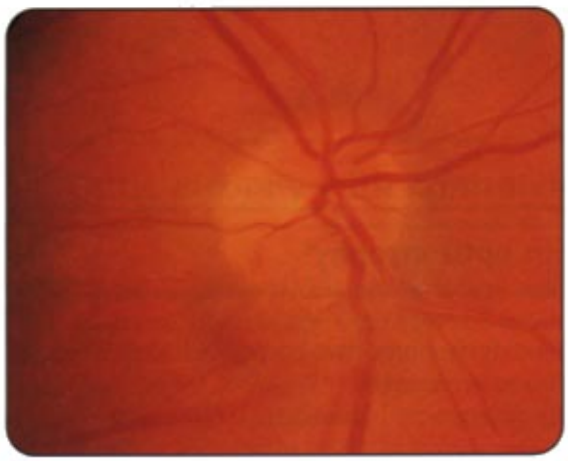

Fig. 11.107

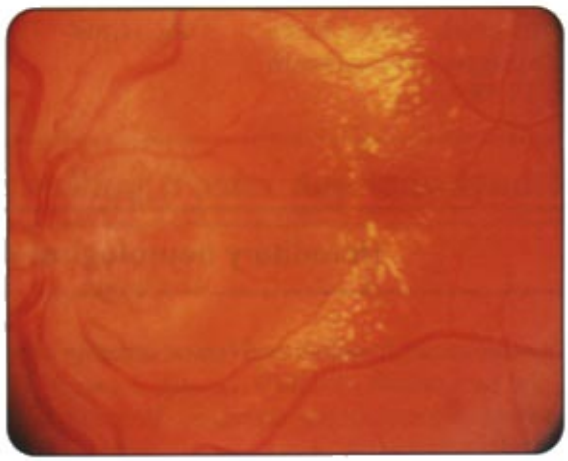

Fig. 11.108

Cavernous angioma

Cavernous angioma is a very rare, unilateral, benign and innocuous tumor that may also involve the retina.

Signs

- Clumps of thin-walled saccular aneurysms filled with dark blood vessels reminiscent of a cluster of grapes (**Fig. 11.109**).

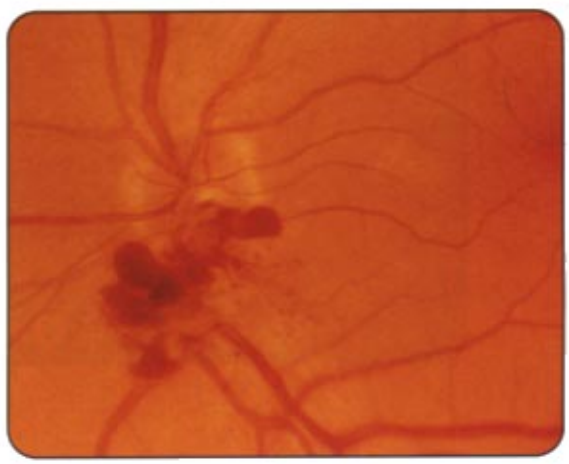

Fig. 11.109

Melanocytoma

Melanocytoma is a very rare, unilateral, benign tumor that typically affects heavily pigmented races.

Signs

- Pigmented disc lesion frequently located eccentrically (**Fig. 11.110**) which may spill over into the adjacent retina (**Figs 11.111, 11.112**).
- The tumor may interfere with cytoplasmic transport and induce disc swelling (**Fig. 11.113**).
- About 15% of melanocytomas increase in size over 5–20 years.

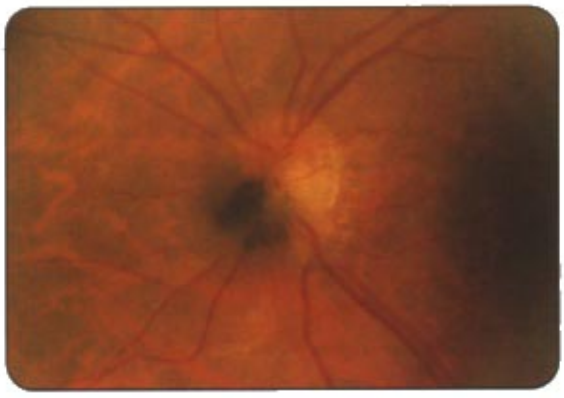

Fig. 11.110

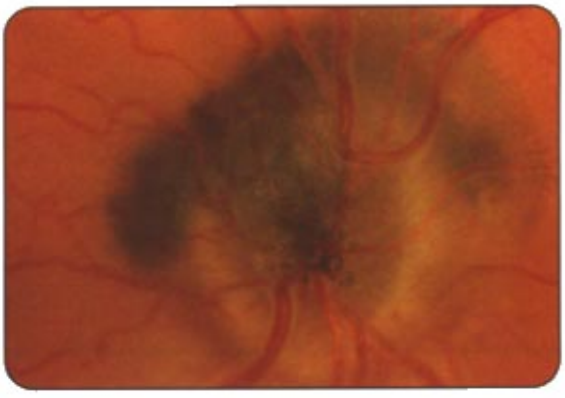

Fig. 11.111

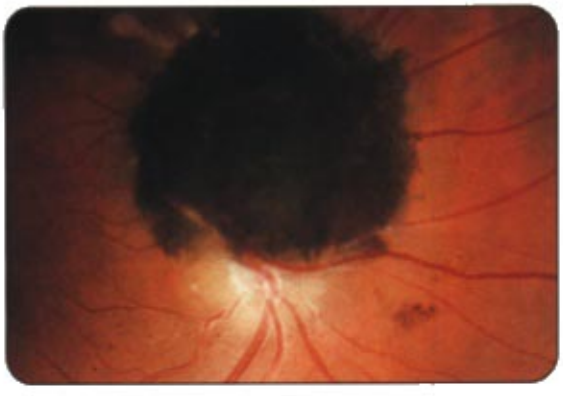

Fig. 11.112

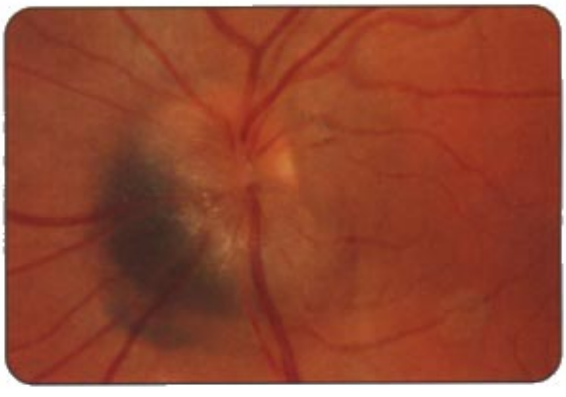

Fig. 11.113

Astrocytoma

Astrocytoma is a rare, usually unilateral, benign and innocuous tumor that may also involve the retina. It is frequently associated with tuberous sclerosis.

Signs

- Semi-translucent mulberry-like lesion (**Fig. 11.114**) that displays autofluorescence.

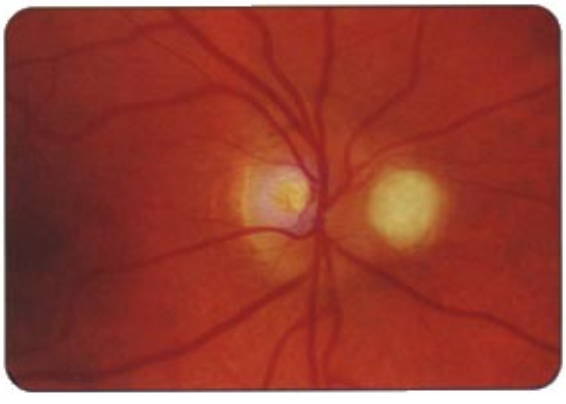

Fig. 11.114

Combined hamartoma of the retina and RPE

Combined hamartoma of the retina and RPE is a very rare, benign but vision-threatening, usually unilateral condition that may also affect the retina. It usually occurs in isolation but may be associated with neurofibromatosis type 2.

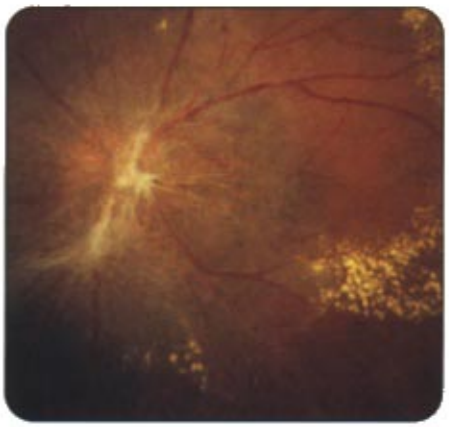

Fig. 11.115

Signs

- Thickened epiretinal membrane overlying a dark-gray-pigmented hamartoma which is associated with stress lines in the surrounding retina (**Fig. 11.115**).
- Peripheral lesions may cause dragging of the adjacent retinal vessels.

Infiltrative lesions

Fig. 11.116

Table 11.8 Infiltrative disc lesions

Neoplastic
• adjacent tumors – retinoblastoma and choroidal melanoma
• systemic malignancies – leukemia, lymphoma, and metastatic carcinoma
Granulomatous and inflammatory
• sarcoid granuloma (Fig. 11.116)
• toxocara (Figs. 11.117, 11.118)
• cytomegalovirus (Fig. 11.119)

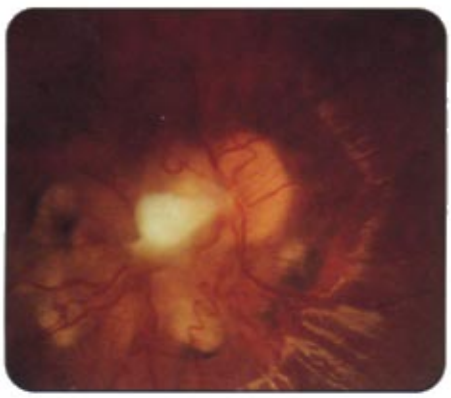

Fig. 11.117

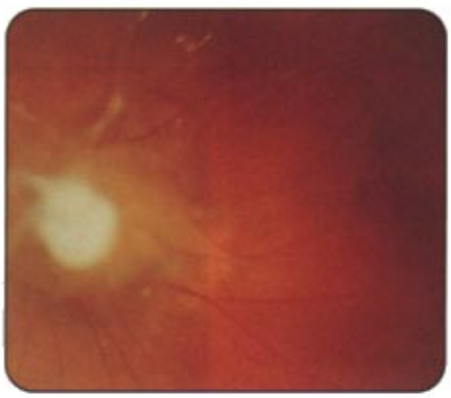

Fig. 11.118

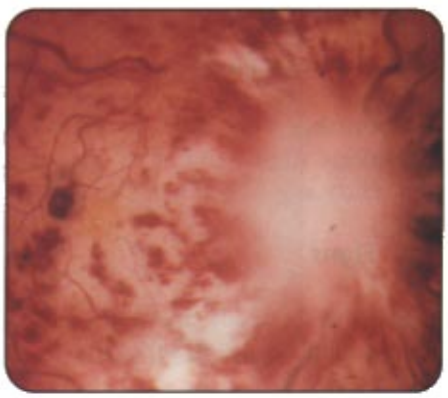

Fig. 11.119

JUXTAPAPILLARY LESIONS

Atrophy

Chronic glaucoma

Signs

- The central zone, beta, borders the peripapillary scleral ring and is characterized by visible sclera and large choroidal blood vessels.
- A peripheral zone, alpha, is characterized by variable irregular hyper- and hypopigmentation of the retinal pigment epithelium (**Fig. 11.120**).

These changes are larger and occur more frequently in glaucomatous than in normal eyes.

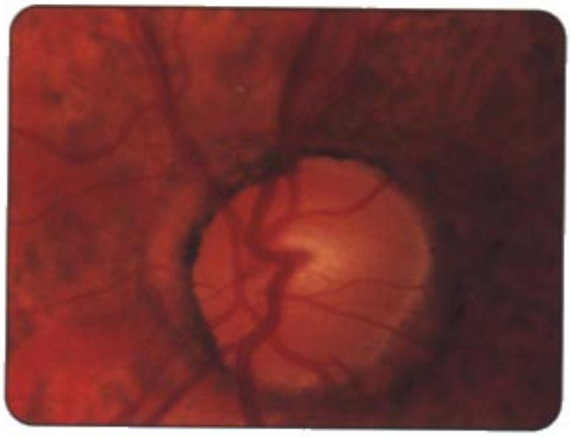

Fig. 11.120

High myopia

Signs

Peripapillary atrophy (**Fig. 11.121**) that may be associated with:

- Maculopathy (**Fig. 11.122**).
- Chorioretinal atrophy (**Fig. 11.123**).

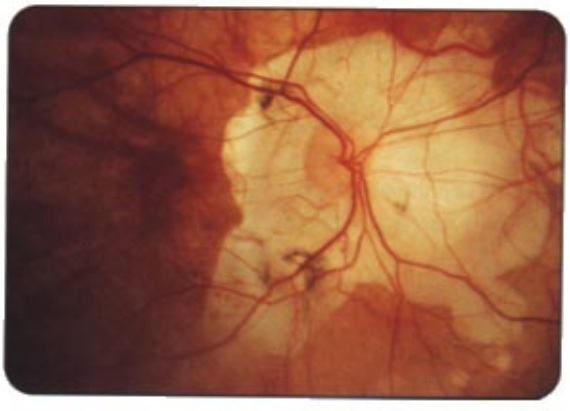

Fig. 11.121

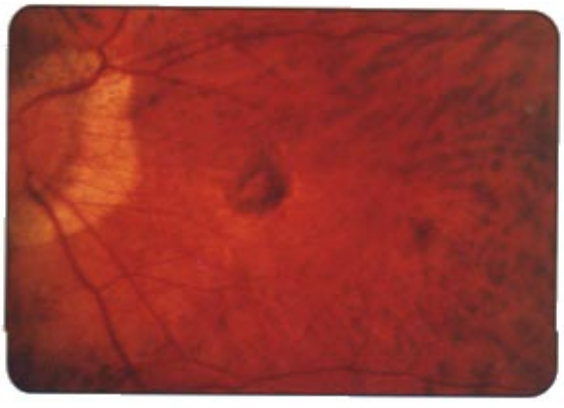

Fig. 11.122

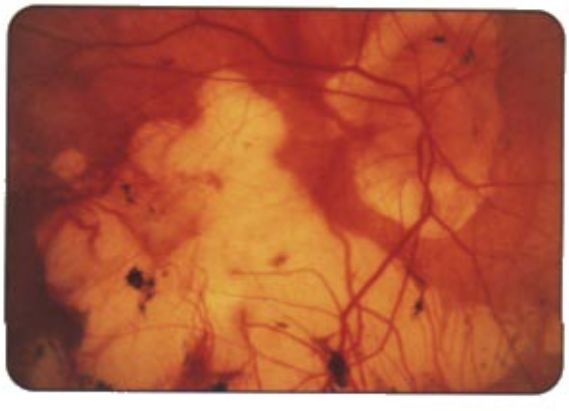

Fig. 11.123

Presumed ocular histoplasmosis syndrome

Signs

Peripapillary atrophy (**Fig. 11.124**) associated with:
- Atrophic punched-out 'histo' spots in the mid-periphery (**Fig. 11.125**) and posterior pole.
- Maculopathy.
- Peripheral linear streaks of chorioretinal atrophy.

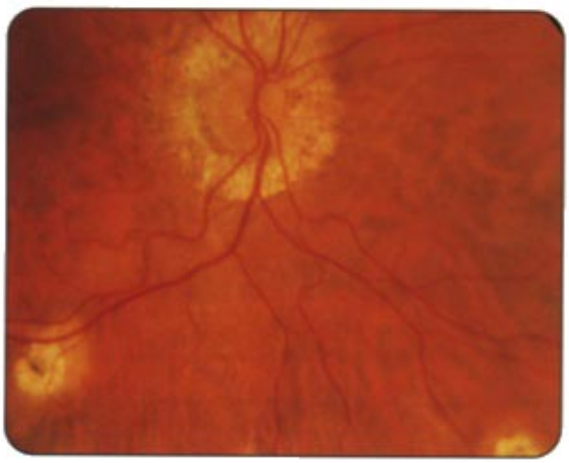

Fig. 11.124

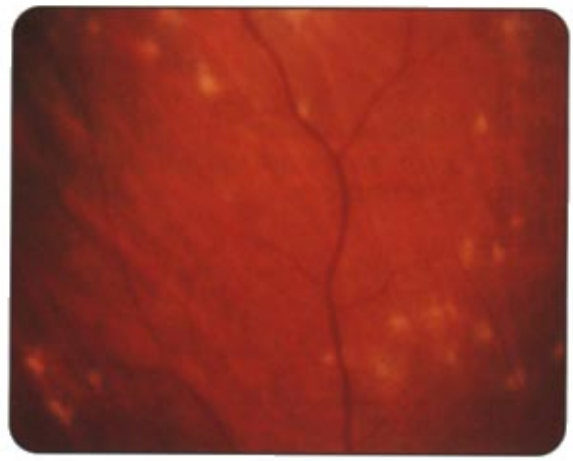

Fig. 11.125

Postinflammatory

Postinflammatory is a common, usually unilateral, chorioretinal scar, most frequently caused by old toxoplasmosis.

Signs

- Peripapillary area of variable size associated with pigmentary changes (**Fig. 11.126**).

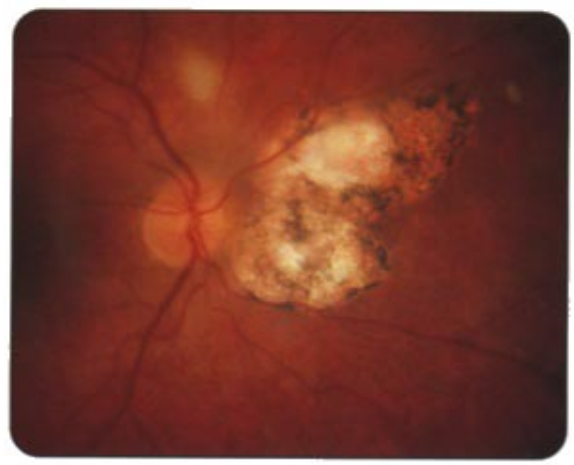

Fig. 11.126

Juxtapapillary coloboma

Juxtapapillary coloboma is an uncommon, unilateral, congenital anomaly.

Signs

- Atrophic area located inferiorly (**Fig. 11.127**).

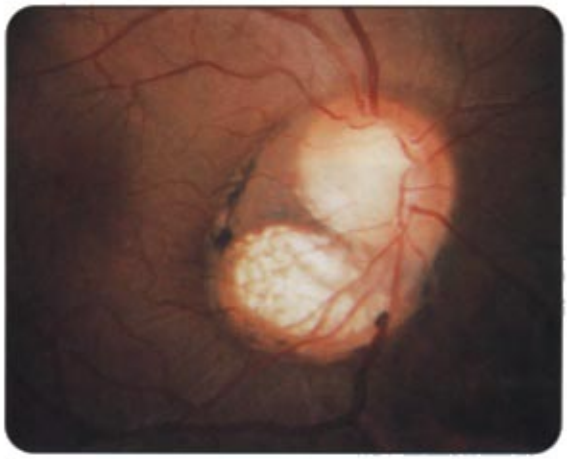

Fig. 11.127

Serpiginous choroidopathy

Serpiginous choroidopathy typically starts at the edge of the disc and spreads outward (**Fig. 11.128**).

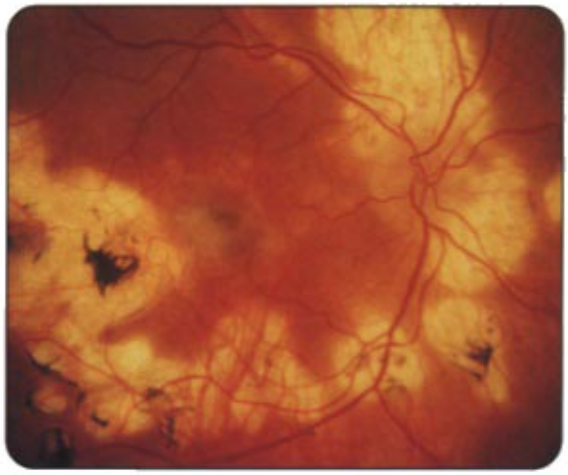

Fig. 11.128

Miscellaneous

Normal variants

- Congenital crescent (**Fig. 11.129**).

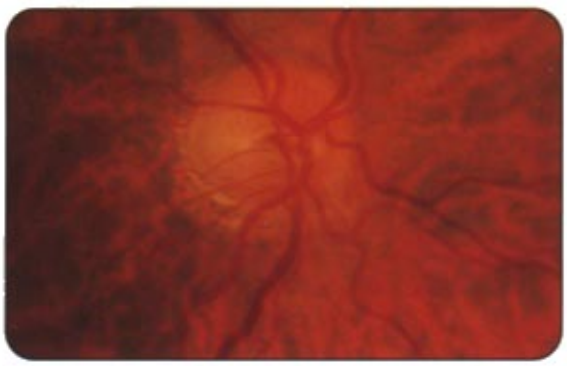

Fig. 11.129

- Pigment crescent (**Fig. 11.130**).

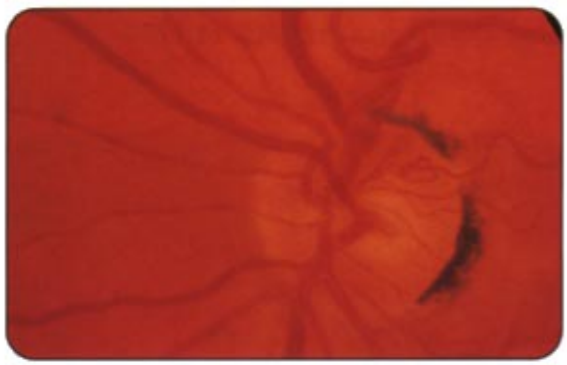

Fig. 11.130

Opaque nerve fibers

Opaque nerve fibres is a congenital condition that may, on cursory examination, be mistaken for disc edema. It is present to some extent in 1% of the general population and is bilateral in 20% of cases.

Signs

- White lesions with feathery edges which conform to the configuration of the retinal nerve fiber layer.
- The fibers may be peripapillary and radiate outward from the disc (**Figs 11.131–11.133**) or they may be peripheral and isolated (**Fig. 11.134**).

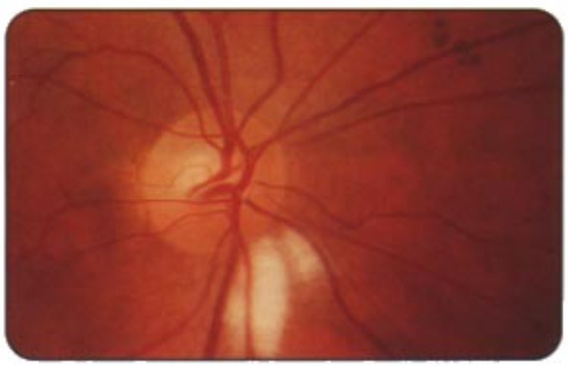

Fig. 11.131

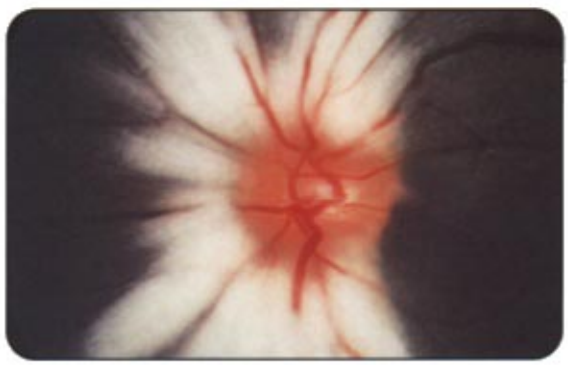

Fig. 11.132

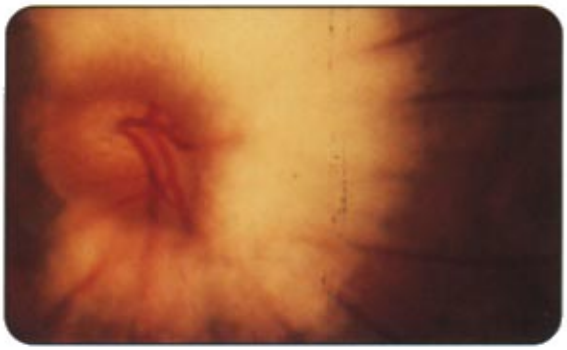

Fig. 11.133

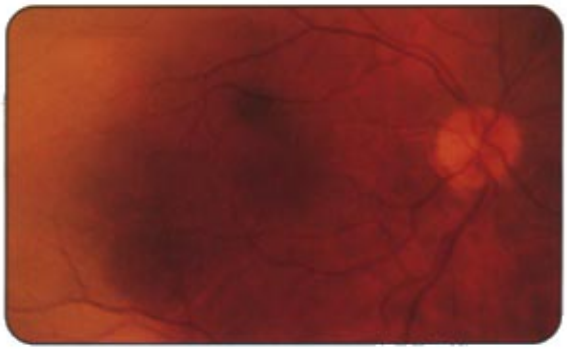

Fig. 11.134

Juxtapapillary choroidal neovascularization

Signs

- Serous retinal elevation associated with hemorrhage, usually located at the temporal border of the disc (**Fig. 11.135**).

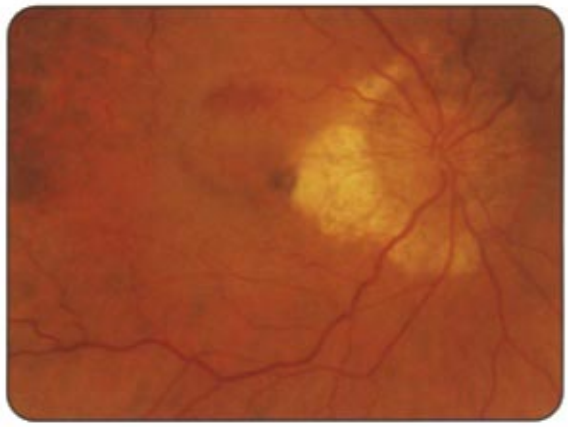

Fig. 11.135

Juxtapapillary angioid streaks

Signs

- Angioid streaks typically encircle the disc (**Fig. 11.136**), then radiate outward.

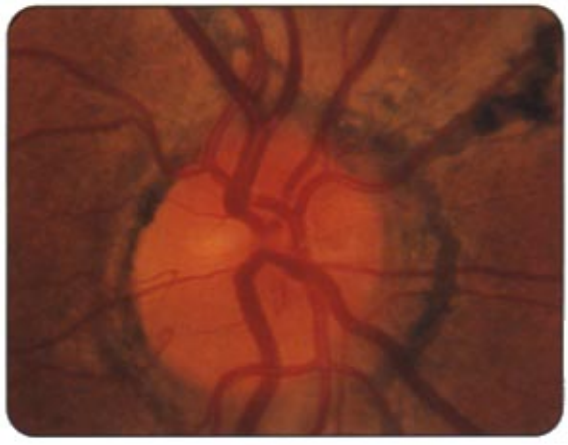

Fig. 11.136

Choroidal osteoma

Choroidal osteoma is a very rare tumor that typically affects young females.

Signs

- Juxtapapillary orange–yellow lesions with scalloped borders which may in some cases encircle the disc (**Fig. 11.137**).

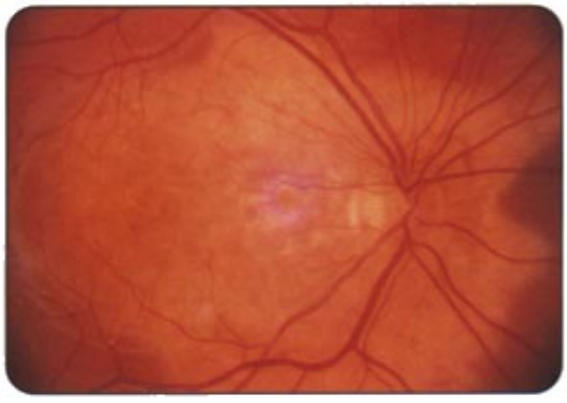

Fig. 11.137

12 The fundus

VASCULAR LESIONS 289

Hemorrhages
- Preretinal hemorrhages
- Retinal flame-shaped hemorrhages
- Roth spots
- Dot and blot hemorrhages
- Subretinal hemorrhages
- Sub-RPE hemorrhages
- Choroidal hemorrhages

Hard exudates
- Arranged in rings or clumps
- Stellate maculopathy
- Subretinal exudates

Cotton-wool spots

Neovascularization

Retinal vasculitis

Retinal emboli
- Fibrinoplatelet emboli
- Cholesterol (Hollenhorst) plaques
- Calcific emboli
- Miscellaneous emboli

Major changes in vascular caliber

Major vascular malformations
- Retinal artery macroaneurysm
- Leber miliary aneurysms
- Coats disease
- Retinal capillary hemangioma
- Retinal cavernous hemangioma
- Retinal racemose hemangioma

INFLAMMATORY LESIONS 309

Idiopathic multifocal white dot syndromes
- Multiple evanescent white dot syndrome
- Punctate inner choroidopathy
- Birdshot retinochoroidopathy
- Acute posterior multifocal placoid pigment epitheliopathy
- Mulitifocal choroiditis with panuveitis syndrome
- Serpiginous choroidopathy
- Diffuse subretinal fibrosis syndrome

Focal infectious posterior uveitis
- Toxoplasmosis
- Toxocariasis
- Candidiasis
- Presumed ocular histoplasmosis syndrome
- Choroidal pneumocystosis

Diffuse infectious retinitis
- Cytomegalovirus retinitis
- Acute retinal necrosis
- Progressive outer retinal necrosis
- Herpes simplex retinitis
- Measles retinitis

Panuveitis
- Sarcoidosis
- Behçet disease
- Vogt–Koyanagi–Harada syndrome
- Sympathetic ophthalmitis
- Tuberculous choroiditis
- Acquired syphilis

ATROPHIC MACULOPATHIES 326

Nonhereditary
- Dry age-related macular degeneration
- Myopic maculopathy

Hereditary
- End-stage Stargardt macular dystrophy
- Central areolar choroidal dystrophy
- Sorby macular dystrophy
- End-stage Best disease
- End-stage North Carolina macular dystrophy

BULL'S-EYE MACULOPATHIES 331

Signs

Causes
- Antimalarials
- Cone dystrophy
- Other causes

EXUDATIVE MACULOPATHIES 333
- Macular edema
- Serous detachment of the neuroretina
- Detachment of the RPE
- Associated with choroidal neovascularization
- Acute idiopathic maculopathy

FIBROTIC MACULOPATHIES 342
- Preretinal macular fibrosis
- Subretinal macular fibrosis

MACULAR HOLE 343
- Idiopathic macular hole
- Other macular holes
- Macular pseudohole

CRYSTALLINE MACULOPATHIES 344

COLORED MACULAR LESIONS 345

Egg-yolk lesions
- Best vitelliform macular dystrophy
- Adult vitelliform macular dystrophy

Cherry-red spot lesions

Dark lesions

SOLITARY FUNDUS LESIONS 348

Dark lesions
- Congenital RPE hypertrophy
- Choroidal naevus
- Choroidal melanoma
- RPE hamartoma
- Black 'sunburst' spots in non-proliferative sickle cell retinopathy

Pale lesions
- Old retinochoroiditis
- Idiopathic chorioretinal atrophy
- Coloboma of retina and choroid
- Congenital albinotic spots
- Choroidal metastatic tumor
- Retinal astrocytoma
- Early retinoblastoma

MULTIPLE YELLOW-WHITE FLECKS 354

Nonhereditary
- Hard drusen
- Soft drusen
- Cuticular (basal laminar) drusen
- Type II mesoangiocapillary membranoproliferative glomerulonephritis

Hereditary
- Familial dominant drusen in young individuals (Doyne honeycomb dystrophy)
- Malattia levantinese
- Fundus flavimaculatus
- Fundus albipunctatus
- Retinitis punctata albescens
- Pattern dystrophy
- Benign flecked retina syndrome
- Alport syndrome
- Early North Carolina macular dystrophy

DIFFUSE PIGMENT CLUMPING 362

Nonhereditary
- Age-related honeycomb (reticular) degeneration
- Advanced choroquine toxicity
- Related to retinal detachment
- Grouped congenital hypertrophy of the RPE
- Rubella retinopathy
- Congenital syphilis

Hereditary
- Typical retinitis pigmentosa
- Atypical retinitis pigmentosa
- Female carriers of choroideremia
- In eyes with angioid streaks

GENERALIZED WHITE FUNDUS 366

Nonhereditary
- Extensive retinal nerve fibre myelination
- High myopia
- Acute retinal ischemia
- Commotio retinae
- Large coloboma of retina and choroid

Hereditary
- Albinism
- Choroideremia
- Gyrate atrophy
- Diffuse choroidal atrophy
- Progressive bifocal chorioretinal atrophy

FOLDS AND LINEAR STREAKS 372
- Chorioretinal folds
- Lacquer cracks
- Pigment demarcation lines in retinal detachment
- Choroidal rupture
- Angioid streaks

RETINAL DETACHMENT 375
- Rhegmatogenous retinal detachment
- Tractional retinal detachment
- Exudative retinal detachment

Differential diagnosis of retinal detachment
- Degenerative retinoschisis
- Choroidal detachment
- Uveal effusion syndrome

POOR VISION BUT NORMAL FUNDUS 380

VASCULAR LESIONS

Hemorrhages

Preretinal hemorrhages

Signs

- Usually solitary and frequently located at the posterior pole, where it obscures all underlying retinal landmarks (**Fig. 12.1a**). On fluorescein angiography, the hemorrhage blocks retinal fluorescence throughout dye transit (**Fig. 12.1b–d**).
- Initially the hemorrhage is round, although later it may settle with gravity, giving rise to the typical boat-shaped crescentic configuration (**Fig. 12.2**).
- In some cases, a large hemorrhage may break through into the vitreous cavity (**Fig. 12.3**).
- Absorption usually occurs from above,causing the upper portion consisting of plasma to become yellow–white in color (**Fig. 12.4**).

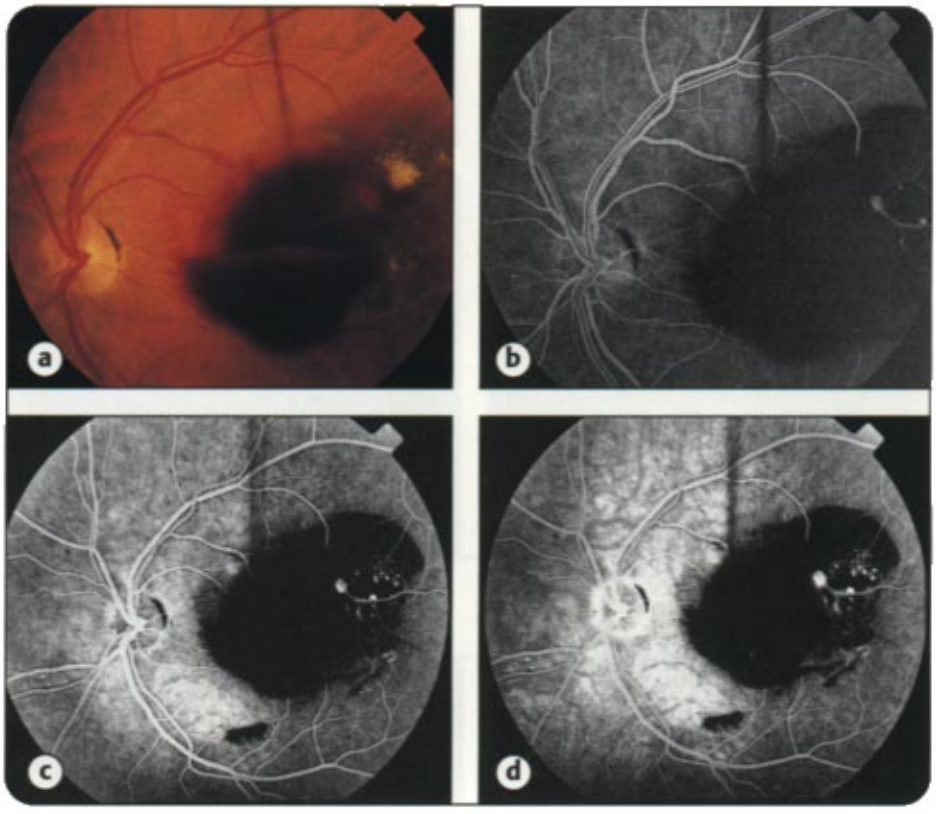

Fig. 12.1

Causes

- Proliferative retinopathies.
- Retinal artery macroaneurysm.
- Trauma.
- Subarachnoid hemorrhage (Terson syndrome).
- Valsalva retinopathy caused by a severe increase in venous pressure brought on by strenuous physical exertion.
- Nonaccidental injury ('shaken-baby' syndrome).

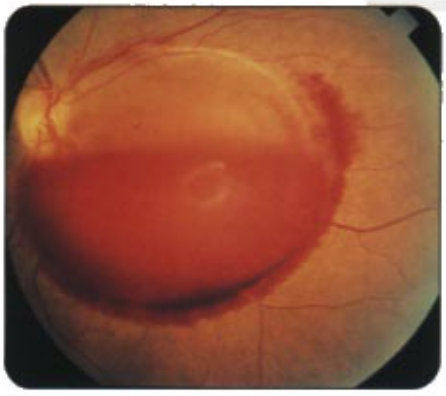

Fig. 12.2

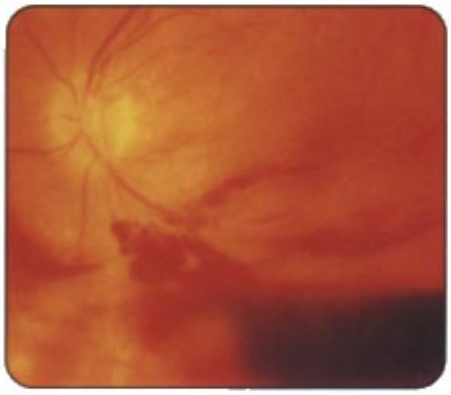

Fig. 12.3

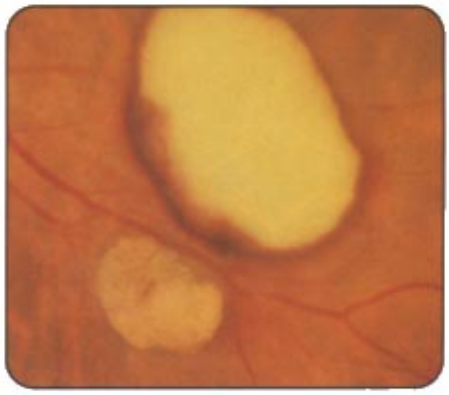

Fig. 12.4

Retinal flame-shaped hemorrhages

Signs

- Bright red, usually multiple, and located either in the retinal nerve fiber layer at the posterior pole or in relation to the optic nerve head, but seldom in the peripheral retina. On fluorescein angiography, the hemorrhages block background choroidal fluorescence throughout dye transit.

Causes

- Retinal vein occlusion.

Figure 12.5a–d shows a fresh left upper hemisphere retinal vein occlusion.
Figure 12.6a–d shows a fresh right inferotemporal branch retinal vein occlusion, with large cotton-wool spots.

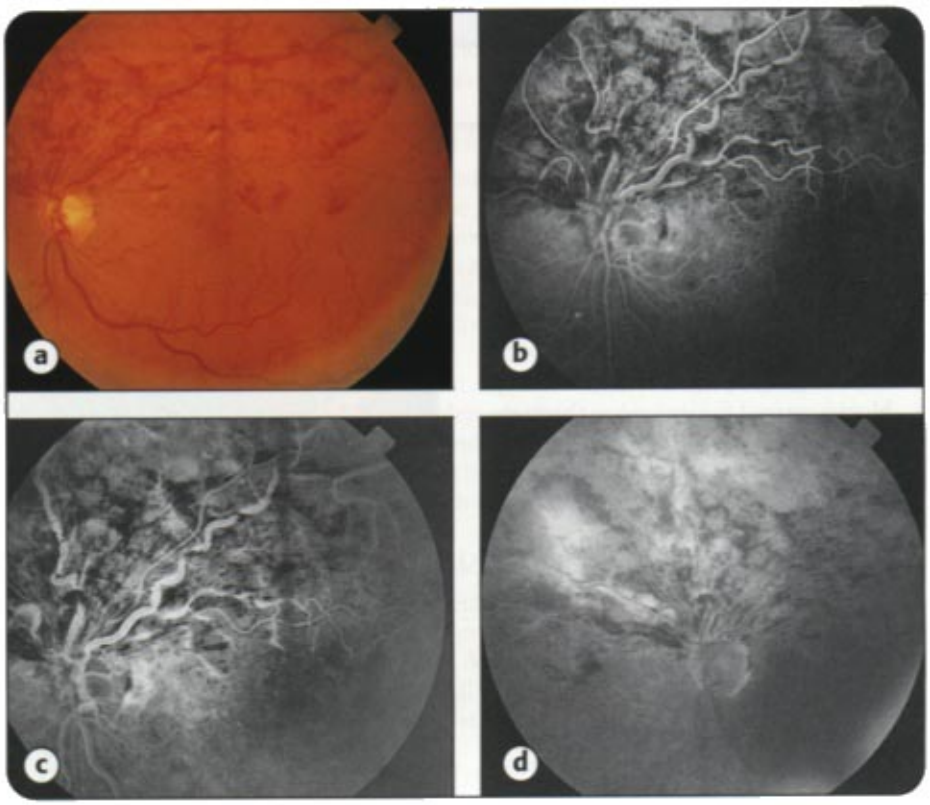

Fig. 12.5

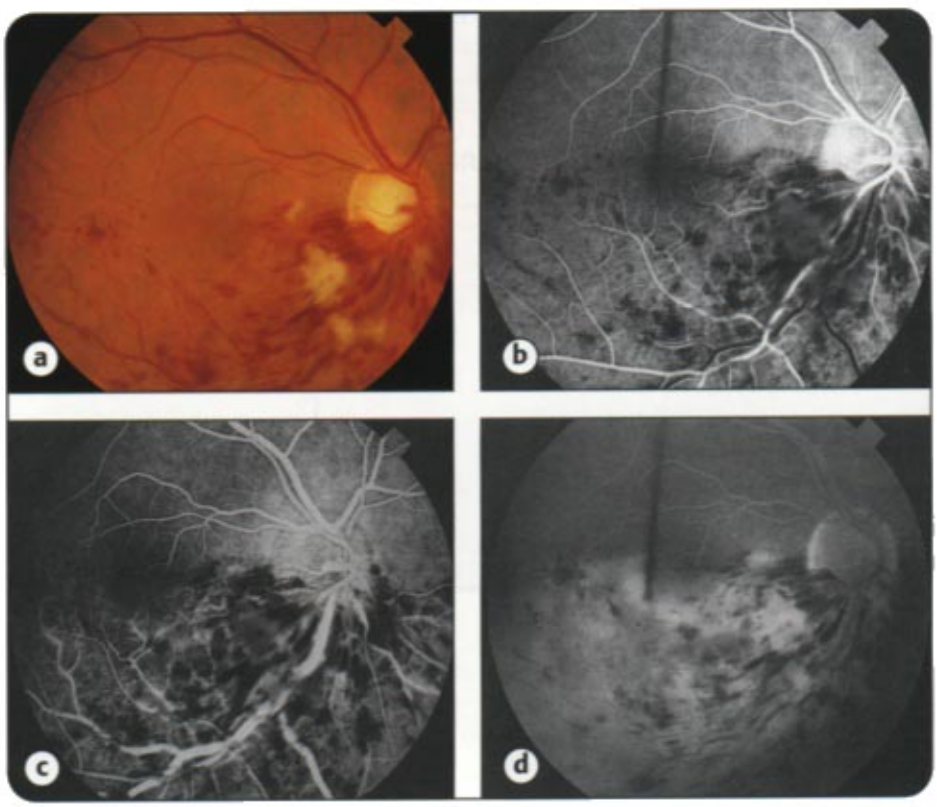

Fig. 12.6

Figure 12.7a shows a fresh non-ischemic central retinal vein occlusion in which the angiogram (**Fig. 12.7b–d**) shows leakage but good perfusion.
Figure 12.8a shows a fresh ischemic central retinal vein occlusion in which the angiogram (**Fig. 12.8b–d**) shows extensive areas of nonperfusion.

- Hypertensive retinopathy (**Fig. 12.9a and b**).
- Background diabetic retinopathy.
- Optic nerve disease – acute papilledema and anterior ischemic optic neuropathy (**Fig. 12.10**).
- Retinal periphlebitis (*see* Table 12.3) – **Fig. 12.11** shows flame-shaped and deep hemorrhages in cytomegalovirus retinitis.

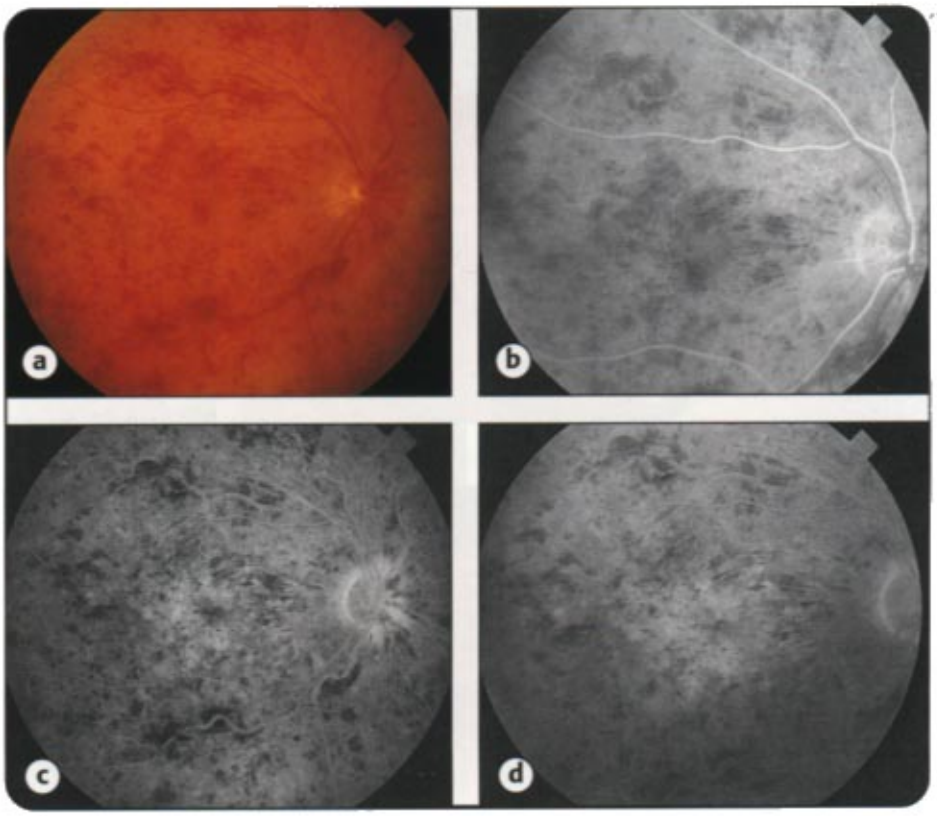

Fig. 12.7

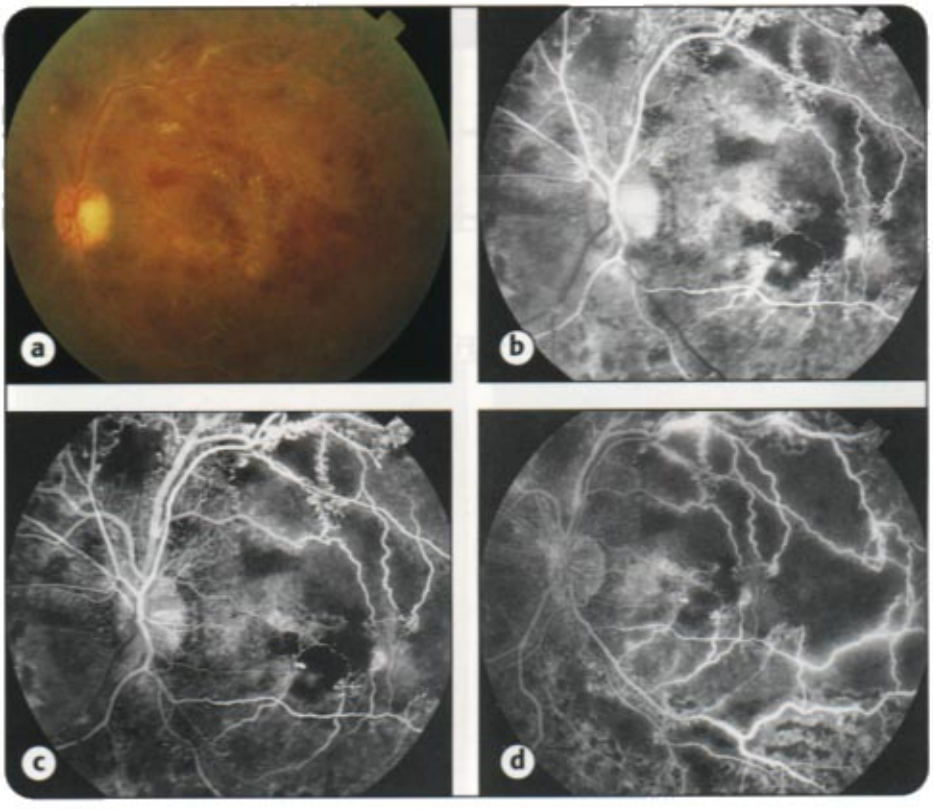

Fig. 12.8

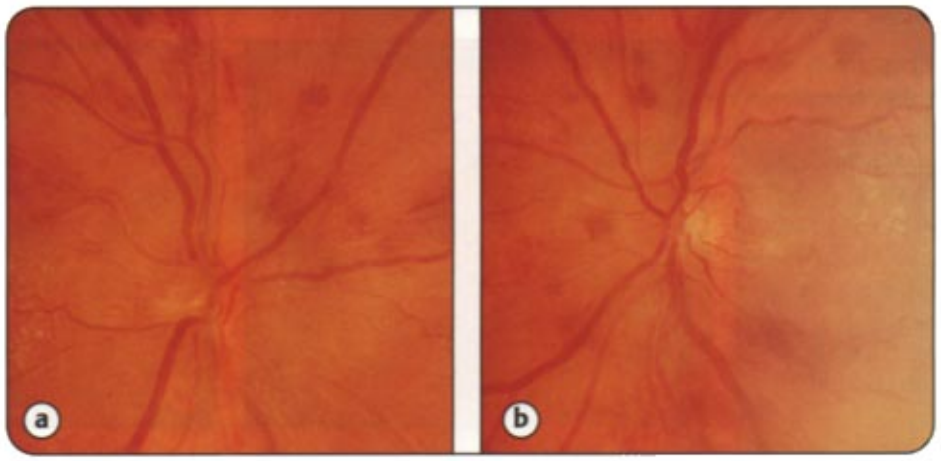

Fig. 12.9

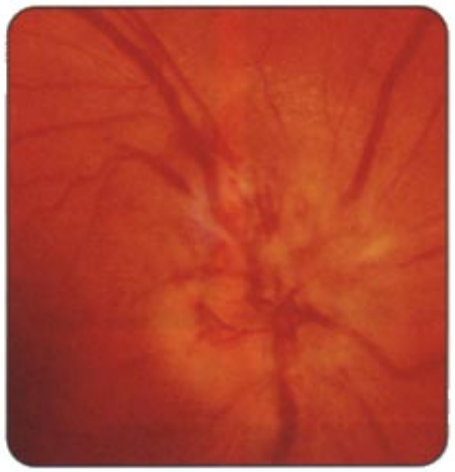

Fig. 12.10

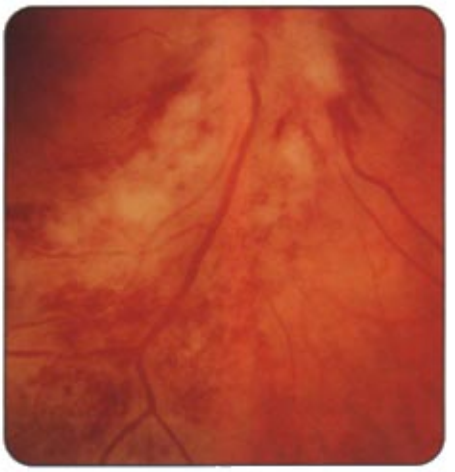

Fig. 12.11

Roth spots

Signs

- Hemorrhages with white centers (**Figs 12.12, 12.13, 12.14**).

Causes

- Severe anemias.
- Leukemias.
- Dysproteinemias.
- HIV retinopathy.
- Bacterial endocarditis.

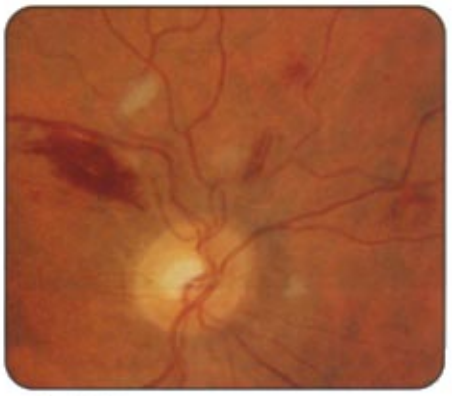

Fig. 12.12

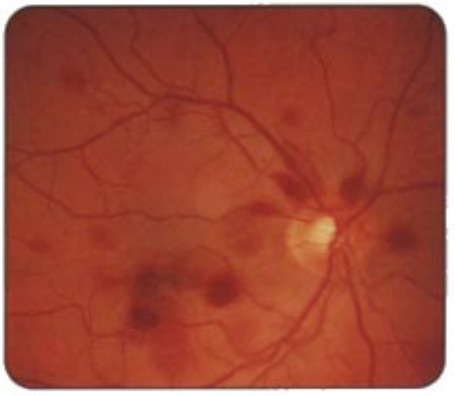

Fig. 12.13

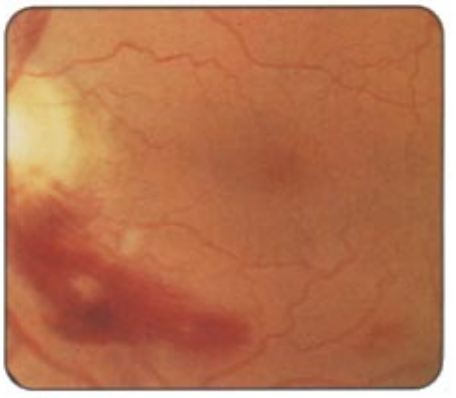

Fig. 12.14

Dot and blot hemorrhages

In dot and blot hemorrhages, the blood originates from the deep retinal capillaries.

Signs

- Dot hemorrhages are small, round, and of uniform density.
- Blot hemorrhages occupy the full thickness of the retina and are larger and darker.
- In the peripheral retina, the retinal nerve fiber layer is thin, so most retinal hemorrhages assume a dot and blot configuration.

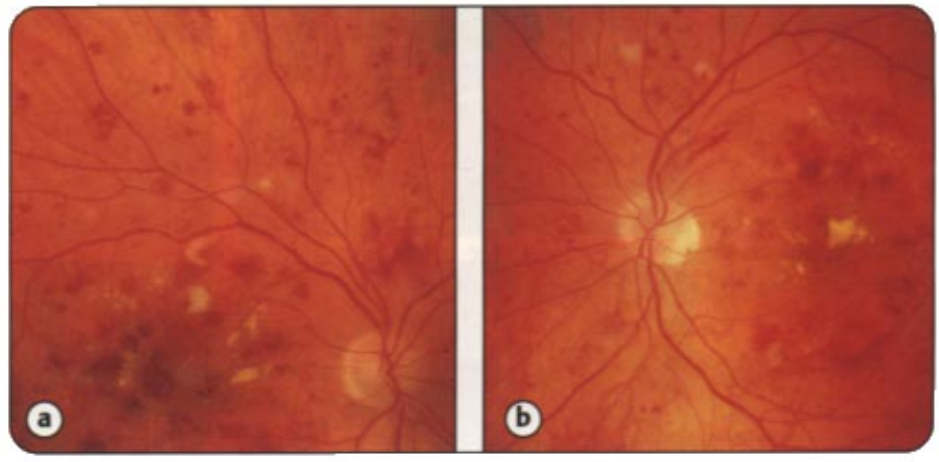

Fig. 12.15

Causes

- In background diabetic retinopathy, multiple dot and blot hemorrhages are seen at the posterior pole (**Figs 12.15a and b, 12.16a and b**).
- Large blotchy hemorrhages are usually indicative of retinal ischemia in retinal vein occlusion, preproliferative diabetic retinopathy, and slow-flow retinopathy (**Fig. 12.17a and b**).

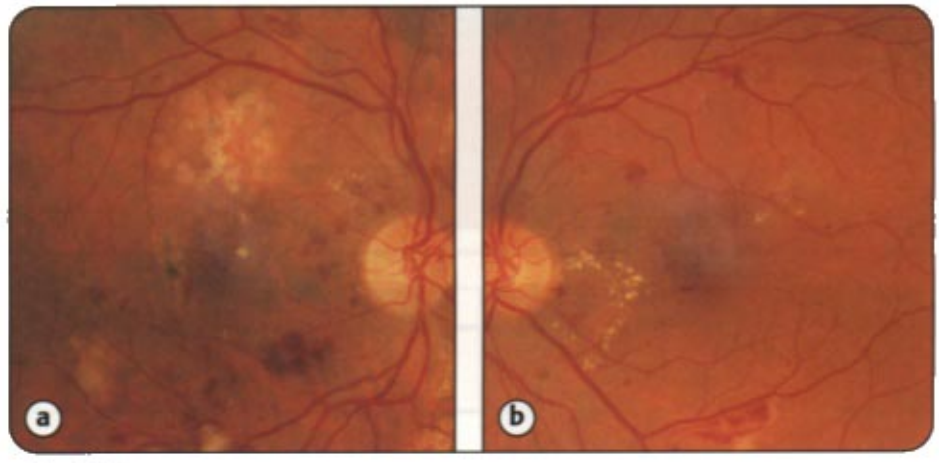

Fig. 12.16

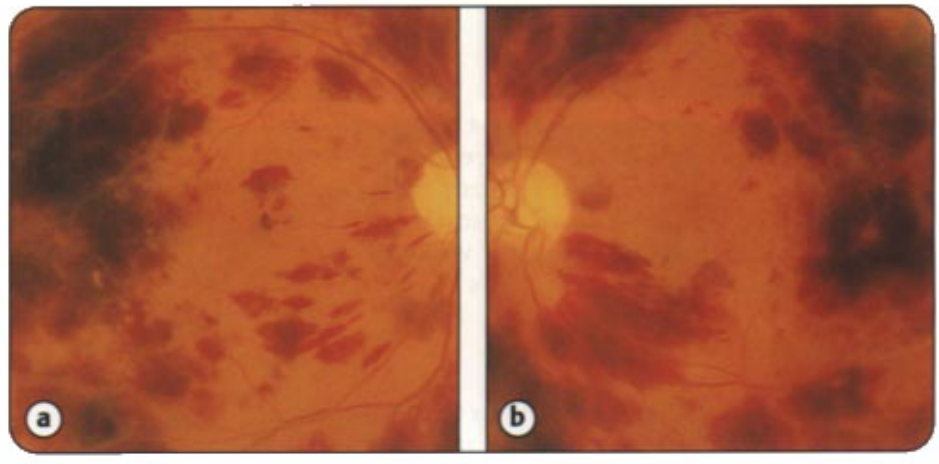

Fig. 12.17

Subretinal hemorrhages

In subretinal hemorrhages, the blood is located between the photoreceptors and the retinal pigment epithelium (RPE).

Signs

- Usually large and bright red with a relatively indistinct outline (**Figs 12.18, 12.19**).
- The retina overlying the hemorrhage is usually slightly elevated, but the retinal vessels are clearly seen.

Causes

- Choroidal neovascularization.
- Ruptured retinal artery macroaneurysm.
- Coats disease.
- Sickle cell anemia.
- Blunt ocular trauma.

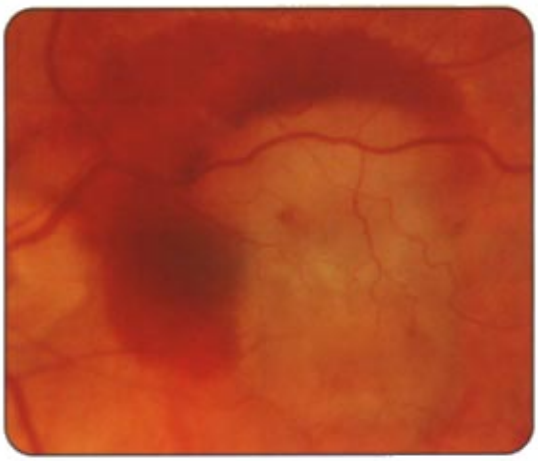

Fig. 12.18

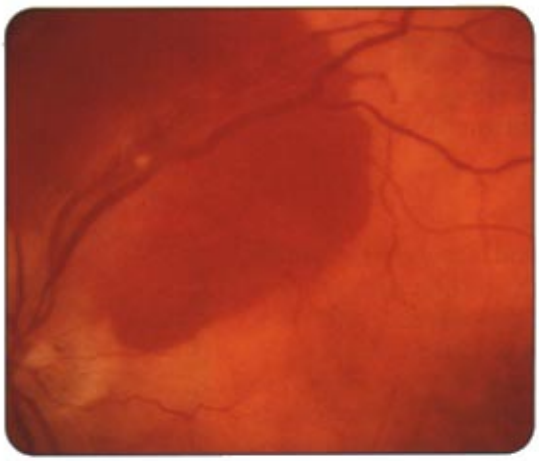

Fig. 12.19

Sub-RPE hemorrhages

In sub-RPE hemorrhages, the blood is derived from the choroid and enters the space between the RPE and Bruch membrane.

Signs

- Usually solitary, very dark-red, almost black, with a well-defined outline (**Fig. 12.20**). **Figure 12.21a** shows a partially absorbed sub-retinal pigment epithelial hemorrhage. The fluorescein angiogram (**Fig. 12.21b–d**) shows that the retinal circulation is well visualized but background choroidal fluorescence is blocked.

Causes

- Choroidal neovascularization is by far the most common (hemorrhagic pigment epithelial detachment).

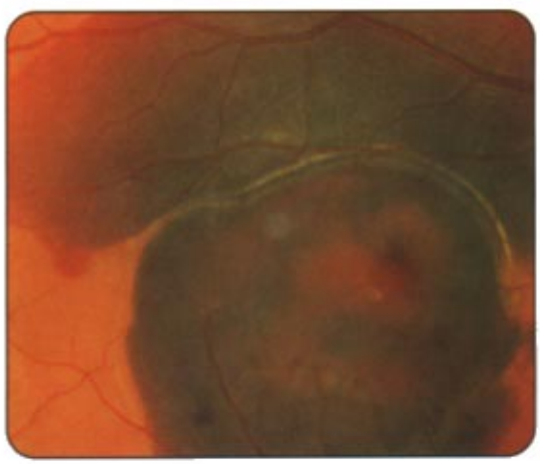

Fig. 12.20

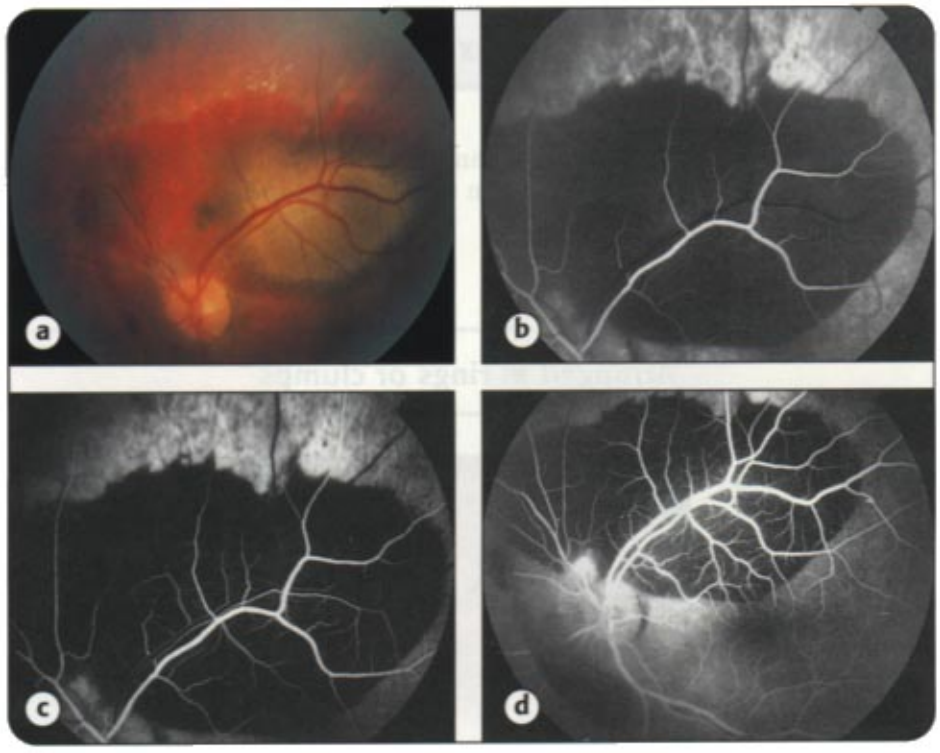

Fig. 12.21

Choroidal hemorrhages

Signs

- Dark red and frequently extensive.

Causes

- Blunt ocular trauma. **Figure 12.22** shows a choroidal rupture and hemorrhage.
- As a complication of drainage of subretinal fluid during retinal detachment surgery.

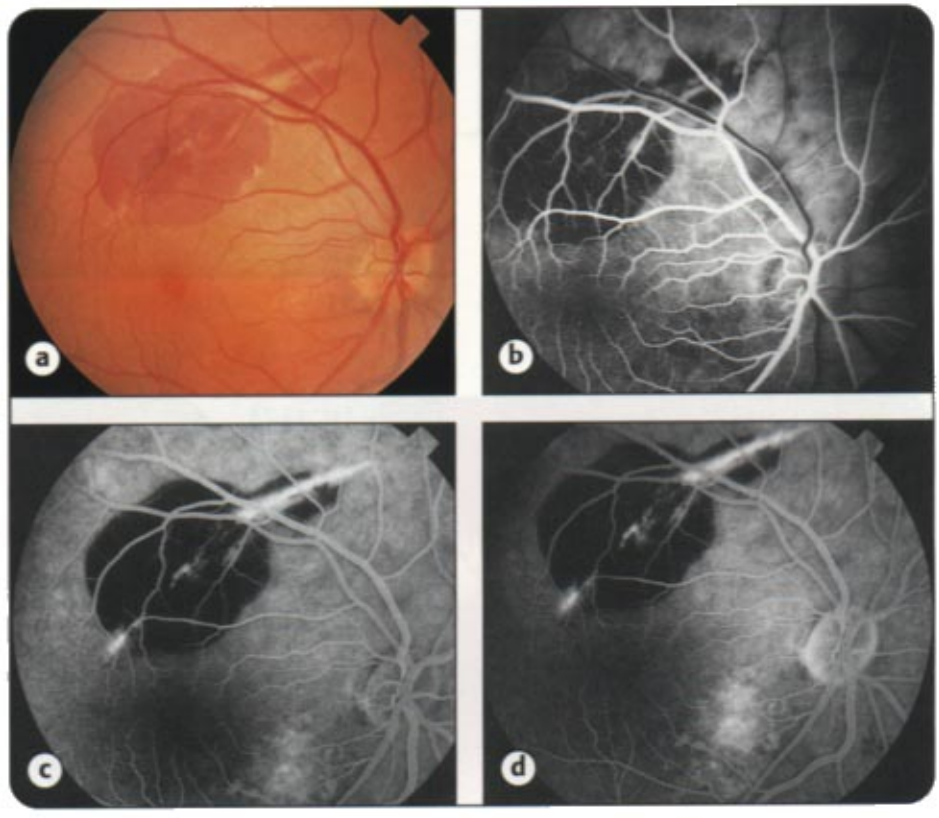

Fig. 12.22

Hard exudates

Hard exudates are yellow waxy plaques with relatively distinct margins which are most frequently seen at the posterior pole. They vary in size and have one of the following configurations:

Arranged in rings or clumps

Hard exudates that are arranged in rings or clumps are the result of chronic leakage from capillaries in the macular area.

Causes

- Diabetes – **Figure 12.23a** shows hard exudates in an eye with exudative maculopathy. On angiography the exudates cause a focal block of background fluorescence (**Fig. 12.23b–d**). Multiple microaneurysms are also seen on the angiogram.
- Old branch retinal vein occlusion (**Fig. 12.24**).
- Retinal artery macroaneurysm (**Fig. 12.25**).
- Radiation retinopathy (**Fig. 12.26**).
- Retinal telangiectasia (**Fig. 12.27**).

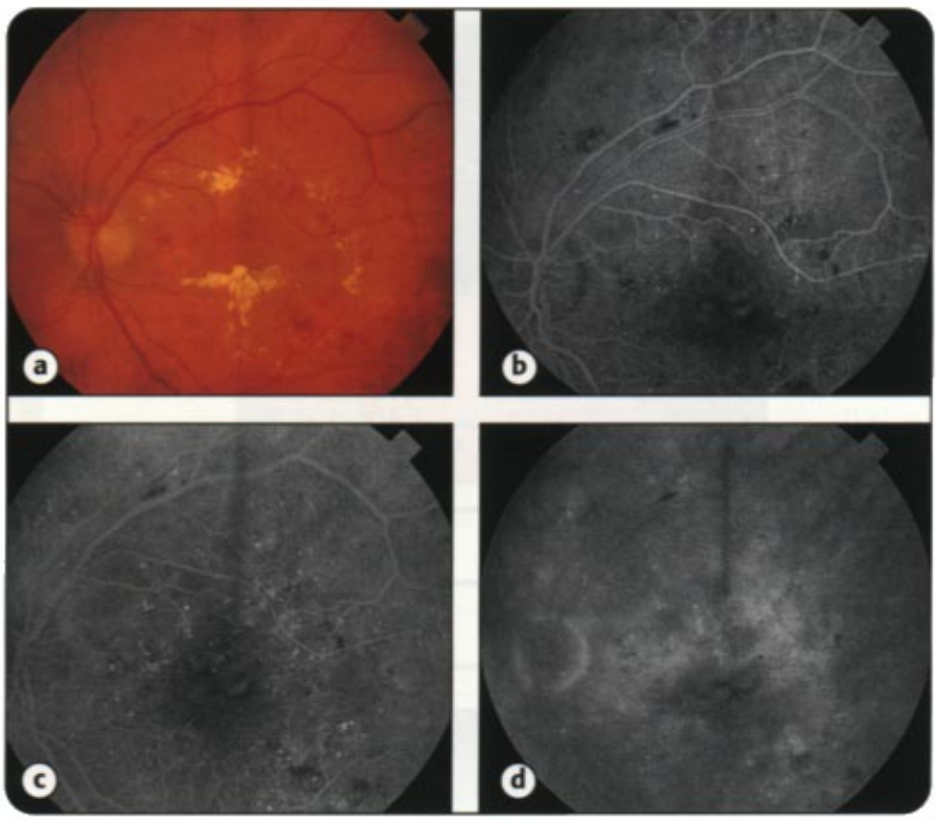

Fig. 12.23

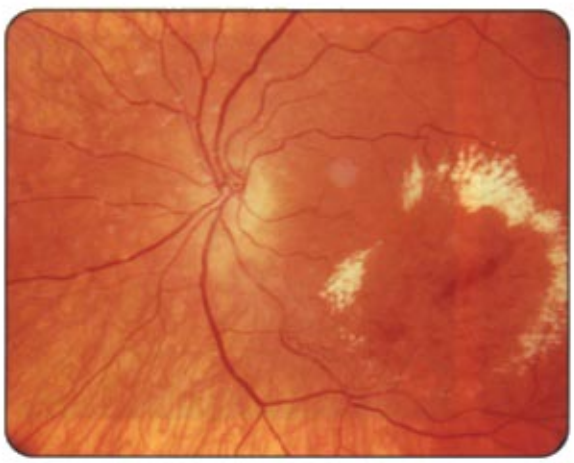

Fig. 12.24

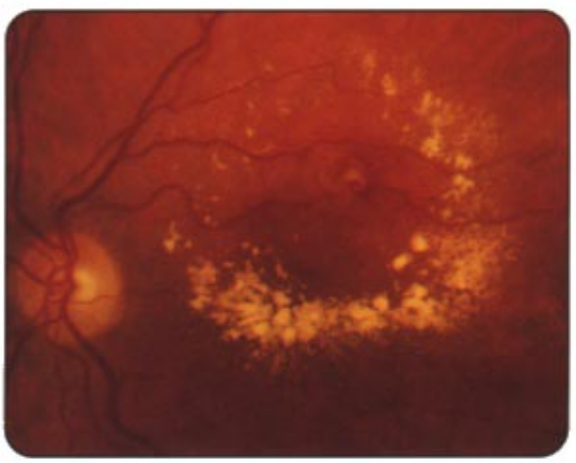

Fig. 12.25

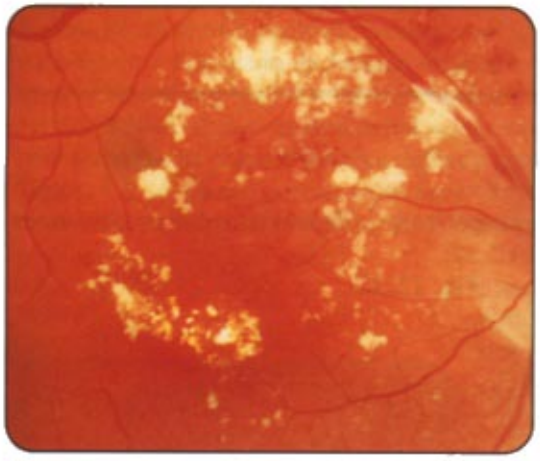

Fig. 12.26

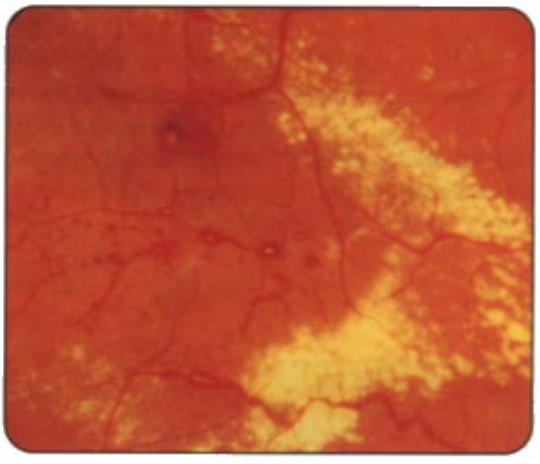

Fig. 12.27

Stellate maculopathy

Signs

- Retinal hard exudates forming either a complete or partial macular star.
- Frequently associated with disc swelling.

Causes

- Hypertension – bilateral (**Fig. 12.28a and b**).
- Papilledema – bilateral but may be asymmetric (**Fig. 12.29a and b**).
- Neuroretinitis – usually unilateral (**Fig. 12.30**).
- Capillary angioma, which may be either on the disc or in the periphery, is frequently associated with macular exudates.

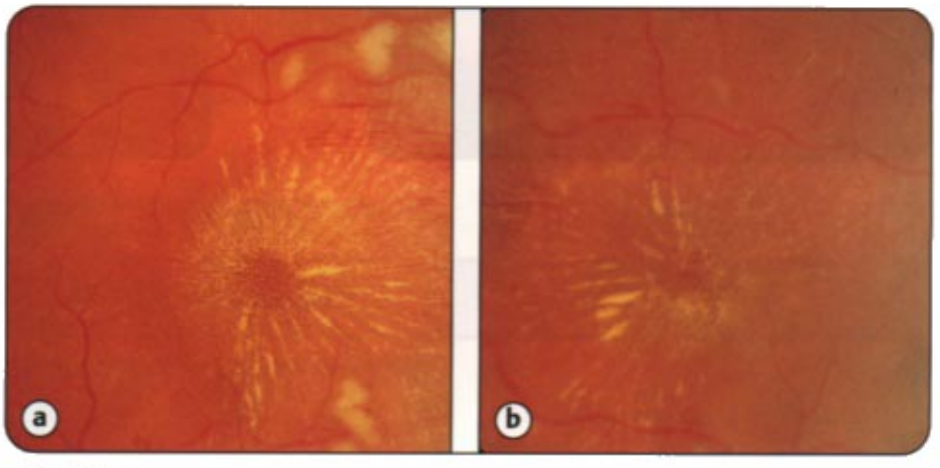

Fig. 12.28

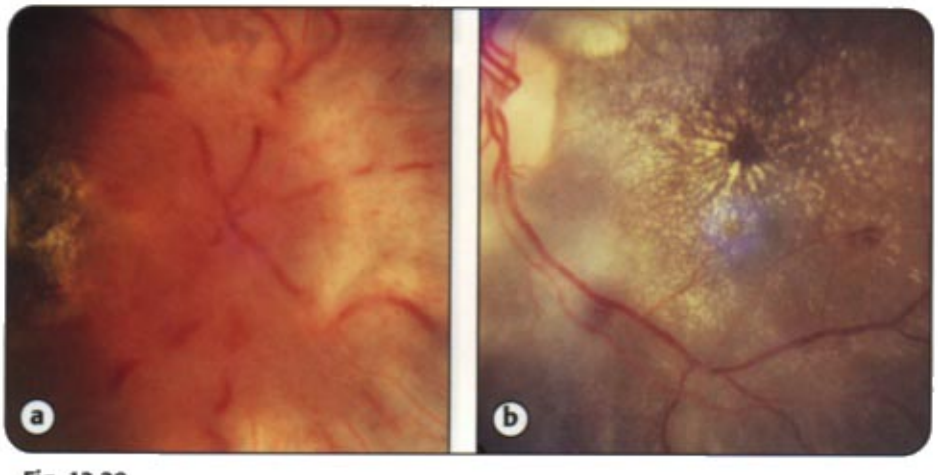

Fig. 12.29

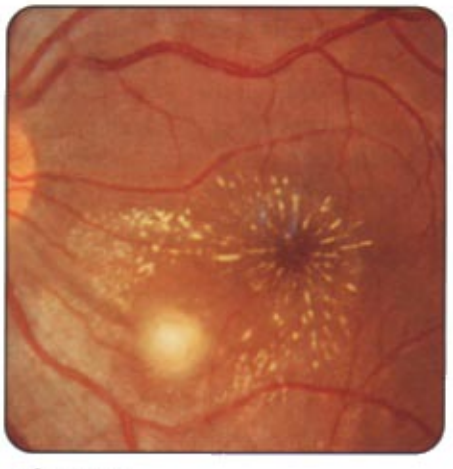

Fig. 12.30

Subretinal exudates

Subretinal exudates are frequently associated with serous elevation of the overlying retina.

Causes

- Chronic leakage from a choroidal neovascular membrane (**Fig. 12.31**).
- Coats disease (**Fig. 12.32**).
- *Toxocara canis*.

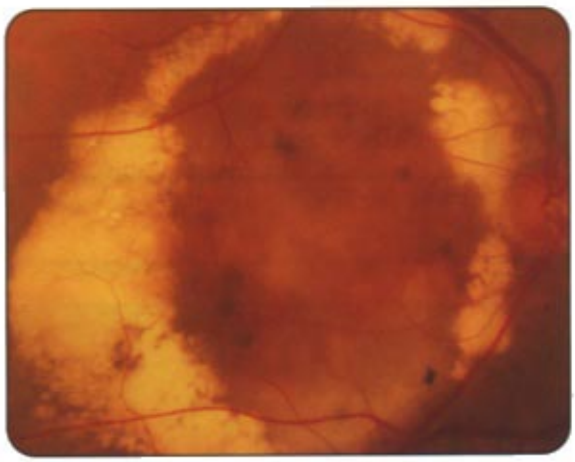

Fig. 12.31

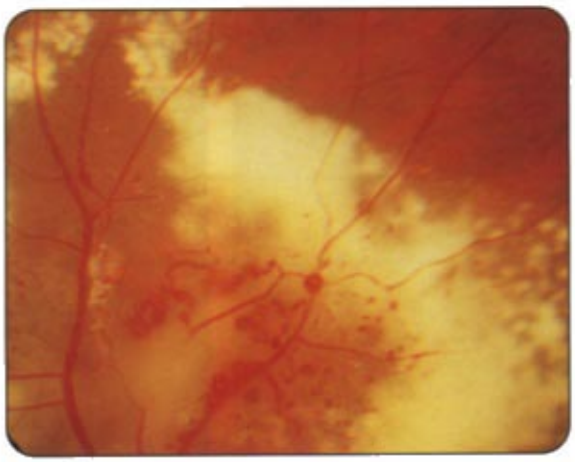

Fig. 12.32

Cotton-wool spots

Cotton-wool spots are caused by localized accumulation of axoplasmic debris in the nerve fiber layer secondary to ischemia.

Signs

- Small, white, superficial lesions with indistinct borders which may obscure a small underlying blood vessel (**Fig. 12.33a**). The fluorescein angiogram (**Fig. 12.33b–d**) of an eye with diabetic retinopathy shows focal areas of capillary nonperfusion associated with the cotton-wool spots.

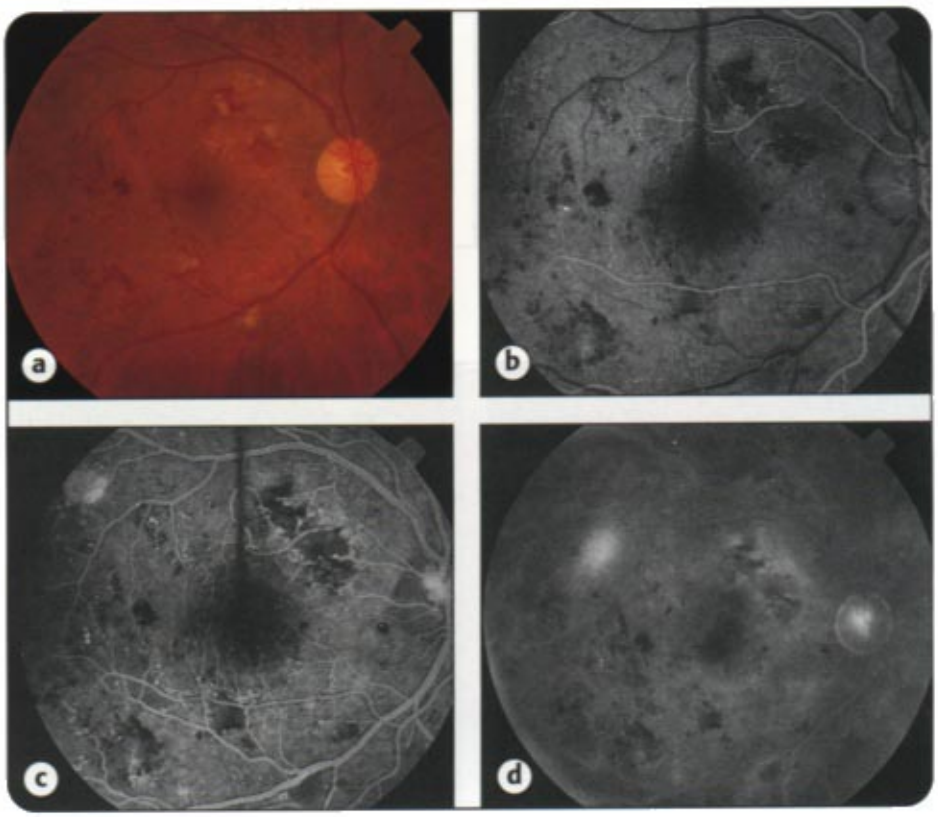

Fig. 12.33

Figure 12.34 shows multiple cotton-wool spots associated with venous dilatation and flame-shaped hemorrhages in a superior hemisphere retinal vein occlusion.
Figure 12.35 shows cotton-wool spots and blot retinal hemorrhages in preproliferative diabetic retinopathy.
Figure 12.36 shows cotton-wool spots and flame-shaped hemorrhages in severe hypertensive retinopathy.
Figure 12.37 shows extensive cotton-wool spots, some of which are confluent, in dermatomyositis.
Figure 12.38 shows multiple cotton-wool spots and a Roth spot in severe anemia.
Figure 12.39 shows multiple small cotton-wool spots in HIV microvasculopathy.
Figure 12.40 shows multiple cotton-wool spots in acute pancreatitis.

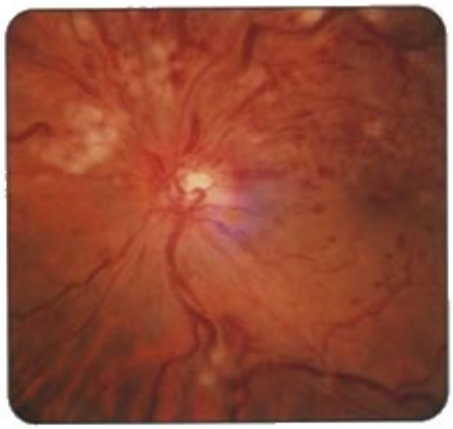

Fig. 12.34

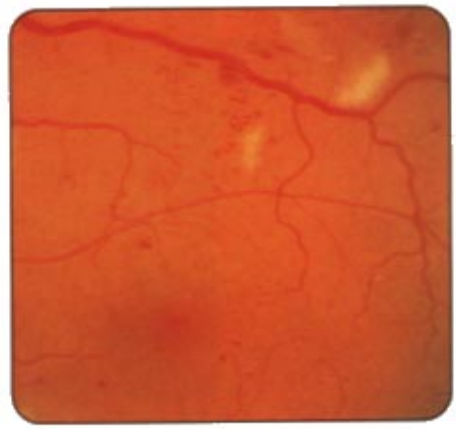

Fig. 12.35

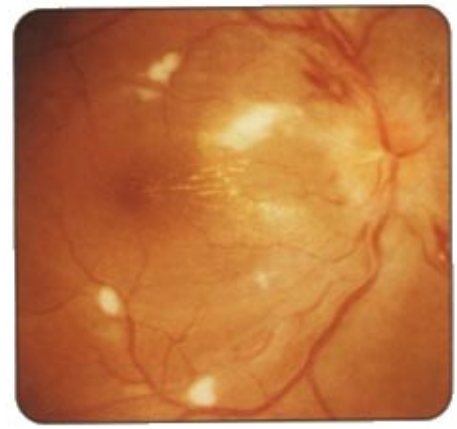

Fig. 12.36

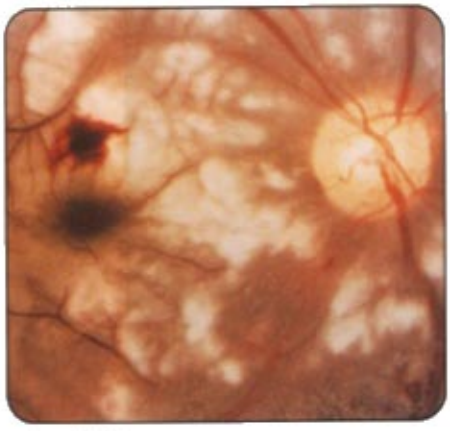

Fig. 12.37

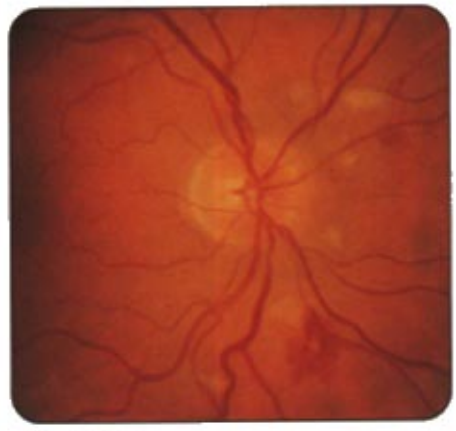

Fig. 12.38

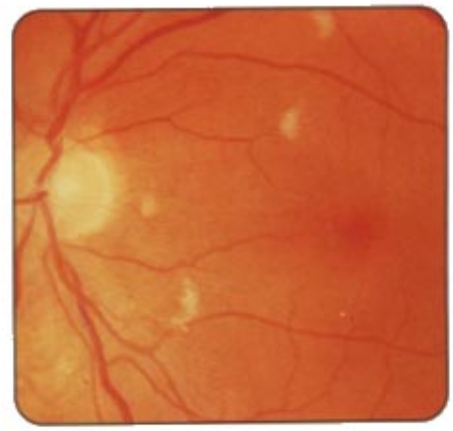

Fig. 12.39

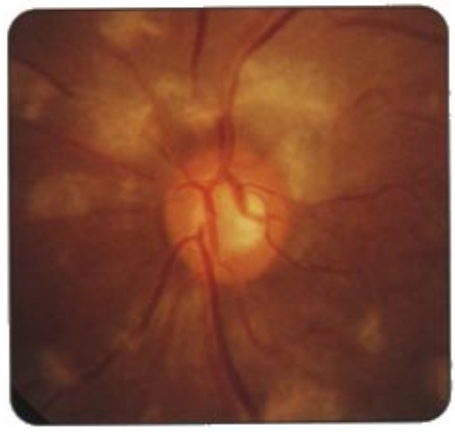

Fig. 12.40

Table 12.1 Causes of cotton-wool spots

Common
• retinal vein occlusion
• preproliferative diabetic retinopathy
• hypertension
• scleroderma
• systemic vasculitides
• HIV microvasculopathy
• microembolic retinal artery occlusion
Uncommon
• ocular ischemic syndromes
• hematological disorders
• antiphosphoid antibody syndrome
• trauma to chest and long bones (Purtscher retinopathy)
• acute pancreatitis

Differential diagnosis

- Congenital opaque nerve fibers – white striations of nerve fibers (**Figs 12.41, 12.42**).
- Retinitis – superficial yellow–white cloudy lesions associated with vitritis, which may be located adjacent to a pigmented scar (**Fig. 12.43**).

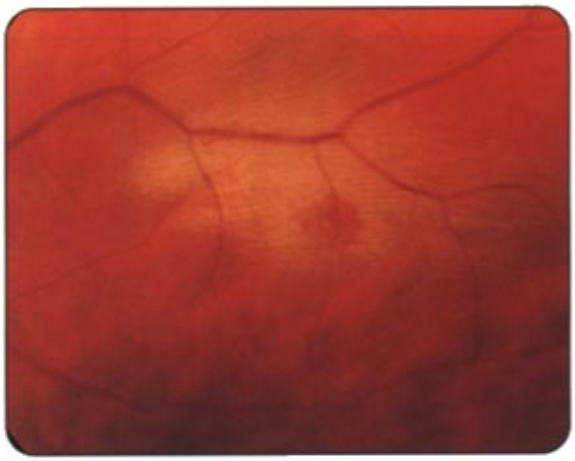

Fig. 12.41

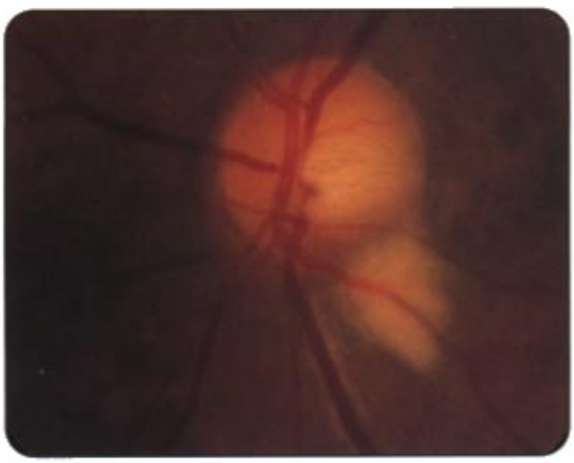

Fig. 12.42

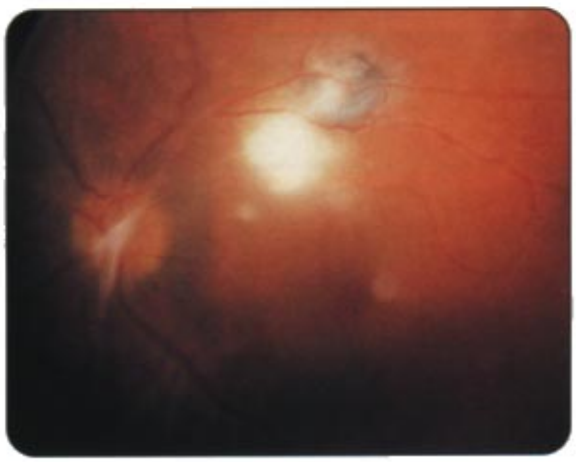

Fig. 12.43

Neovascularization

Signs

- Neovascularization may involve the optic nerve head.
- Neovascularization may involve the central retina along the major vascular arcades.
- Neovascularization may involve mainly the peripheral retina – where, it frequently assumes a 'seafan' configuration.
- The new vessels may be flat or elevated and they may be bare or associated with variable fibrosis.
- Contraction of fibrous tissue may give rise to tractional retinal detachment.

Figure 12.44 shows moderate disc neovascularization in proliferative diabetic retinopathy.
Figure 12.45 shows very severe disc new vessels in proliferative diabetic retinopathy.
Figure 12.46 shows retinal neovascularization following a right superotemporal branch vein occlusion.
Figure 12.47a–d shows peripheral 'seafan' neovascularization in proliferative sickle cell retinopathy. The new vessels leak during the late phase of the angiogram (**Fig. 12.47c and d**).
Figure 12.48 shows peripheral 'seafan' neovascularization in chronic myeloid leukemia.
Figure 12.49 shows severe retinal neovascularization and vascular occlusion in Eales disease.
Figure 12.50 shows fibrovascular proliferation along the superotemporal arcade in proliferative diabetic retionopathy.
Figure 12.51 shows peripheral fibrous proliferation in proliferative sickle cell retinopathy.

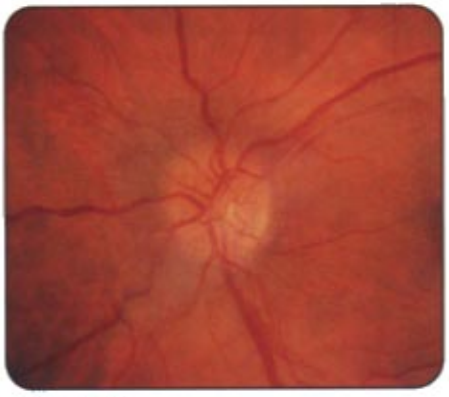

Fig. 12.44

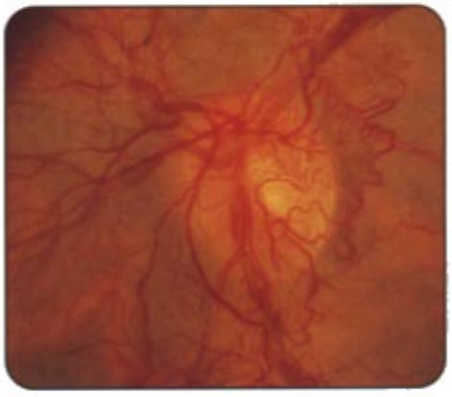

Fig. 12.45

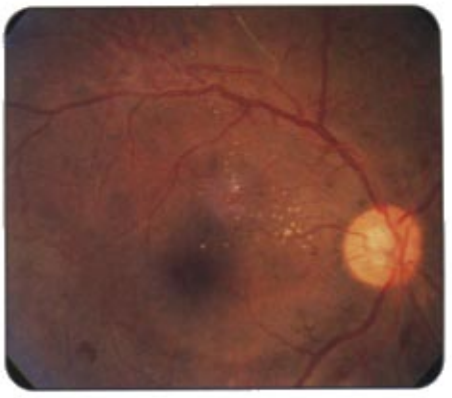

Fig. 12.46

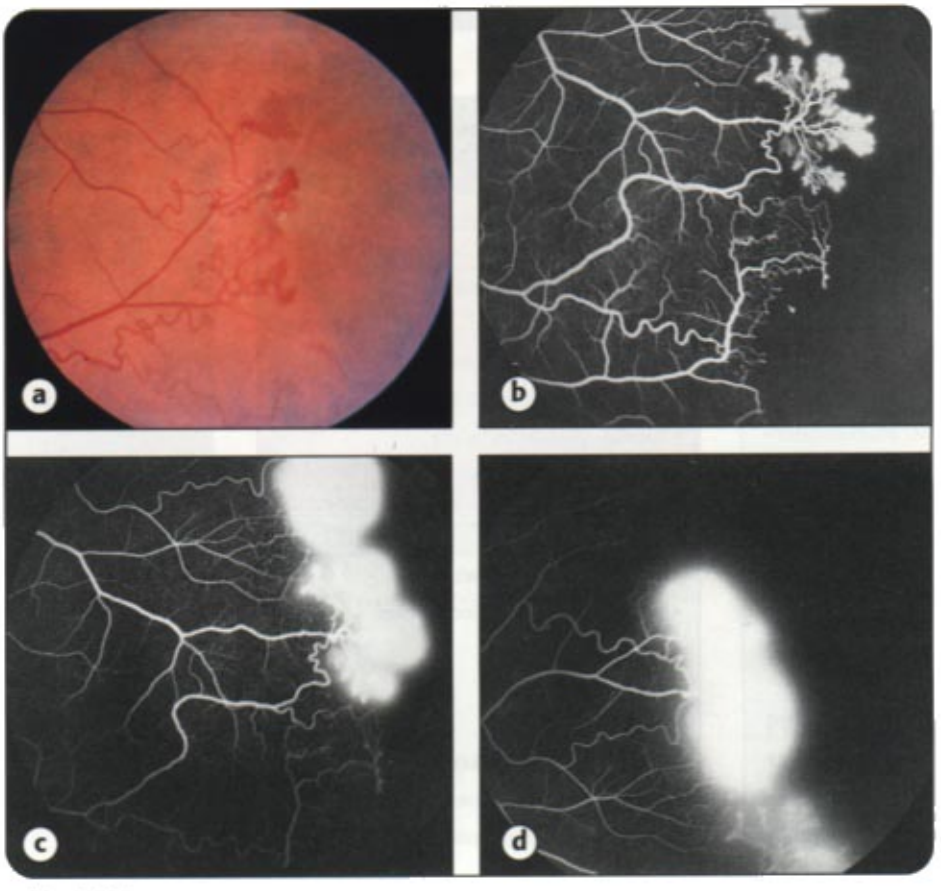

Fig. 12.47

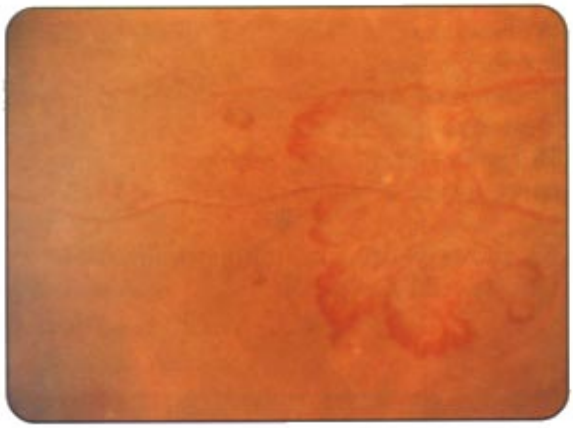

Fig. 12.48

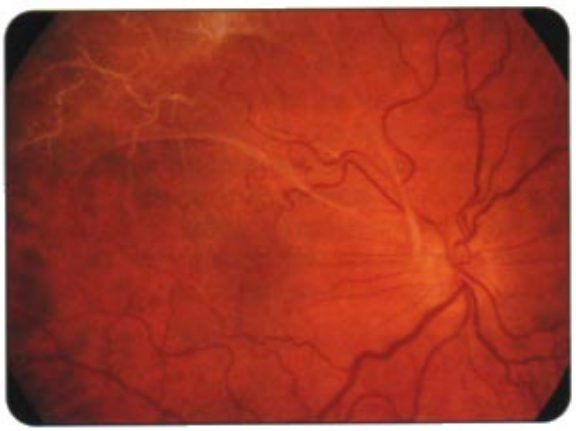

Fig. 12.49

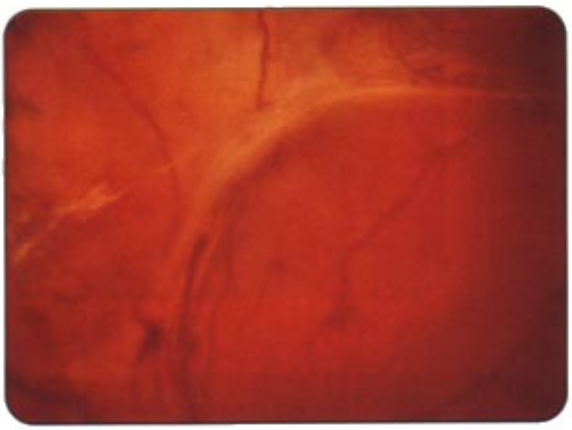

Fig. 12.50

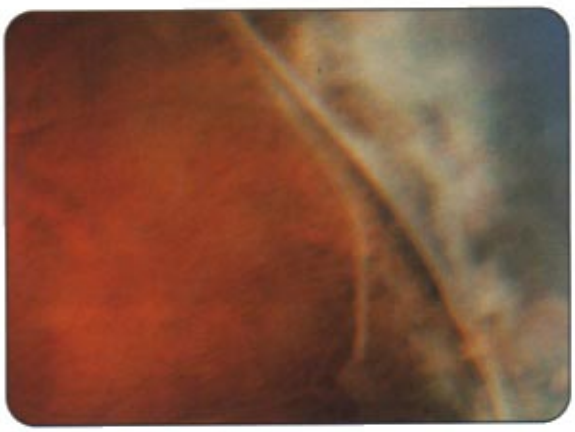

Fig. 12.51

Table 12.2 Causes of neovascularization

Central
• proliferative diabetic retinopathy
• old retinal vein occlusion
• retinal vasculitis
• retinal artery occlusion
• ocular ischemic syndromes
• carotid–cavernous fistula
• radiation retinopathy
Peripheral
• retinopathy of prematurity
• sickling hemoglobulinopathies
• Eales disease
• sarcoidosis
• intermediate uveitis
• chronic myeloid leukemia
• familial exudative vitreoretinopathy
• incontinentia pigmenti (Bloch–Sulzberger syndrome)

Retinal vasculitis

Signs

- Vasculitis may involve the retinal vein (periphlebitis) or, less commonly, the arterioles (periarteritis).
- Active vasculitis is characterized by a fluffy white haziness (cuffing) surrounding the blood column, which can be seen better on fluorescein angiography.
- Severe periphlebitis may give rise to branch vein occlusion, and severe periarteritis may result in permanent branch artery occlusion.

Figure 12.52 shows mild periphlebitis in sarcoidosis.
Figure 12.53 shows severe periphlebitis and 'candle-wax drippings' in sarcoidosis.
Figure 12.54 shows a fluorescein angiogram of periphlebitis in which there is staining of the vessel wall.
Figure 12.55 shows periphlebitis in cytomegalovirus retinitis.
Figure 12.56 shows extensive retinal arteriolar occlusion associated with periarteritis in polyarteritis nodosa.

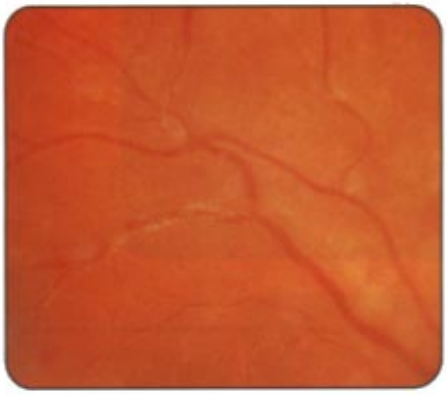

Fig. 12.52

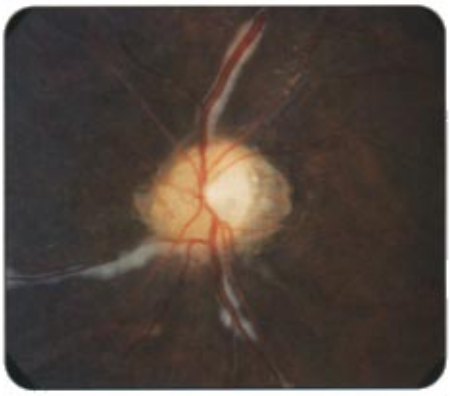

Fig. 12.53

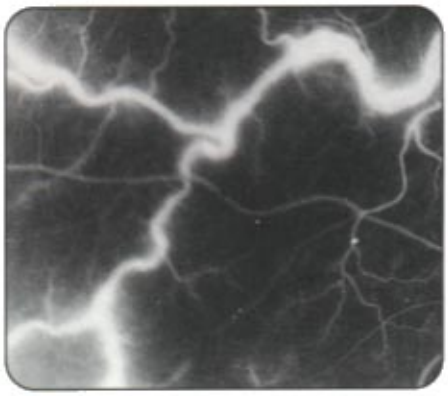

Fig. 12.54

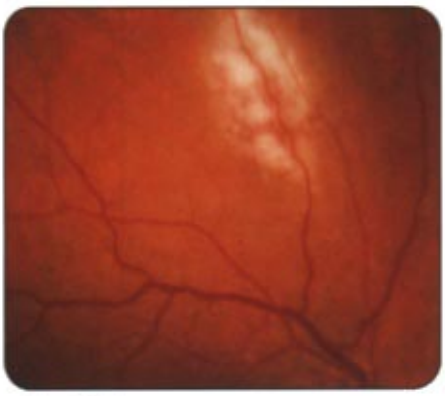

Fig. 12.55

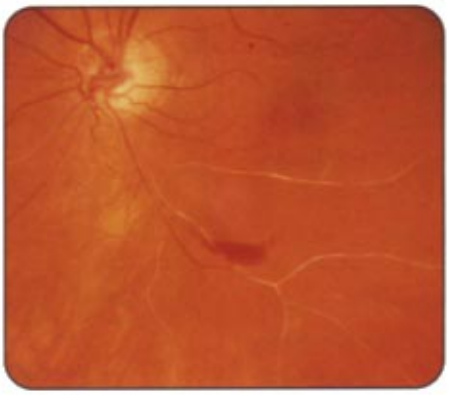

Fig. 12.56

Table 12.3 Causes of retinal vasculitis

Periphlebitis
• sarcoidosis
• Behçet disease
• cytomegalovirus retinitis
• acute retinal necrosis
• intermediate uveitis
• birdshot retinochoroidopathy
• multiple sclerosis
• tuberculosis
• Eales disease
• frosted branch angiitis
• idiopathic (primary)
Periarteritis
• systemic lupus erythematosus
• dermatomyositis
• polyarteritis nodosa
• Wegener granulomatosis

Retinal emboli

Fibrinoplatelet emboli

Fibrinoplatelets are most commonly associated with carotid disease and cause amaurosis fugax. Occasionally they may give rise to major arterial occlusion.

Signs

- Dull gray, elongated particles which are usually multiple (**Fig. 12.57**).
- Occasionally they fill the entire lumen of an arteriole (**Fig. 12.58**).

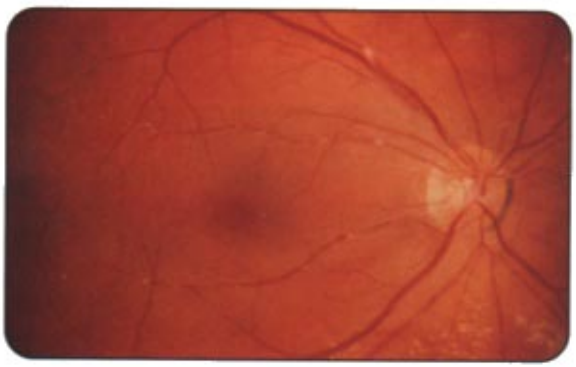

Fig. 12.57

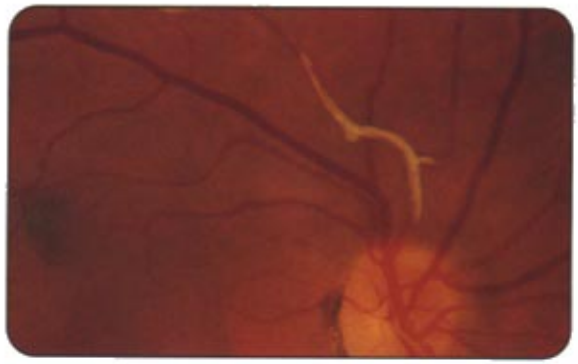

Fig. 12.58

Cholesterol (Hollenhorst) plaques

Cholesterol plaques are associated with carotid artery disease but seldom cause symptoms.

Signs

- Small, glistening, usually multiple particles typically located at bifurcations of vessels (**Fig. 12.59**).

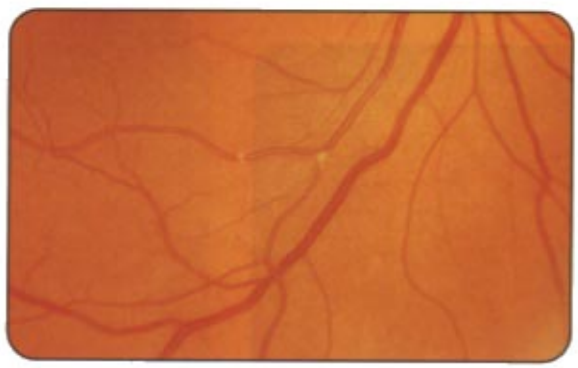

Fig. 12.59

Calcific emboli

Calcific emboli may be associated with cardiac valvular disease and may give rise to major arterial occlusion.

Signs

- White, solitary particle which is most commonly located on the disc (**Fig. 12.60**).

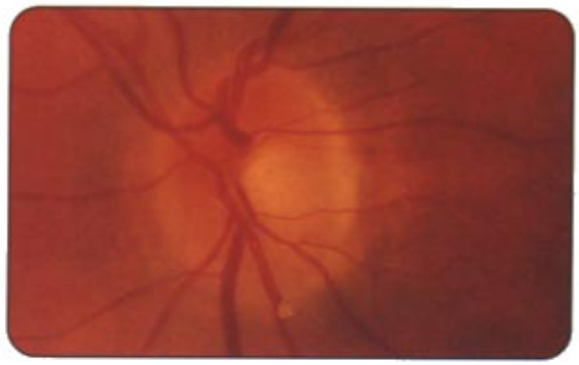

Fig. 12.60

Miscellaneous emboli

- Heart valve vegetations in bacterial endocarditis.
- Myxomatous material from an atrial myxoma.
- Fat emboli seen in trauma to the chest and long bones (Purscher retinopathy) (**Fig. 12.61**).
- Fat emboli in acute pancreatitis (*see* **Fig. 12.40**).
- Talc emboli in intravenous drug abusers.
- Metastatic tumor.
- Amniotic fluid.

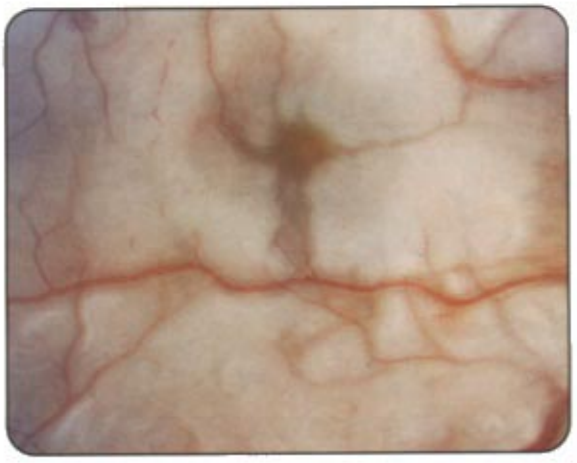

Fig. 12.61

Major changes in vascular caliber

Table 12.4 Causes of major changes in vascular caliber

Arterial attenuation	Venous dilatation and/or tortuosity
• systemic hypertension	• incipient central retinal vein occlusion
• retinal artery occlusion	• retinal vein occlusion
• severe diffuse retinal disease	• preproliferative diabetic retinopathy
• rare systemic syndromes (e.g. Batten–Vogt, Zellweger)	• hyperviscosity syndrome
	• ocular ischemic syndrome
	• carotid–cavernous fistula
Combined venous and arterial dilatation and tortuosity	• inherited venous beading
	• primary antiphospholipid antibody syndrome
• nanophthalmos	• increased intraorbital pressure
• retinopathy of prematurity (plus disease)	• Fabry disease
• retinal capillary hemangioma	• mannosidosis
• retinal racemose hemangioma	
• Moroteaux–Lamy syndrome	

Figure 12.62 shows arteriolar attenuation in advanced retinitis pigmentosa.
Figure 12.63 shows venous beading and dilatation, as well as disc new vessels, in diabetic retinopathy.
Figure 12.64a and b shows arterial tortuosity in inherited venous beading.
Figure 12.65 shows arterial and venous tortuosity in racemose hemangioma.

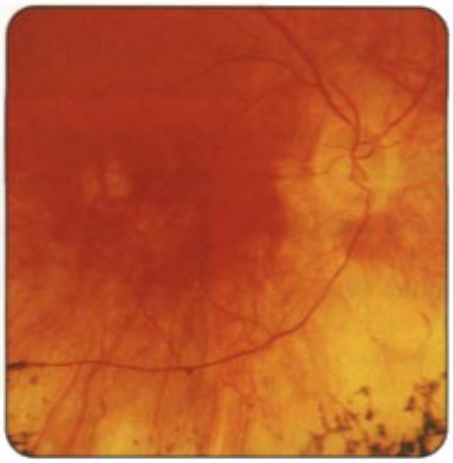

Fig. 12.62

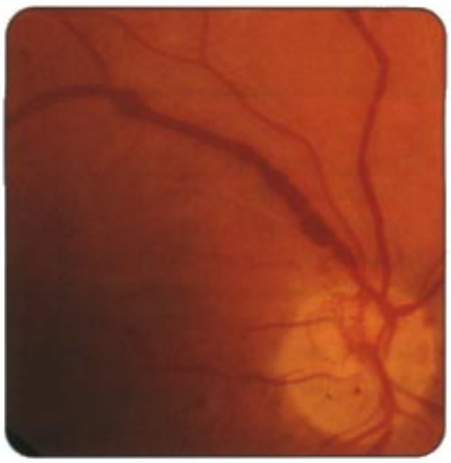

Fig. 12.63

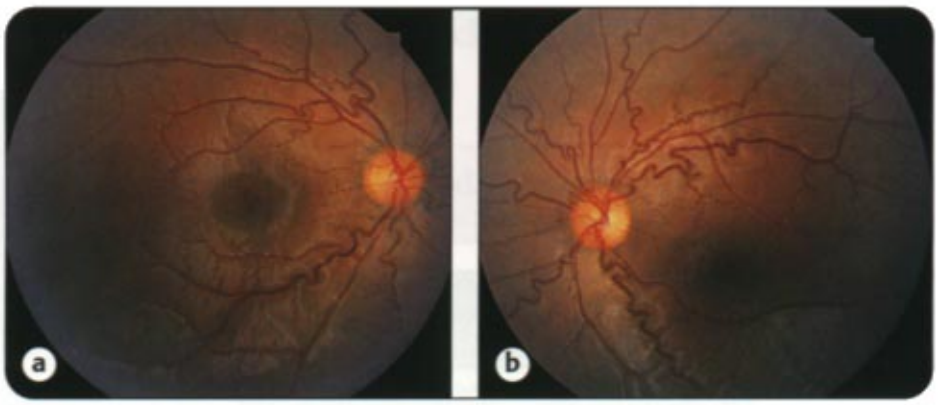

Fig. 12.64

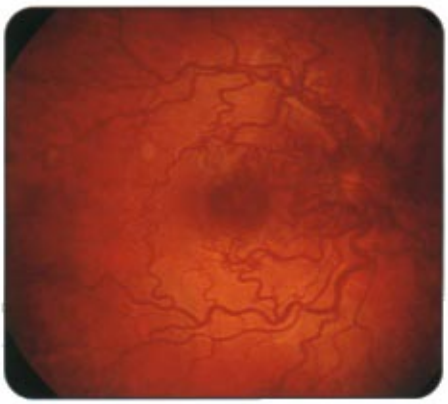

Fig. 12.65

Major vascular malformations

Retinal artery macroaneurysm

Retinal artery macroaneurysm is an uncommon, usually unilateral condition which typically affects elderly females with hypertension.

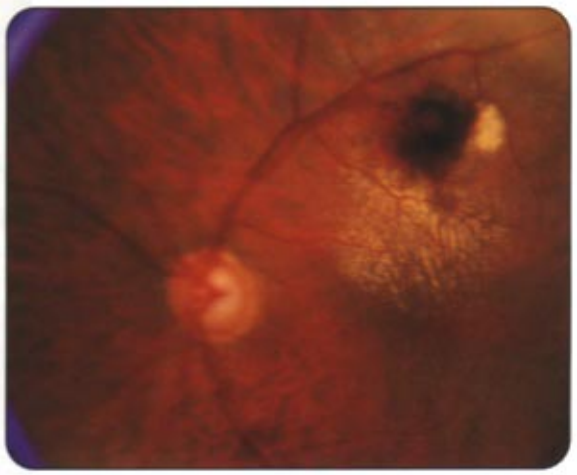

Fig. 12.66

Signs

- Localized fusiform or saccular dilatation usually arising within the first three orders of bifurcation.
- More than one lesion is seen in the same eye in about 20% of cases.

Complications

- Hard exudate formation and hemorrhage (**Fig. 12.66**).
- Rarely, secondary retinal branch vein occlusion.

Leber miliary aneurysms

Leber miliary aneurysm is a rare, unilateral, developmental vascular anomaly that usually presents in adult life. It is thought to represent a milder form of Coats disease.

Signs

- Fusiform and saccular vascular dilatations, mainly affecting the temporal periphery (**Fig. 12.67**).

Complications

- Hard exudate formation and hemorrhage.

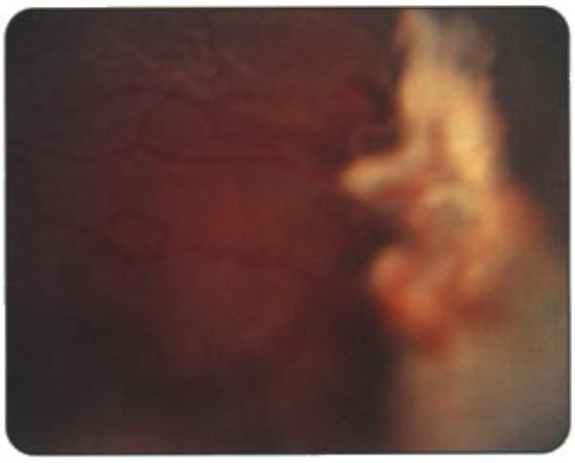

Fig. 12.67

Coats disease

Coats disease is a rare, severe, unilateral, developmental vascular anomaly which typically presents at about the age of 8 years with leukocoria (white pupil) or strabismus. It is more common in boys than in girls.

Signs

- Vascular dilatation, tortuosity, and aneurysm formation at the posterior pole and periphery (**Fig. 12.68**).
- Extensive subretinal exudation (**Fig. 12.69**).

Complications

- Exudative retinal detachment and a retrolental mass which may give rise to leukocoria.
- Cataract.
- Rubeosis iridis.

Differential diagnosis

- Late-onset retinoblastoma.
- *Toxocara canis*, which may also be associated with hard exudate formation.

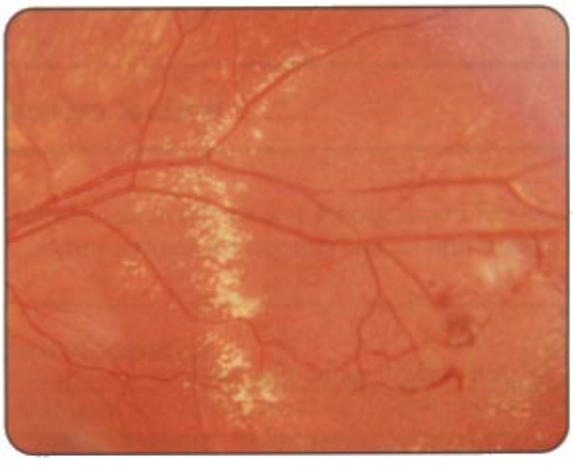

Fig. 12.68

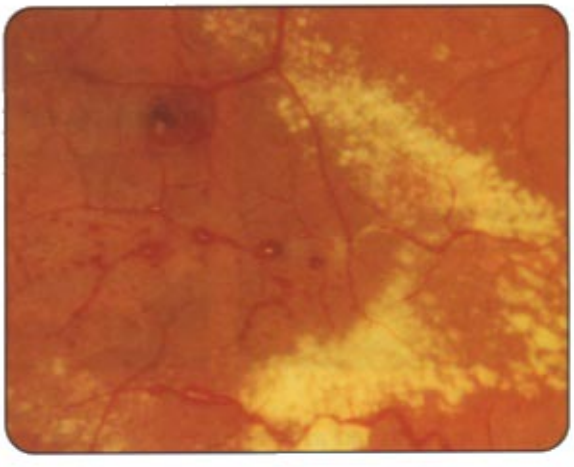

Fig. 12.69

Retinal capillary hemangioma

Retinal capillary hemangioma is an uncommon, benign tumor which may be multiple. Both eyes are affected in 50% of cases. About 25% of patients have systemic lesions (von Hippel–Lindau syndrome).

Signs (in chronological order)

- Small red nodule between an arteriole and venule (**Fig. 12.70**).
- Large round orange–red tumor associated with dilated and tortuous artery and vein (**Fig. 12.71**).

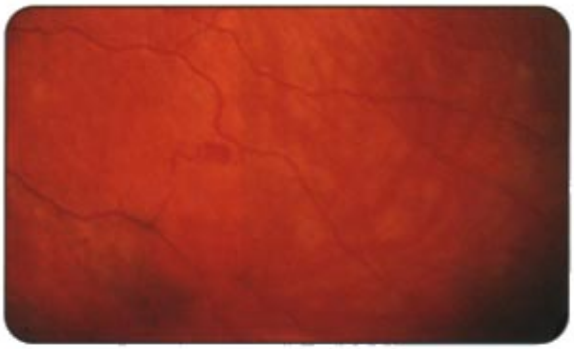

Fig. 12.70

Complications

- Hard exudates near the lesion and at the macula.
- Vitreous hemorrhage.
- Exudative retinal detachment.

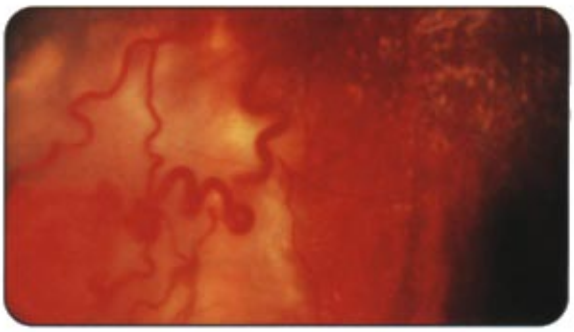

Fig. 12.71

Retinal cavernous hemangioma

Retinal cavernous hemangioma is a very rare, congenital, unilateral, asymptomatic and innocuous, benign tumor.

Signs

- Clumps of thin-walled, grape-like saccular aneurysms filled with blood, which may occasionally leak (**Fig. 12.72**).

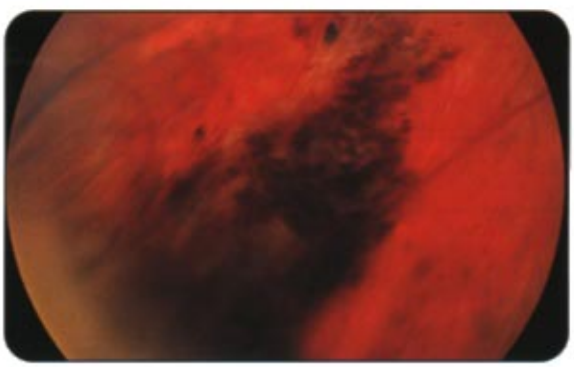

Fig. 12.72

Retinal racemose hemangioma

Retinal racemose hemangioma is a very rare, usually unilateral, congenital arteriovenous malformation that may occur in isolation or in association with systemic lesions (Wyburn–Mason syndrome).

Signs

- Grossly dilated and tortuous arteries and veins which are more numerous than in a normal fundus (**Fig. 12.73**; *also see* **Fig. 12.65**).

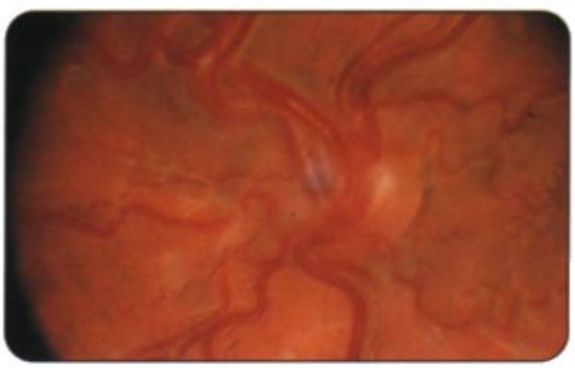

Fig. 12.73

INFLAMMATORY LESIONS

Idiopathic multifocal white dot syndromes

Multiple evanescent white dot syndrome

Multiple evanescent white dot syndrome (MEWDS) is a rare, usually unilateral condition that typically affects young women and has an excellent prognosis.

Signs

- Mild vitritis.
- Numerous, very small white dots at the level of the RPE most prominent at the posterior pole and mid-periphery (**Fig. 12.74a**) which are better visualized on fluorescein angiogram (**Fig. 12.74b**).
- Orange granularity around the fovea.
- Enlarged blind spot.

Residua

- The blind spot may remain enlarged and the macula may remain abnormal but visual acuity is normal.

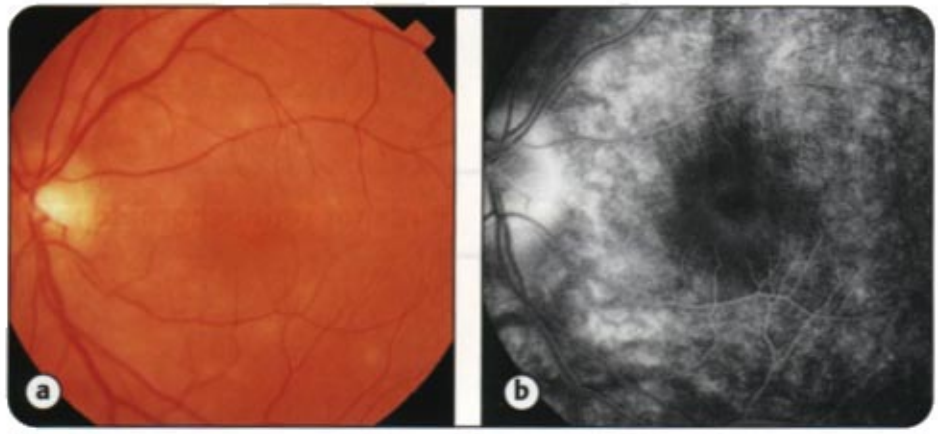

Fig. 12.74

Punctate inner choroidopathy

Punctate inner choroidopathy (PIC) is an uncommon, usually eventually bilateral condition that typically affects young women with myopia and has a guarded prognosis.

Signs

- No vitritis.
- Small, yellow, indistinct choroidal lesions all of same age at the posterior pole (**Fig. 12.75a and b**).
- A serous retinal detachment may be associated with plentiful lesions.

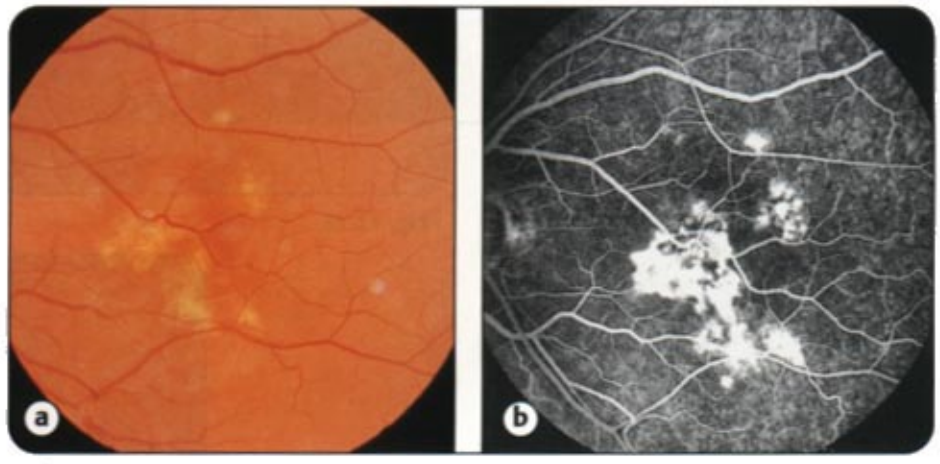

Fig. 12.75

Residua

- Sharply demarcated, dumb-bell-shaped choroidal scars (**Fig. 12.76a and b**).
- Occasional choroidal neovascularization (*see* **Fig. 12.181a–d**).

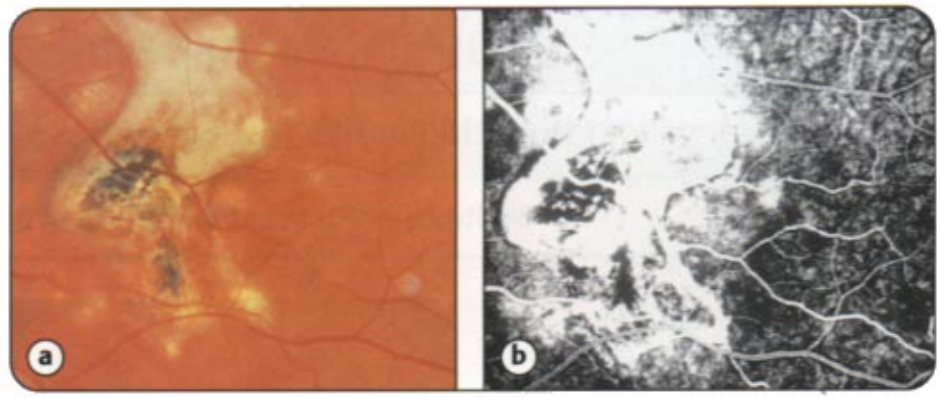

Fig. 12.76

Birdshot retinochoroidopathy

Birdshot retinochoroidopathy is an uncommon, usually bilateral condition that typically affects middle-aged individuals who carry HLA-A29. The prognosis is guarded.

Signs

- Moderate vitritis.
- Moderate size, creamy-yellow, deep ovoid spots with indistinct edges, which radiate from the optic disc toward the equator (**Fig. 12.77a and b, 12.78a**).
- Fluorescein angiography shows late intraretinal and disc leakage (**Fig. 12.78b**).

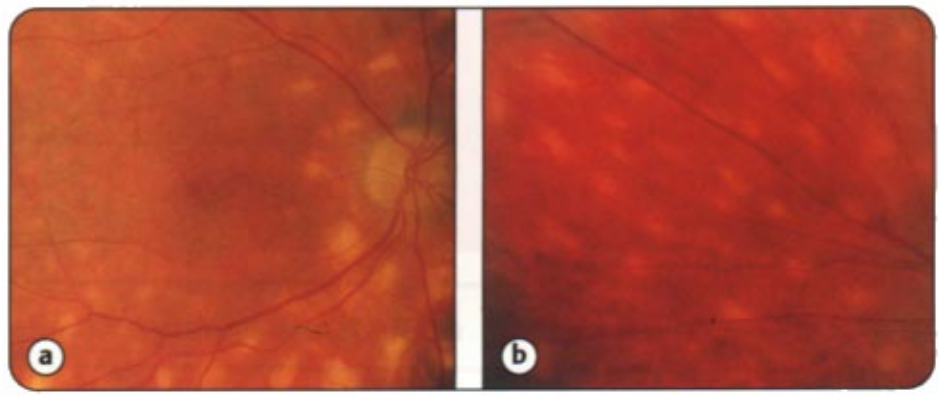

Fig. 12.77

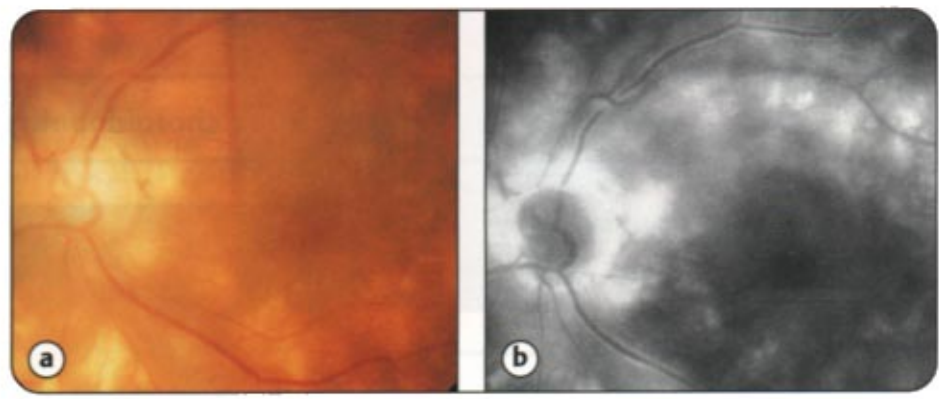

Fig. 12.78

Residua

- Circumscribed atrophic depigmented scars (**Fig. 12.79**).
- Occasional choroidal neovascularization.

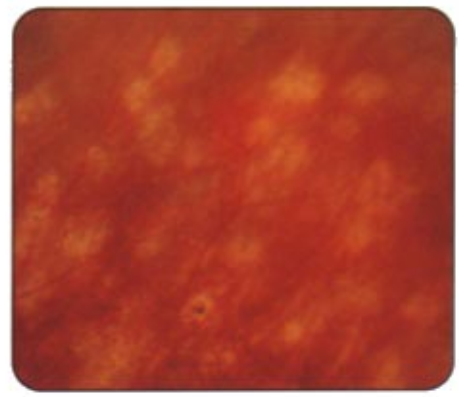

Fig. 12.79

Acute posterior multifocal placoid pigment epitheliopathy

Acute posterior multifocal placoid pigment epitheliopathy (APMPPE) is an uncommon, bilateral condition which typically affects young adults and has an excellent prognosis.

Signs

- Mild vitritis.
- Large, cream-colored, placoid lesions at the level of the RPE located at the posterior pole and midperiphery (**Fig. 12.80a and b**).
- Fluorescein angiography shows initial dense hypofluorescence (**Fig. 12.80c and d**) followed by late diffuse staining (**Fig. 12.80e and f**).

Residua

- Diffuse pigment scars but usually good visual acuity (**Fig. 12.81a and b**).

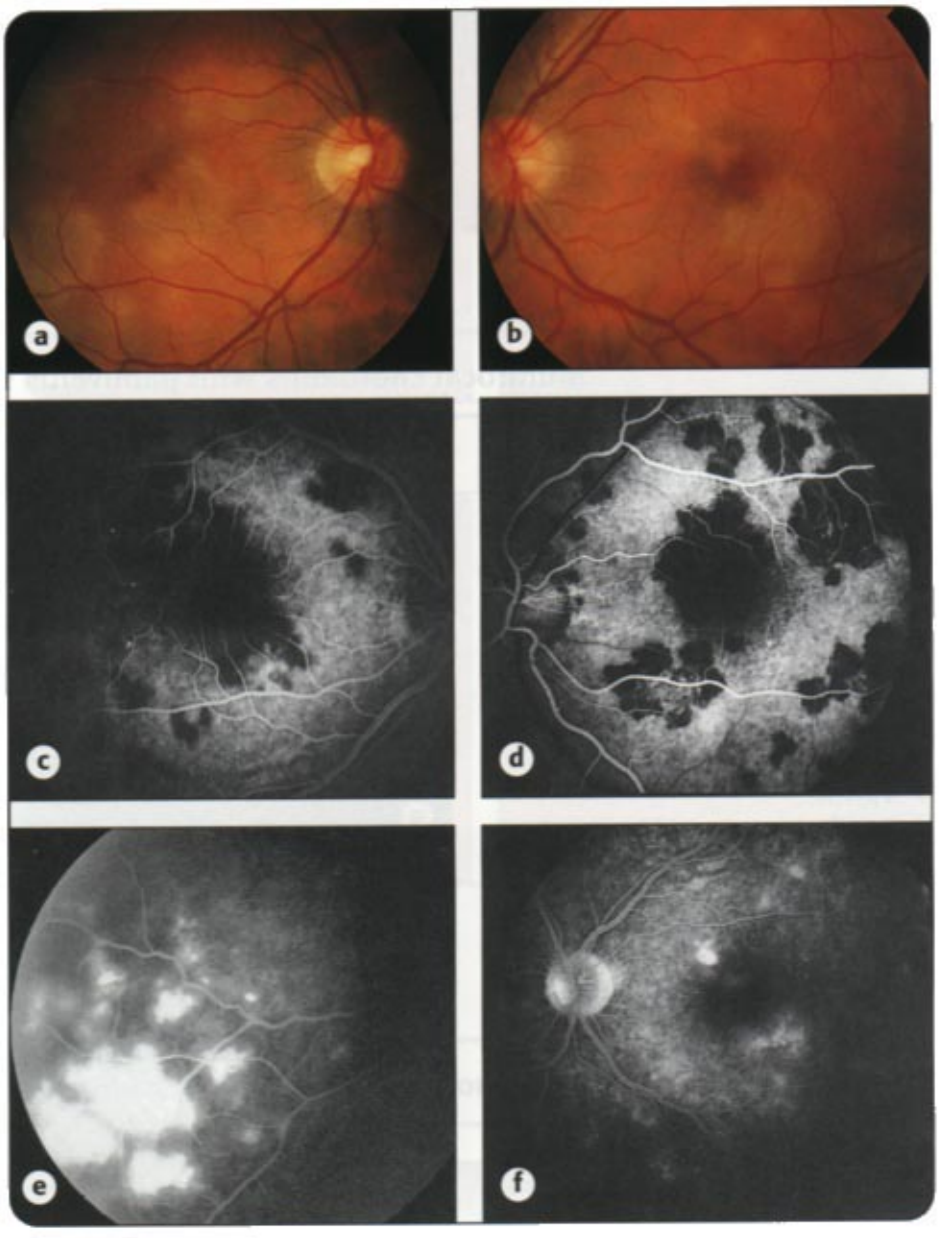

Fig. 12.80

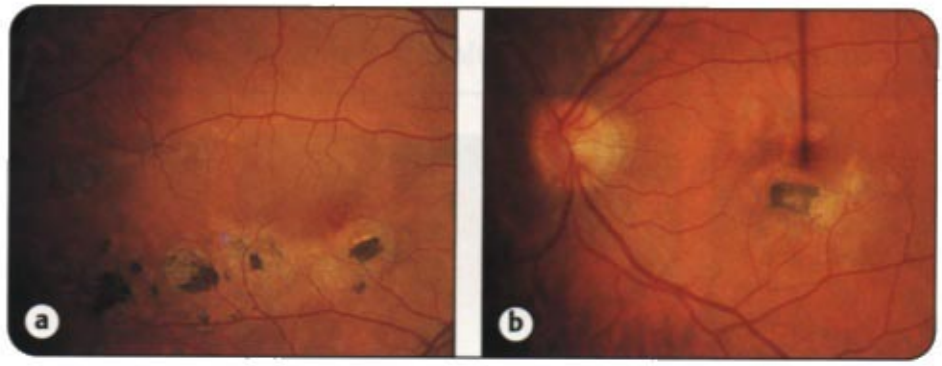

Fig. 12.81

Multifocal choroiditis with panuveitis syndrome

Multifocal choroiditis with panuveitis syndrome is an uncommon, unilateral or bilateral, frequently recurrent condition which may occur at any age but typically affects middle-aged women. It has a fair prognosis.

Signs

- Anterior uveitis.
- Vitritis.
- Small, discrete, old and fresh choroidal lesions located at the midperiphery and fewer at the posterior pole (**Fig. 12.82a and b**).
- The lesions are more apparent on fluorescein angiography (**Fig. 12.82c and d**). Acute lesions either block or fill early, and stain late.

Residua

- Atrophic spots with variable pigmentation mixed with fresh lesions (**Fig. 12.83a and b**).
- Occasional choroidal neovascularization.
- Subretinal fibrosis (rarely).

Dx Differential diagnosis

- Sarcoidosis.
- Acute lesions may resemble PIC but the latter is not associated with vitritis.
- Presumed ocular histoplasmosis may show similar lesions but is not associated with vitritis.

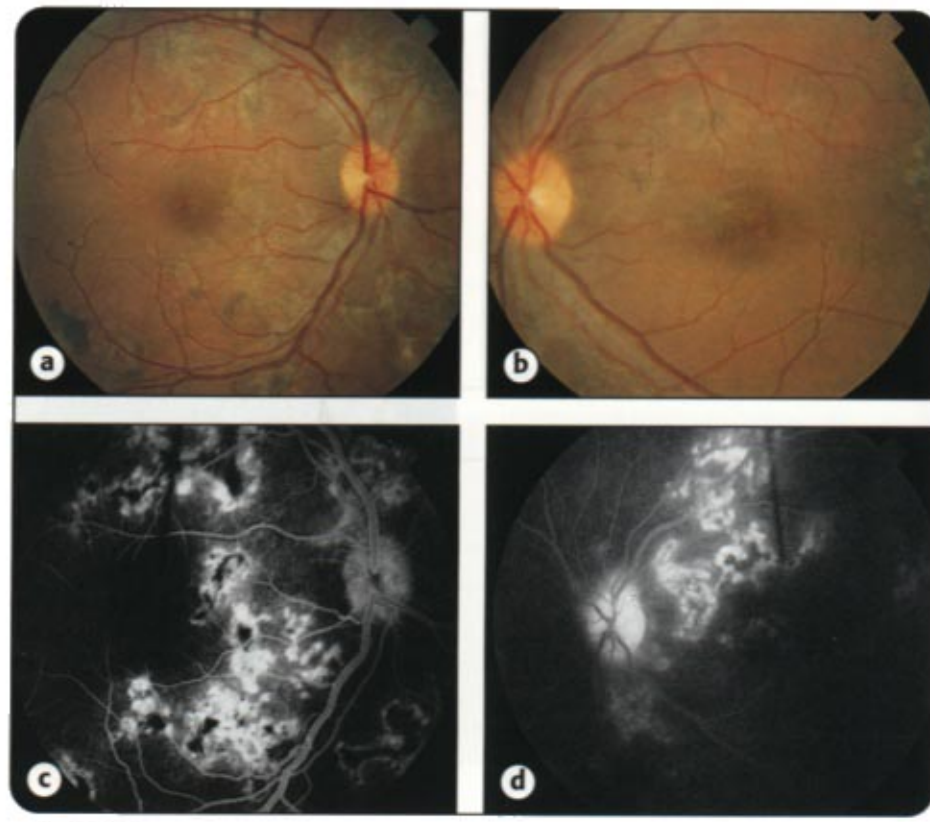

Fig. 12.82

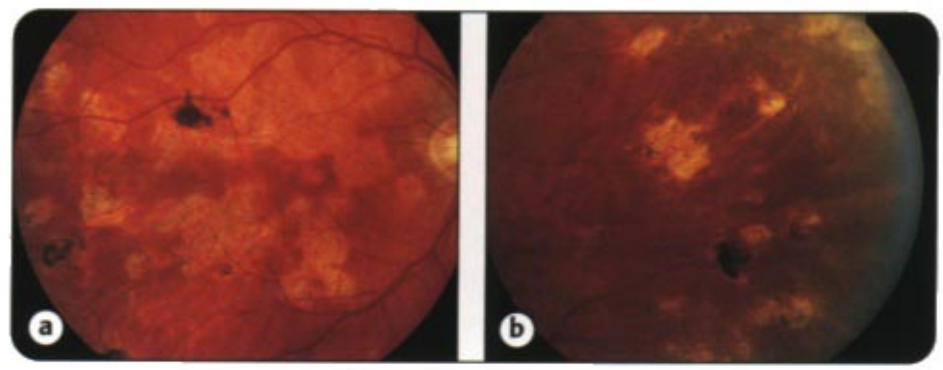

Fig. 12.83

Serpiginous choroidopathy

Serpiginous choroidopathy is an uncommon, eventually bilateral condition which typically affects middle-aged individuals and has a poor prognosis.

Signs

- Mild vitritis.
- Geographic choroidal lesions with hazy borders, which typically spread outward from the disc (**Fig. 12.84a and b**).

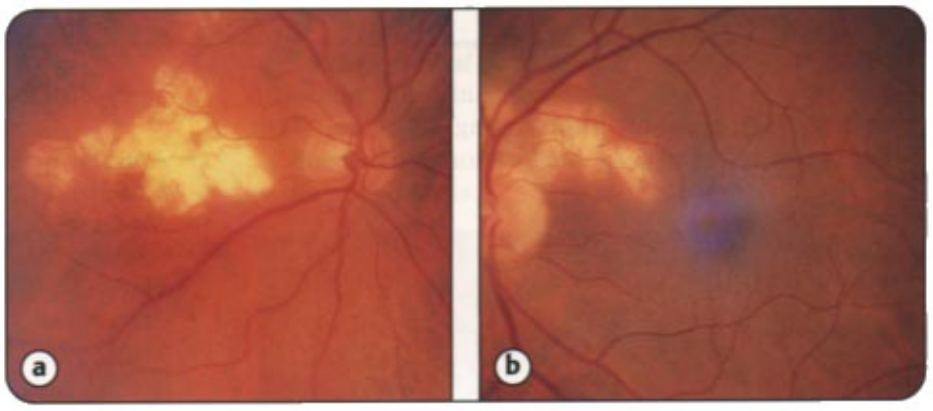

Fig. 12.84

Residua

- Scalloped, punched-out atrophic areas (**Fig. 12.85a and b**).
- Occasional choroidal neovascularization.

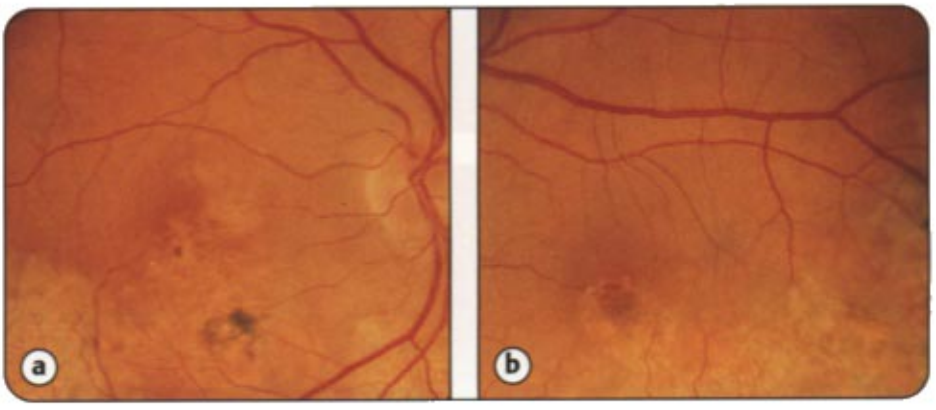

Fig. 12.85

Diffuse subretinal fibrosis syndrome

Diffuse subretinal fibrosis syndrome is a rare, bilateral, recurrent condition which affects young adults. It has a poor prognosis.

Signs

- Vitritis.
- Yellow, indistinct subretinal lesions which coalesce into dirty-yellow mounds.

Residua

- Subretinal opaque bands (**Fig. 12.86**).

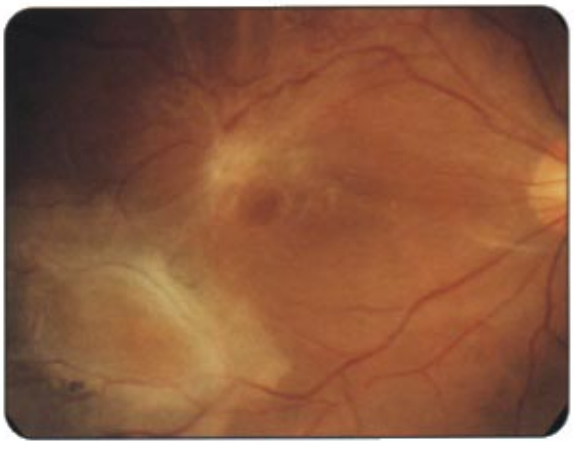

Fig. 12.86

Focal infectious posterior uveitis

Toxoplasmosis

Toxoplasmosis is a common, usually unilateral, protozoan infection which typically affects young individuals. The visual prognosis depends on the location of the lesion.

Typical signs

- Anterior uveitis which may be granulomatous. **Figure 12.87** shows mutton-fat keratic precipitates.
- Moderate to severe vitritis.
- Solitary focal retinitis adjacent to an old scar (**Figs 12.88–12.91a**).
- Fluorescein angiography shows extensive late leakage of dye (**Fig. 12.91d**).

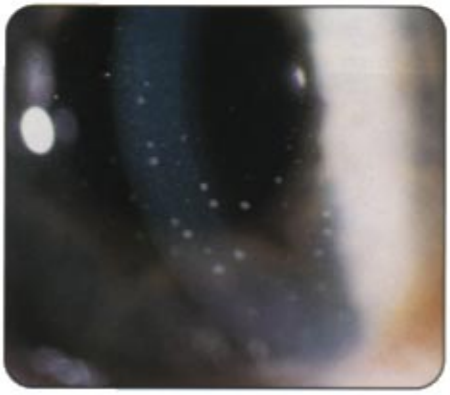

Fig. 12.87

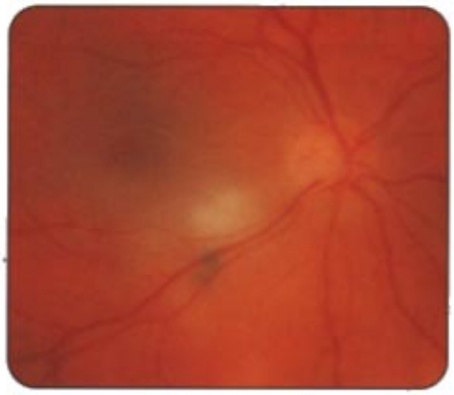

Fig. 12.88

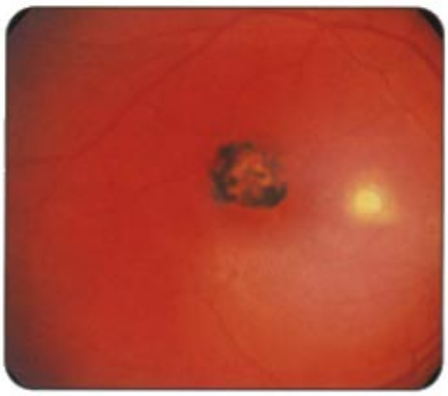

Fig. 12.89

Fig. 12.90

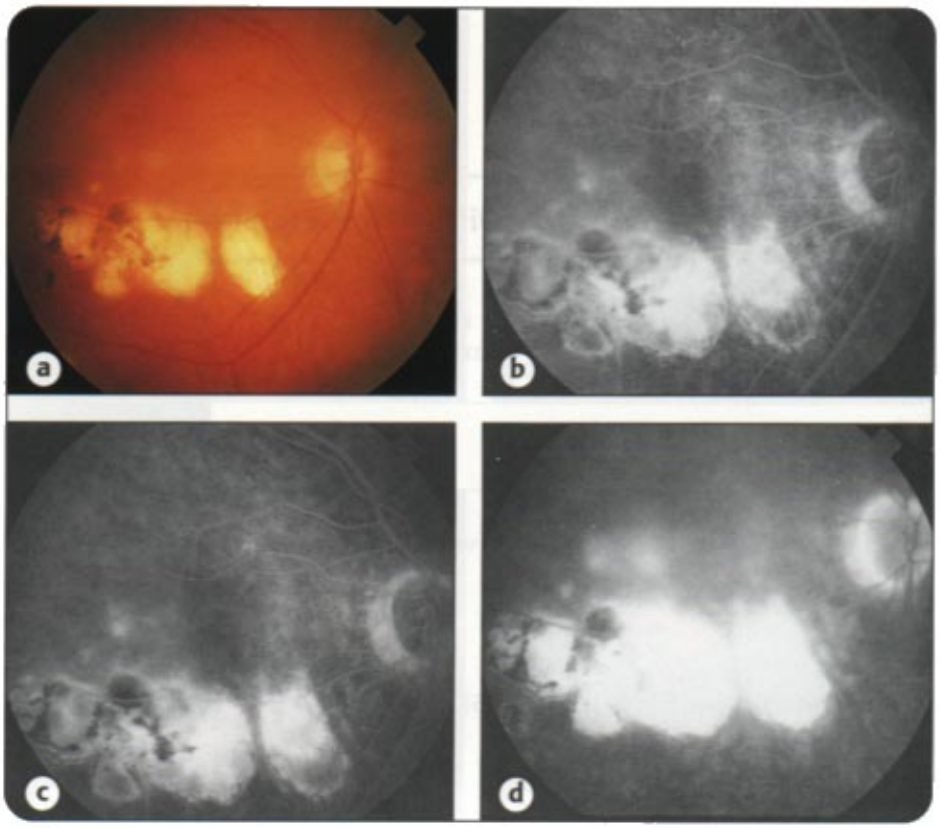

Fig. 12.91

Atypical signs

- Not associated with old scars in acquired toxoplasmosis (**Fig. 12.92**).
- Multifocal (**Fig. 12.93**) in patients with AIDS.
- Bilateral.

Residua

- Punched-out atrophic scar associated with variable pigmentation (**Figs 12.94, 12.95**).

Dx Differential diagnosis of solitary focal retinitis

- Candidiasis.
- Toxocariasis.
- Cryptococcosis.

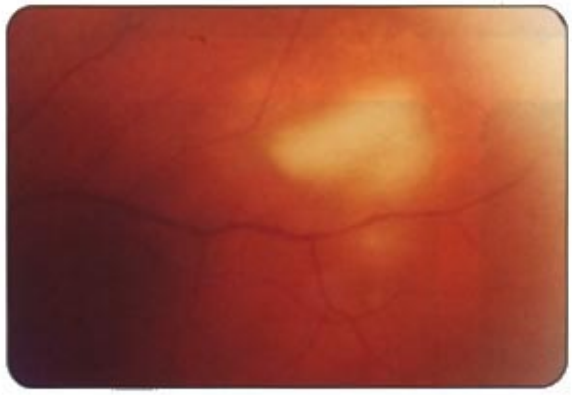

Fig. 12.92

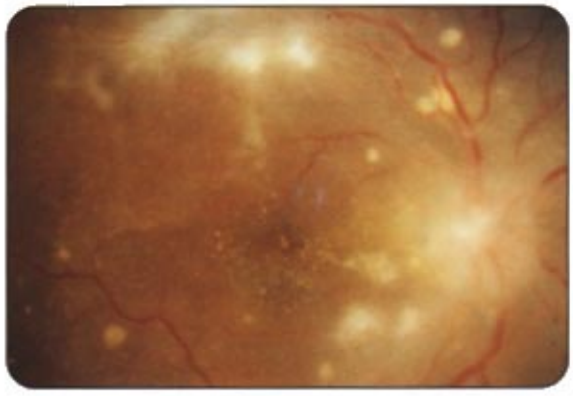

Fig. 12.93

Fig. 12.94

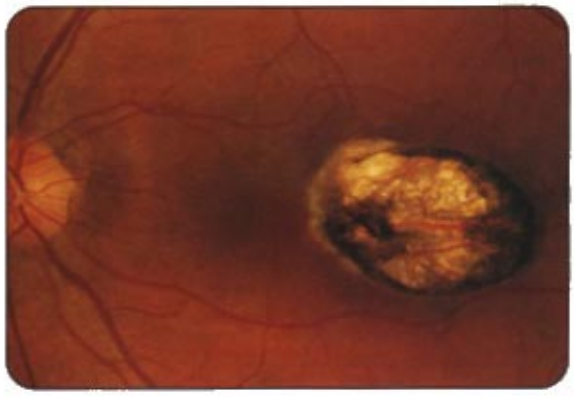

Fig. 12.95

Toxocariasis

Toxocariasis is a very rare, worm infection that is acquired in early childhood. Ocular involvement is always unilateral.

Signs

- Juxtapapillary granuloma (**Figs 12.96, 12.97**).
- Posterior-pole granuloma (**Figs 12.98, 12.99**).
- Peripheral granuloma (**Fig. 12.100**), which may be associated with 'dragging' of the disc (**Fig. 12.101**).
- Chronic endophthalmitis.

Dx Differential diagnosis

- Retinoblastoma may resemble the posterior-pole and peripheral granulomas.

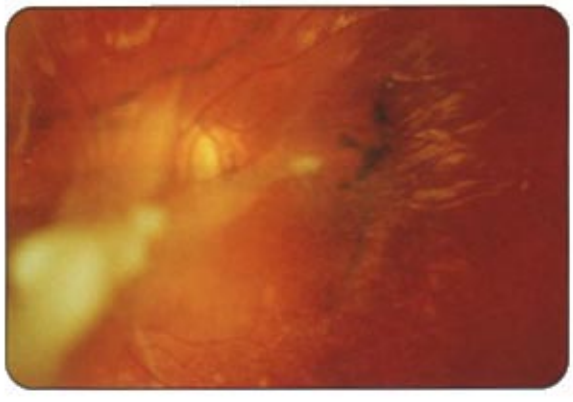

Fig. 12.96

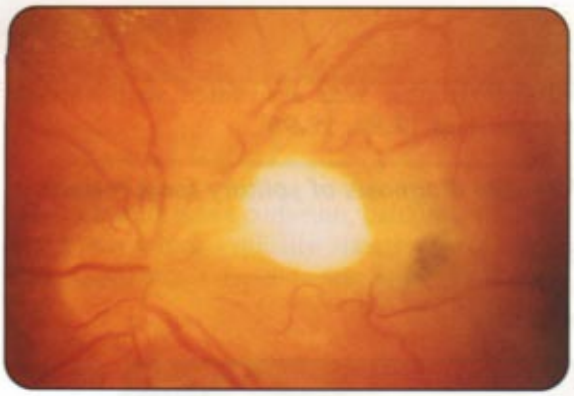

Fig. 12.97

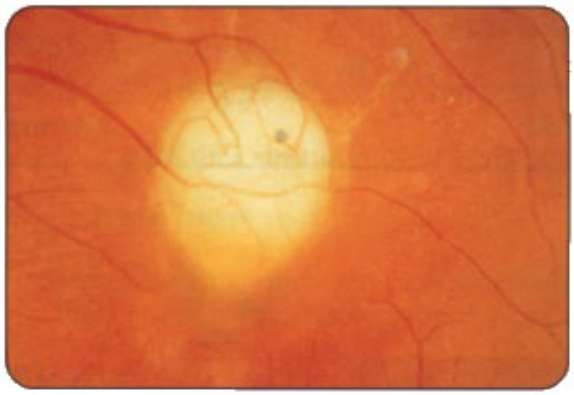

Fig. 12.98

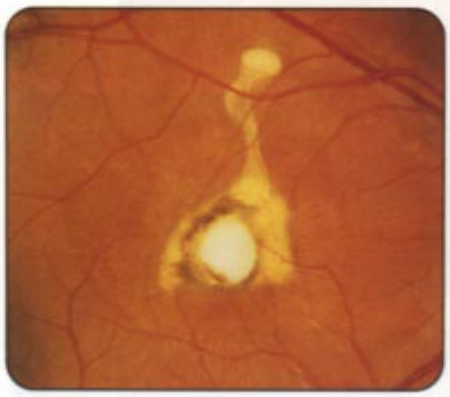

Fig. 12.99

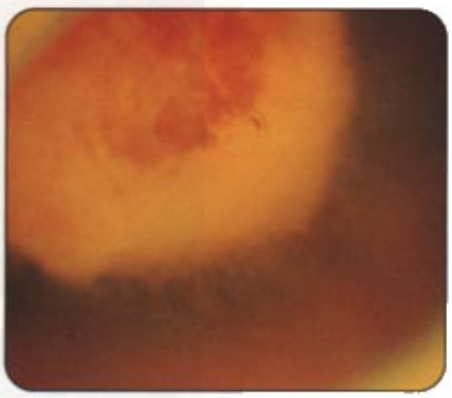

Fig. 12.100

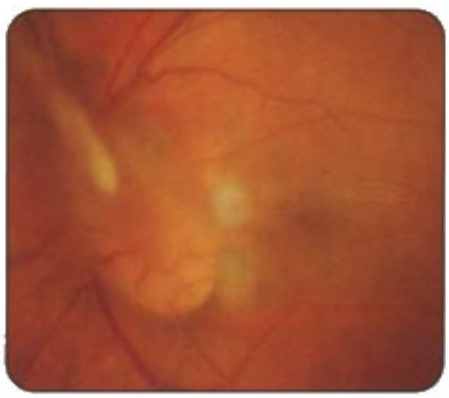

Fig. 12.101

Candidiasis

Candidiasis is an uncommon, unilateral or bilateral, fungal infection which typically affects drug addicts, patients with long-term indwelling catheters, or those on hyperalimentation.

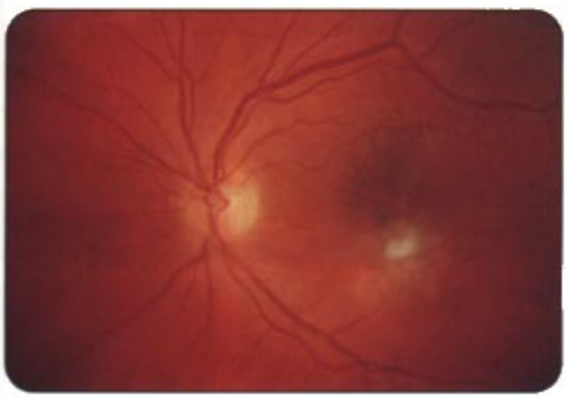

Fig. 12.102

Signs

- Deep, white retinal infiltrates which may be solitary (**Fig. 12.102**) or multiple.
- Vitritis (**Fig. 12.103**) associated with fluffy white 'cotton' balls (**Figs 12.104, 12.105**).

Other causes of vitreous cotton balls

- Intermediate uveitis.
- Sarcoidosis.
- Lyme disease.
- Whipple disease.
- Seedings of endophytic retinoblastoma.

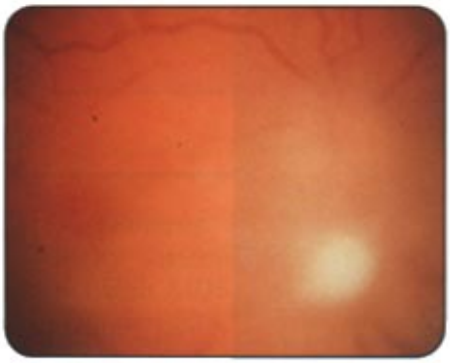

Fig. 12.103

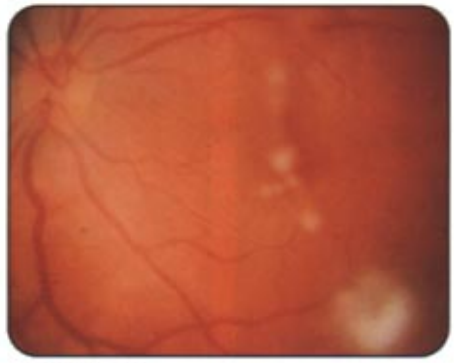

Fig. 12.104

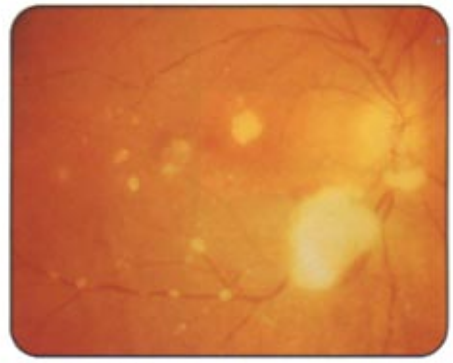

Fig. 12.105

Presumed ocular histoplasmosis syndrome

Presumed ocular histoplasmosis syndrome is a fungal infection which is common in certain parts of the USA but rare in Europe. Both eyes are usually affected. In the absence of choroidal neovascularization, the visual prognosis is good.

Signs

- No vitritis.
- Peripapillary atrophy (**Fig. 12.106**) and small, multifocal, discrete, hypopigmented chorioretinal lesions (histo spots) outside the vascular arcades (**Figs 12.107, 12.108**).
- Peripheral pigmented linear streaks (**Fig. 12.109**).

Causes of visual loss

- Macular choroidal neovascularization (**Fig. 12.110a–d**). The membrane is seen during the early phase of the angiogram (**Fig. 12.110b**) and stains late (**Fig. 12.110d**).

Differential diagnosis

- Paving-stone degeneration may resemble the peripheral atrophic lesions.
- Degenerative myopia may cause both the atrophic lesions and choroidal neovascularization.
- Punctate inner choroidopathy.
- Inactive multifocal choroiditis with panuveitis syndrome.

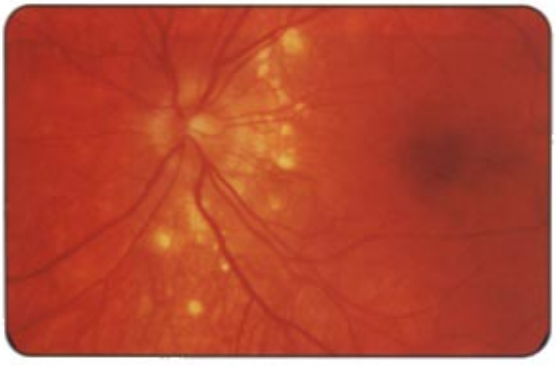

Fig. 12.106

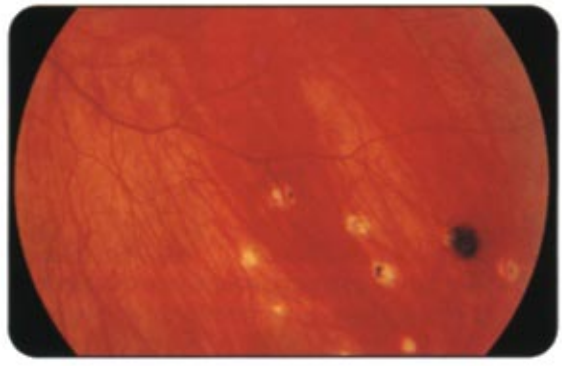

Fig. 12.107

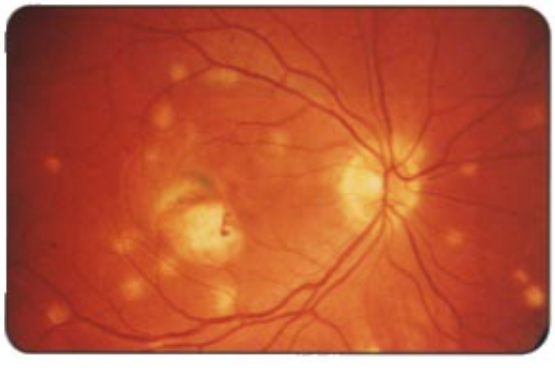

Fig. 12.108

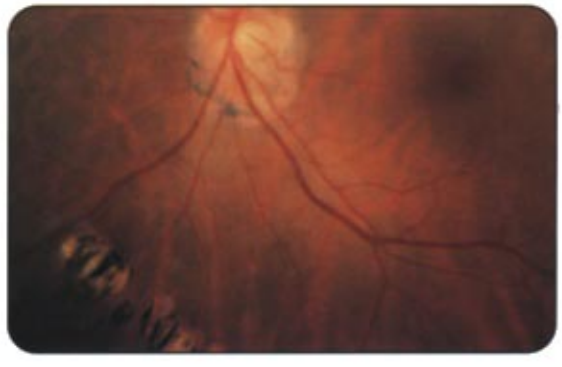

Fig. 12.109

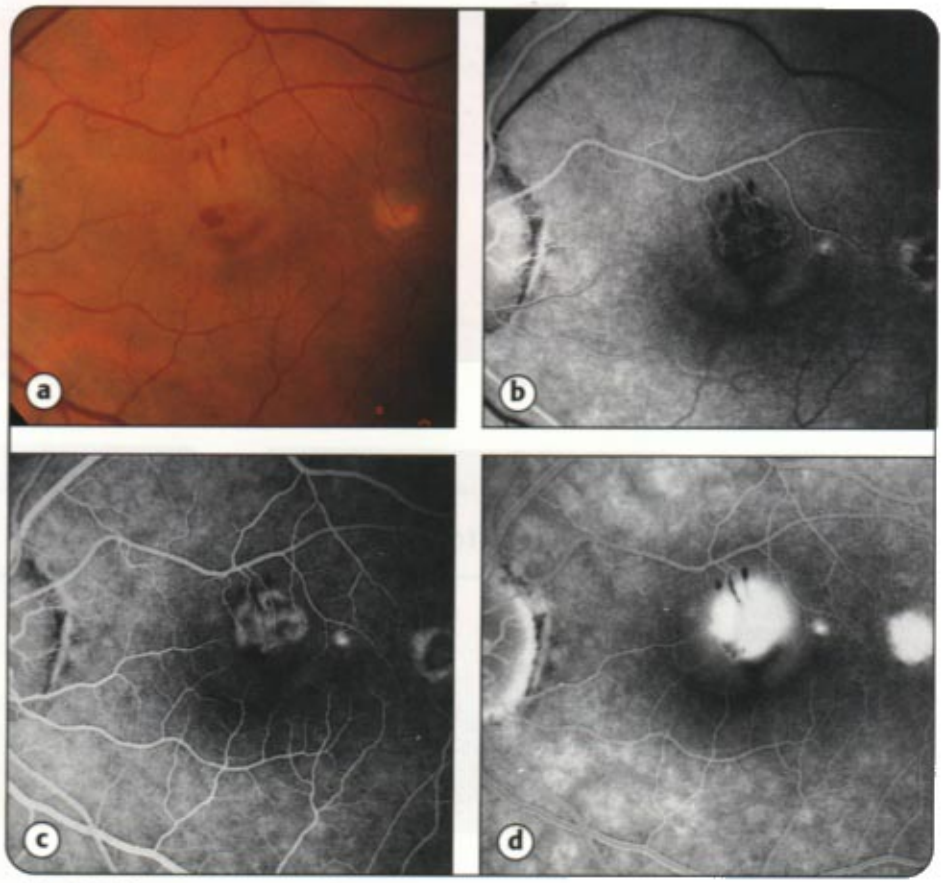

Fig. 12.110

Choroidal pneumocystosis

Choroidal pneumocystosis is a rare, usually bilateral infection with *Pneumocystis carinii* which affects patients with AIDS. It has a good visual prognosis.

Signs

- No vitritis.
- Large, multifocal, slightly elevated, round or oval, plaque-like, yellowish choroidal lesions predominantly involving the posterior pole (**Fig. 12.111**).

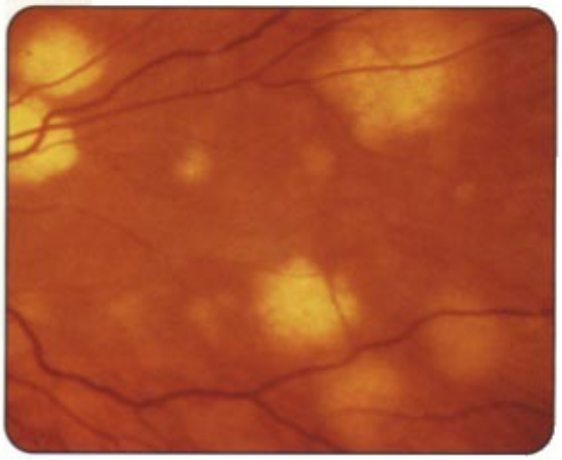

Fig. 12.111

Diffuse infectious retinitis

Cytomegalovirus retinitis

Cytomegalovirus (CMV) retinitis eventually affects up to 25% of patients with AIDS. It is bilateral in approximately 50% of cases.

Signs

- Mild vitritis.
- Slowly progressive, yellow–white retinal infiltration, with advancing brushfire-like borders (**Figs 12.112, 12.113**).
- Yellow–white areas of retinitis and hemorrhage which typically start at the posterior pole and then frequently spread along the vascular arcades (**Figs 12.114, 12.115**).

Signs of decreased activity

- Fewer hemorrhages.
- Less whitening.
- Development of diffuse atrophy and pigmentary changes (**Fig. 12.116**).

Causes of visual loss

- Macular involvement (**Fig. 12.117**).
- Optic nerve involvement (**Fig. 12.118**).
- Retinal detachment (**Fig. 12.119**).

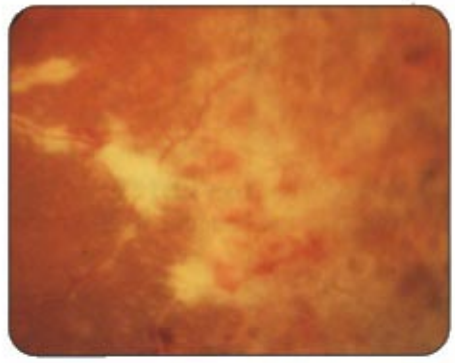

Fig. 12.112

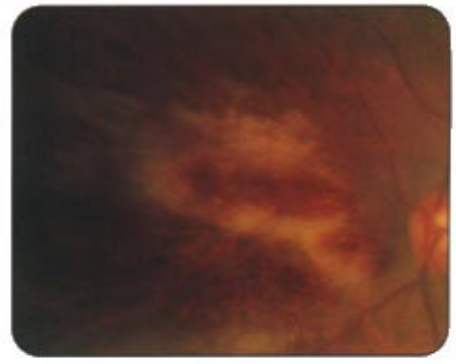

Fig. 12.113

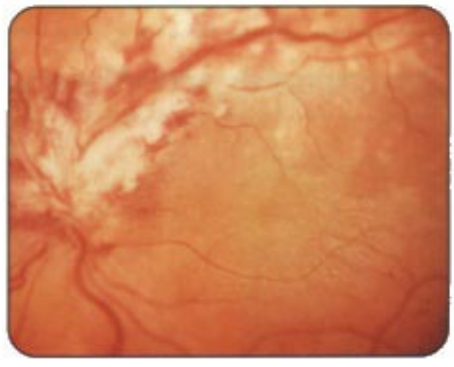

Fig. 12.114

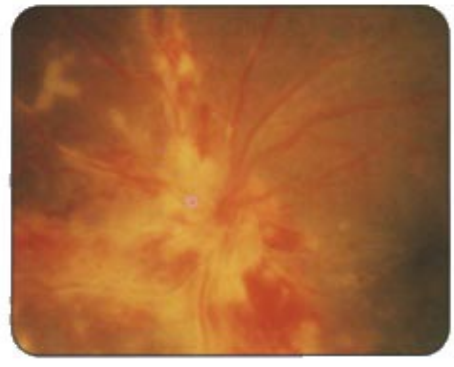

Fig. 12.115

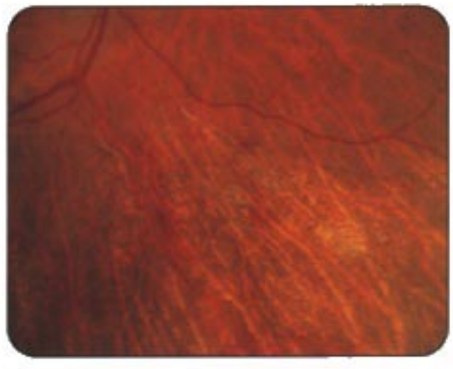

Fig. 12.116

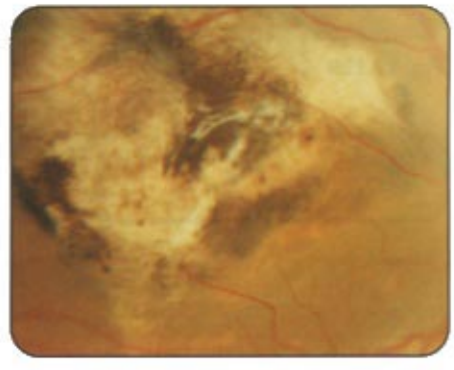

Fig. 12.117

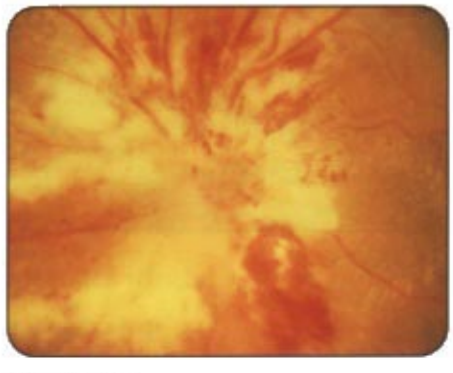

Fig. 12.118

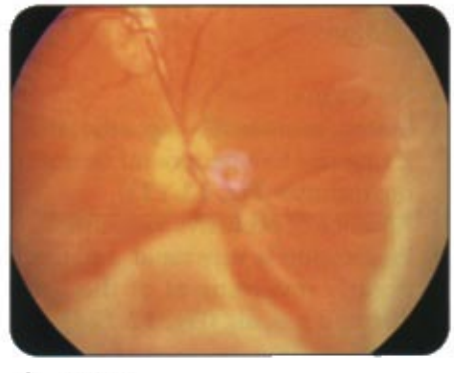

Fig. 12.119

Acute retinal necrosis

Acute retinal necrosis (ARN) is a rare infection caused by the varicella-zoster virus. It is bilateral in 60% of cases and usually affects otherwise healthy individuals.

Signs

- Anterior uveitis.
- Vitritis (**Fig. 12.120**).
- Confluent, peripheral occlusive necrotizing vasculitis (**Figs 12.121, 12.122**).
- Progression is faster than in CMV infection but the macula itself is usually spared.

Causes of visual loss

- Optic nerve involvement.
- Retinal detachment.

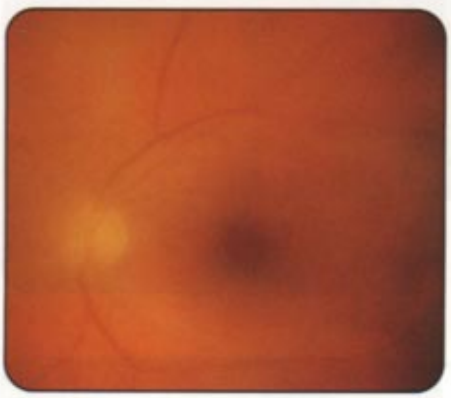

Fig. 12.120

Fig. 12.121

Fig. 12.122

Progressive outer retinal necrosis

Progressive outer retinal necrosis (PORN) is a severe variant of ARN which affects patients with AIDS. It is bilateral in 70% of cases.

Signs

- No vitritis.
- Deep multifocal opacification, giving rise to a peripheral necrotizing retinitis (**Fig. 12.123**).
- Absence of occlusive vasculitis.
- Progression is very rapid, and there is early involvement of the optic disc and macula (**Fig. 12.124**).

Fig. 12.123

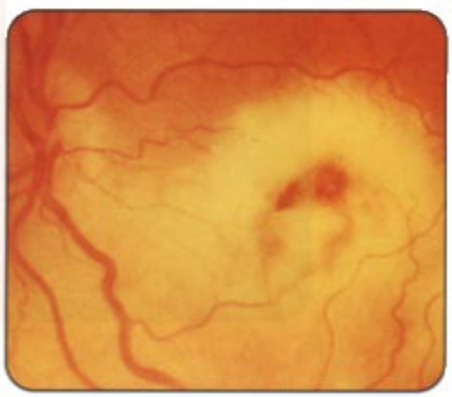

Fig. 12.124

Herpes simplex retinitis

Herpes simplex retinitis is a very rare condition that may occur in healthy as well as immunocompromised individuals. It may be associated with herpetic meningitis.

Fig. 12.125

Signs

- Rapidly progressive, full-thickness necrotizing retinitis characterized by retinal opacification and perivasculitis (**Fig. 12.125**).

Measles retinitis

Patients with subacute sclerosing panencephalitis caused by measles frequently have fundus lesions associated with optic atrophy.

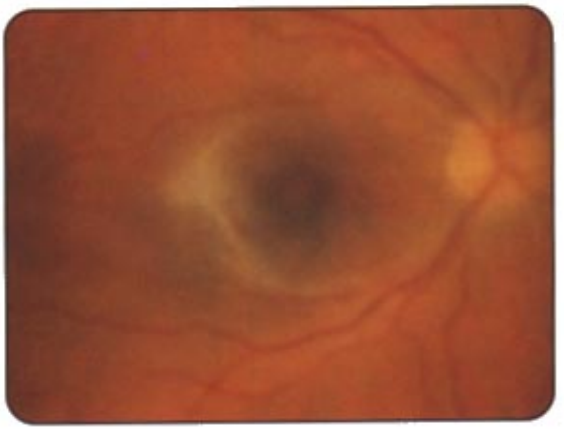

Fig. 12.126

Signs

- Bilateral necrotizing retinitis involving the macula (**Fig. 12.126**).

Residua

- Macular pigmentary changes and optic atrophy.

Panuveitis

The following conditions may be associated with anterior uveitis and posterior uveitis.

Sarcoidosis

Sarcoidosis is a common, multisystem disorder which affects the eyes in about 30% of cases.

Signs

- Anterior uveitis, which is most frequently chronic and granulomatous but may occasionally be nongranulomatous.
- Retinal periphlebitis (*see* **Fig. 12.52**), which may result in minor venous occlusion.
- Retinal periphlebitis with 'candle-wax drippings' (**Figs 12.127, 12.128**).
- Discrete, gray–white to waxy-yellow retinal and preretinal nodules (Lander sign) (**Fig. 12.129**).
- Multifocal choroidal infiltrates, which are more frequent in the inferior fundus (**Fig. 12.130a and b**).
- Solitary choroidal granulomas (**Fig. 12.131**) are seen occasionally.
- Vitreous snowballs.
- Peripheral retinal neovascularization.
- Optic disc edema and hemorrhages (**Fig. 12.132a and b**).
- Optic disc granuloma (**Fig. 12.133**).

Dx Differential diagnosis

- Multifocal choroiditis with panuveitis.
- Birdshot retinochoroidopathy.

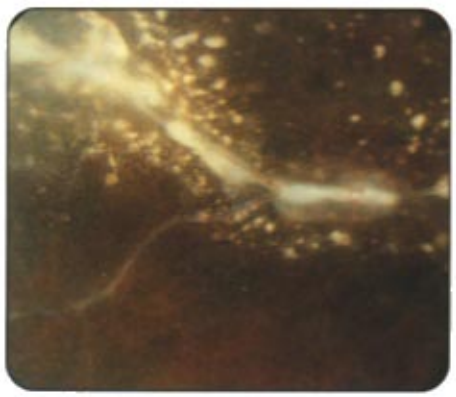

Fig. 12.127

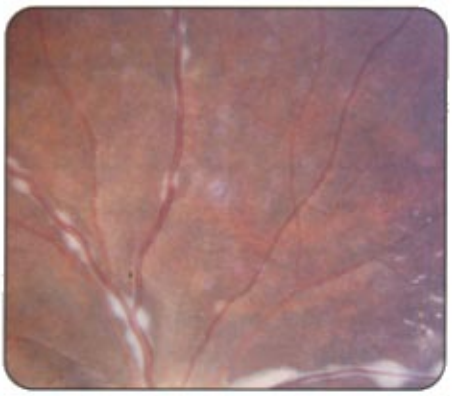

Fig. 12.128

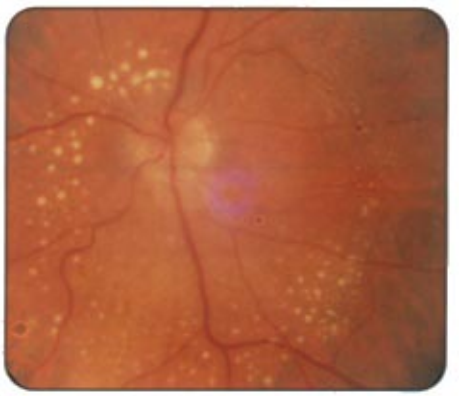

Fig. 12.129

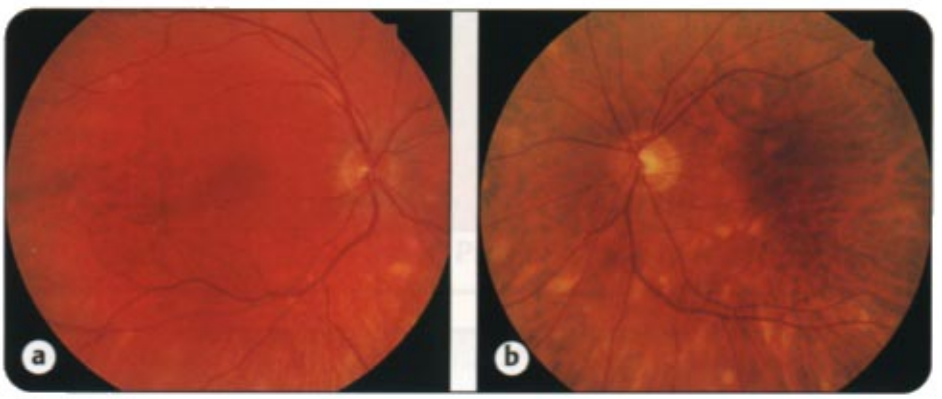

Fig. 12.130

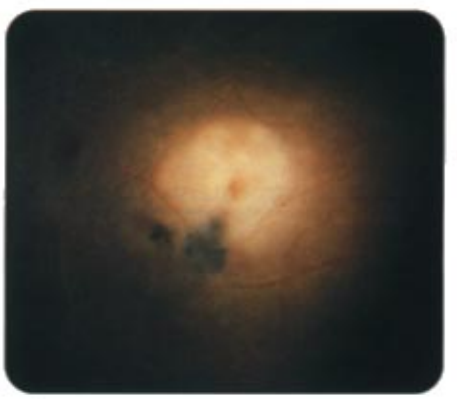

Fig. 12.131

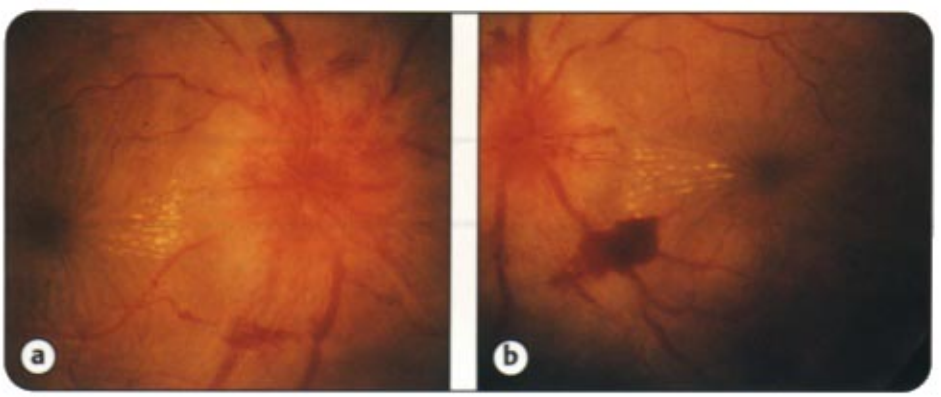

Fig. 12.132

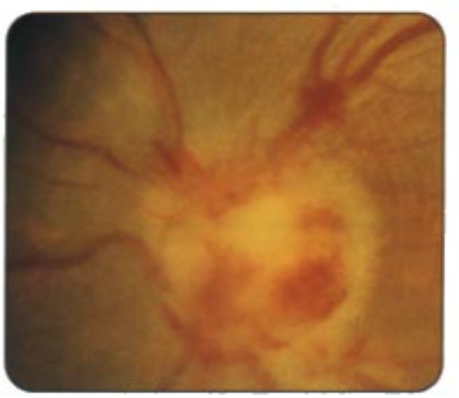

Fig. 12.133

Behçet disease

Behçet disease is an uncommon conditon characterized by recurrent oral and genital ulceration, skin lesions, and uveitis. Both eyes are frequently affected and the visual prognosis is guarded.

Signs

- Acute anterior uveitis with hypopyon.
- Vitritis.
- White, necrotic, superficial retinal infiltrates (**Fig. 12.134**).
- Vasculitis and intraretinal hemorrhages (**Fig. 12.135**).
- Optic disc edema and hemorrhages (**Fig. 12.136**).
- Diffuse retinal and cystoid macular edema.

Residua

- Macular scarring and vascular attenuation (**Fig. 12.137**).
- Retinal atrophy and optic atrophy (**Fig. 12.138**).

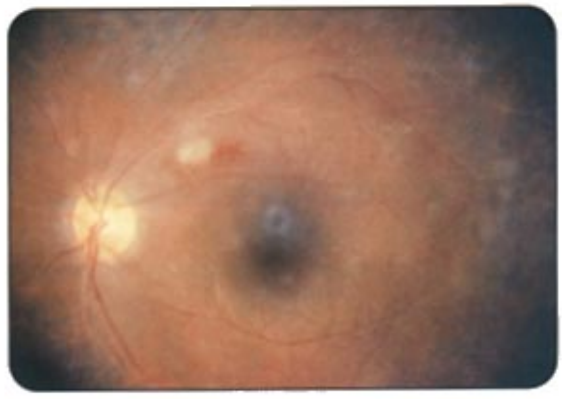

Fig. 12.134

Fig. 12.135

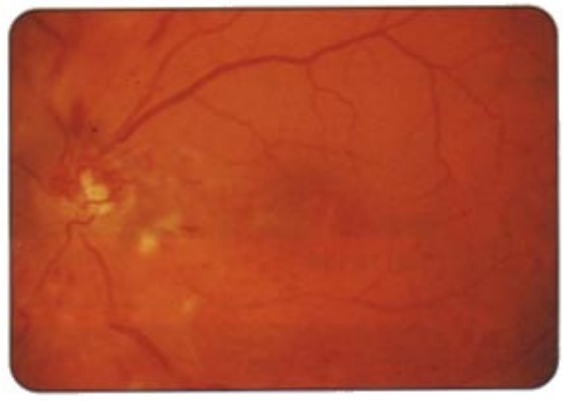

Fig. 12.136

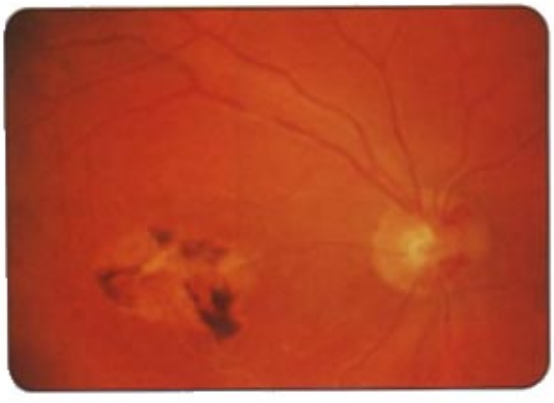

Fig. 12.137

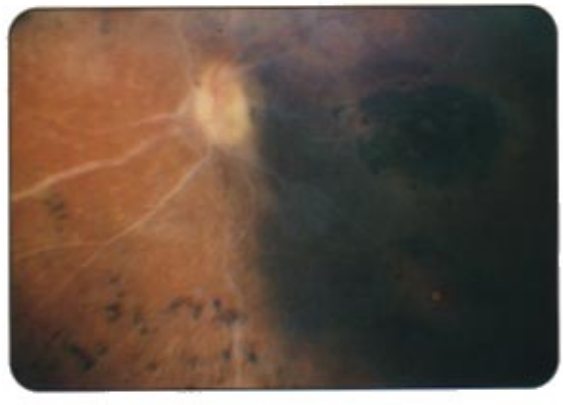

Fig. 12.138

Vogt–Koyanagi–Harada syndrome

Vogt–Koyanagi–Harada syndrome is a rare, idiopathic condition which typically affects Asians, Afro-Caribbeans, and Japanese. Bilateral ocular involvement is the rule. The prognosis is moderately good.

Signs

- Chronic granulomatous anterior uveitis (Vogt–Koyanagi syndrome).
- Multifocal choroiditis (**Fig. 12.139a and b**).
- Multiple detachments of the sensory retina (**Fig. 12.140a and b**).
- Exudative retinal detachment (Harada disease).

Residua

- Yellow–white, well-circumscribed atrophic spots, giving rise to a 'sunset glow' appearance.

Dx Differential diagnosis

- Other causes of exudative retinal detachment.

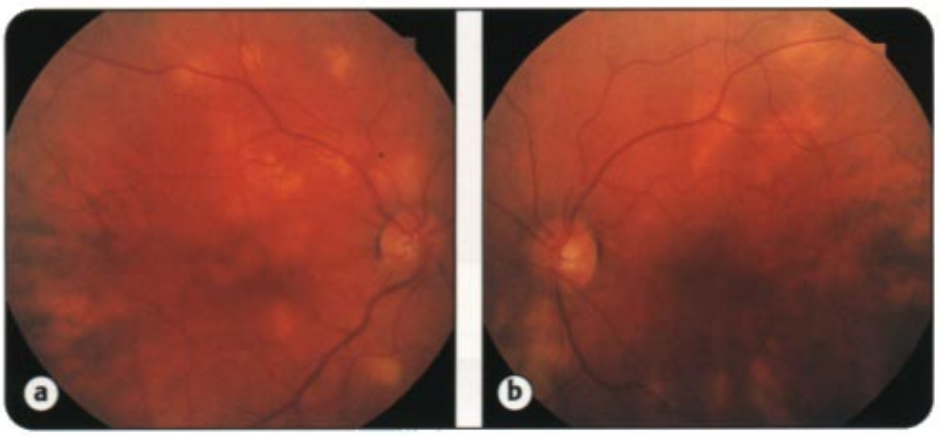

Fig. 12.139

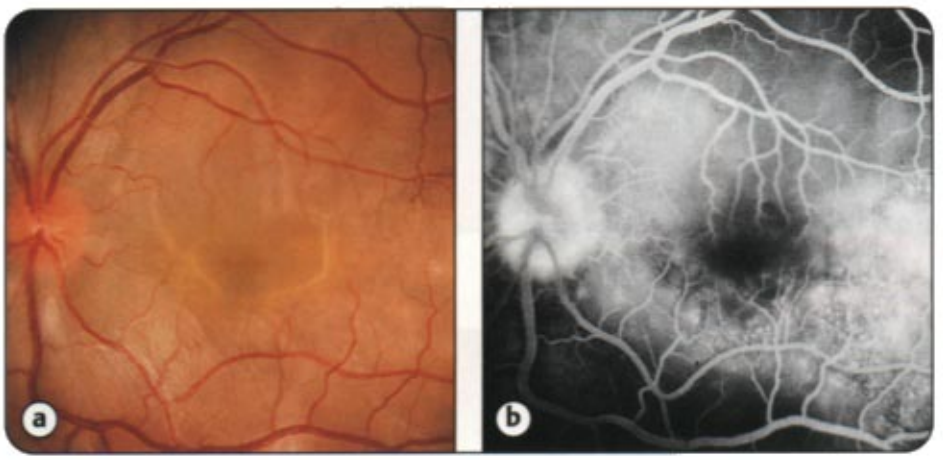

Fig. 12.140

Sympathetic ophthalmitis

Sympathetic ophthalmitis is a very rare, bilateral, granulomatous panuveitis which typically occurs following penetrating trauma. It carries a moderate prognosis.

Signs

- Chronic granulomatous anterior uveitis.
- Multifocal choroiditis involving the entire fundus (**Figs 12.141, 12.142**).

Residual features

- Multifocal chorioretinal scarring (**Fig. 12.143**).

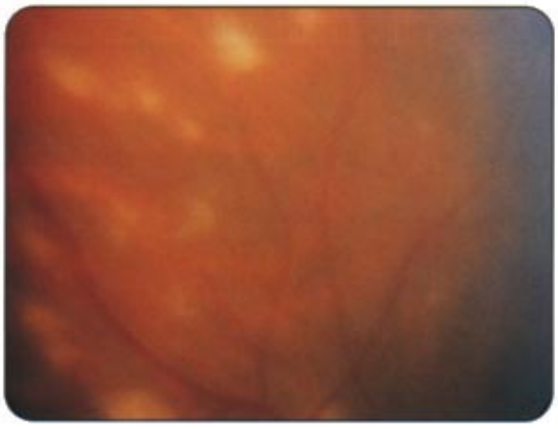

Fig. 12.141

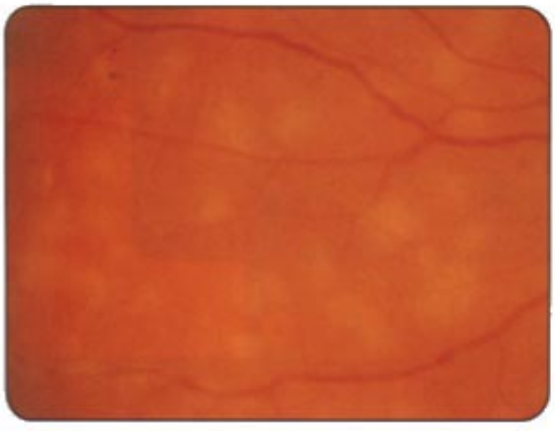

Fig. 12.142

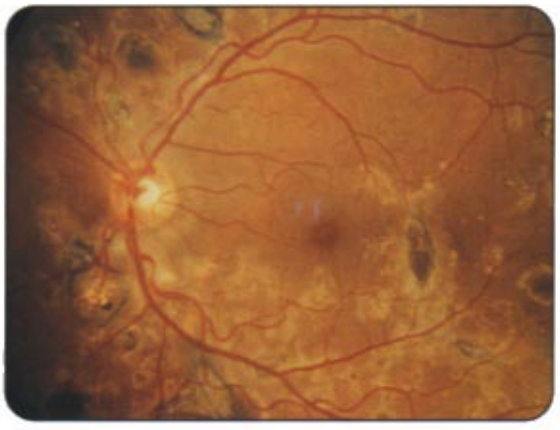

Fig. 12.143

Tuberculous choroiditis

Tuberculous choroiditis is an uncommon cause of uveitis.

Signs

- Chronic granulomatous anterior uveitis.
- Multifocal choroiditis (**Fig. 12.144a and b**).
- Retinal vasculitis similar to that seen in sarcoidosis.
- Solitary choroidal granulomas (**Fig. 12.145**) are seen occasionally.

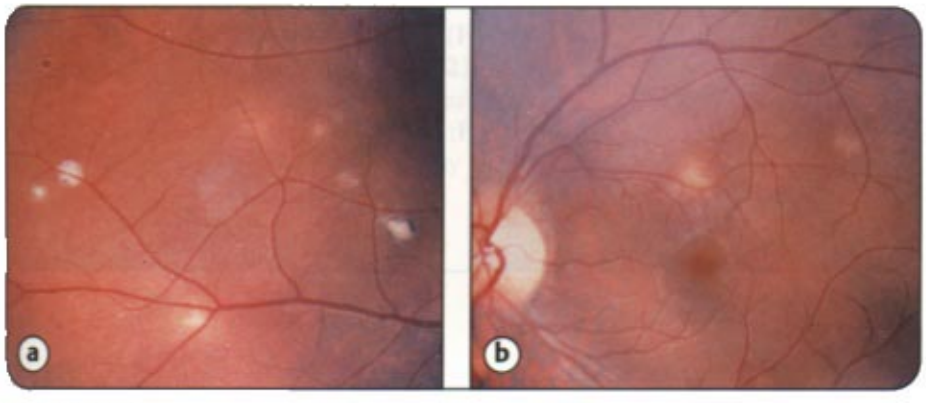

Fig. 12.144

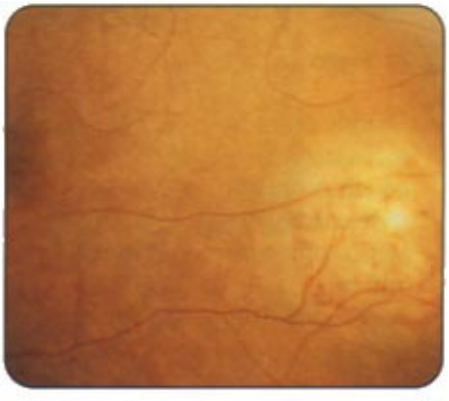

Fig. 12.145

Acquired syphilis

Acquired syphilis is a rare cause of uveitis.

Signs

- Anterior uveitis, which may be acute or chronic granulomatous.
- Neuroretinitis (*see* **Fig. 12.30**).
- Solitary or multifocal, large, deep, yellowish or gray placoid lesions with faded centers, at the posterior pole.
- Secondary serous retinal detachment (**Fig. 12.146**) may occur in some cases.

Residua

- Hypopigmented chorioretinal scars and pigment migration (**Fig. 12.147**).

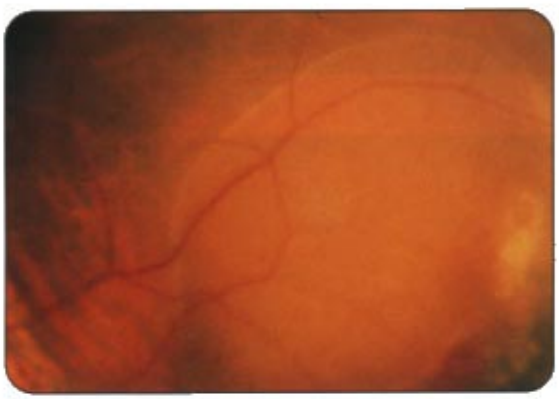

Fig. 12.146

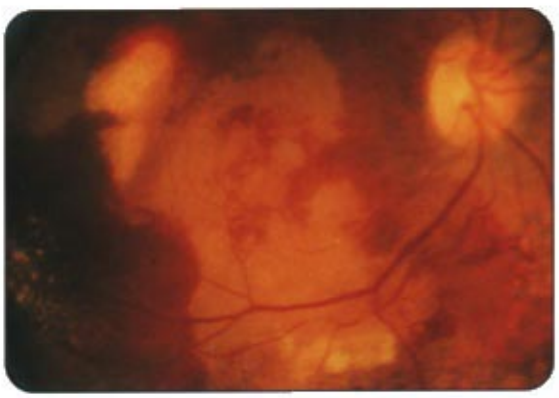

Fig. 12.147

ATROPHIC MACULOPATHIES

Nonhereditary

Dry age-related macular degeneration

Dry age-related macular degeneration is a very common condition which usually presents after the sixth decade of life. It is the most common form of age-related macular degeneration. It is usually bilateral but may be asymmetric.

Signs

- Sharply delineated areas of hypopigmentation or depigmentation in which choroidal vessels are more visible than in the surrounding area (geographic atrophy) (**Fig. 12.148a and b**).
- Associated drusen are usually evident.

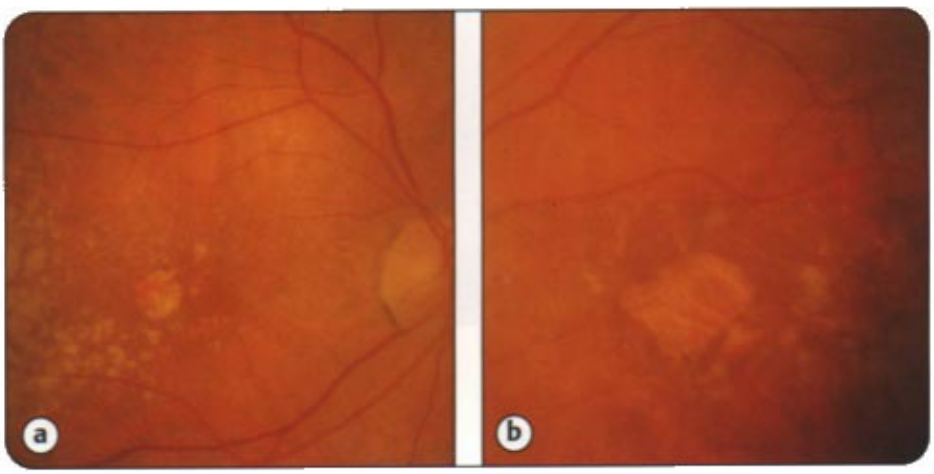

Fig. 12.148

Figure 12.149a and b shows widespread macular pigment atrophy and drusen – which becomes more evident on fluorescein angiography (**Fig. 12.149c and d**).

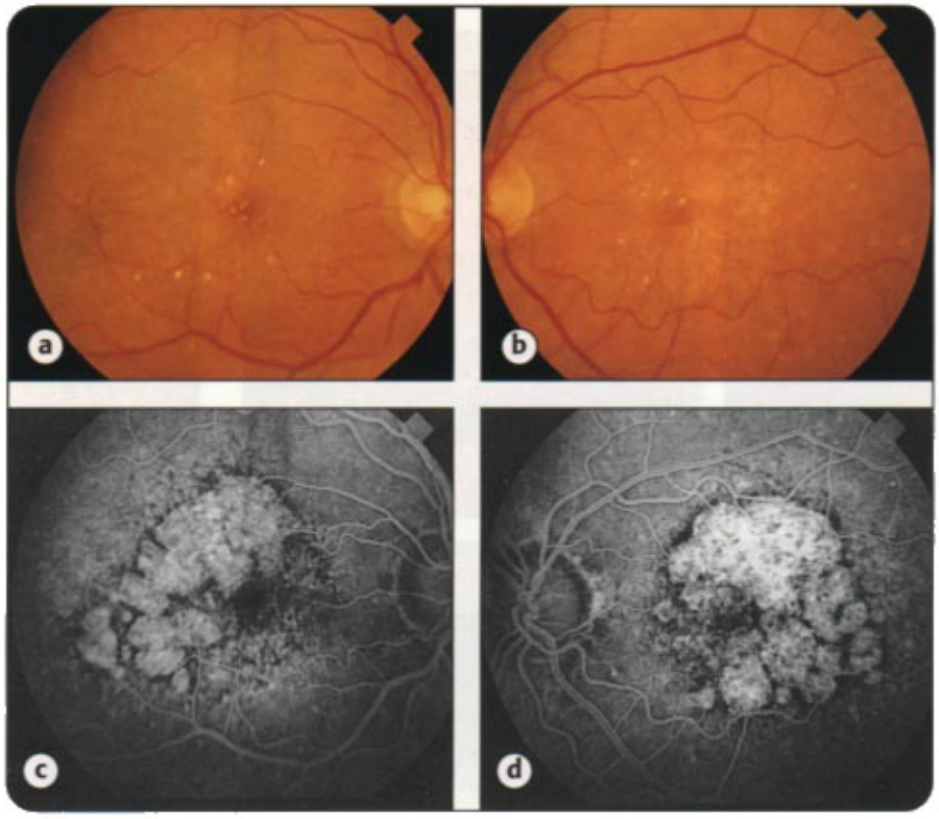

Fig. 12.149

Myopic maculopathy

Myopic maculopathy is usually a bilateral but asymmetric condition which affects highly myopic adults.

Signs

- Atrophic maculopathy (**Fig. 12.150**).
- Lacquer cracks at the posterior pole (**Fig. 12.151**).
- Macular hemorrhage (**Fig. 12.152**) which may be associated with choroidal neovascularization.
- Pigmented (Fuchs) spot at the fovea (**Fig. 12.153**).
- Macular hole (**Fig. 12.154**) which may give rise to retinal detachment.

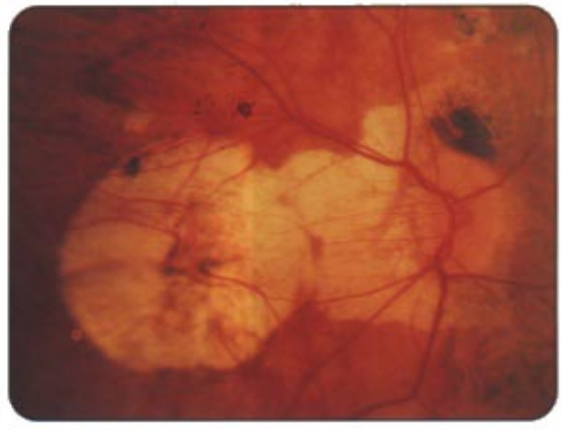

Fig. 12.150

Look for

- Tilted disc and peripapillary atrophy (**Fig. 12.155 a and b**).
- Posterior staphyloma (**Fig. 12.156**).
- Peripheral chorioretinal atrophy (**Fig. 12.157**).
- Lattice degeneration.
- Retinal breaks (**Fig. 12.158**).
- Retinal detachment.
- Vitreous degeneration.

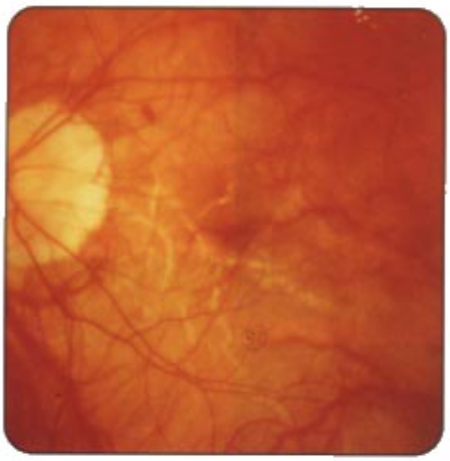

Fig. 12.151

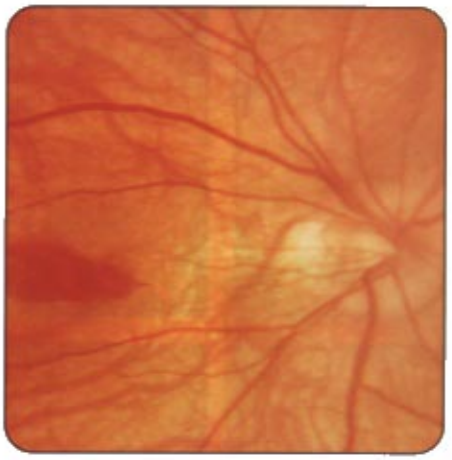

Fig. 12.152

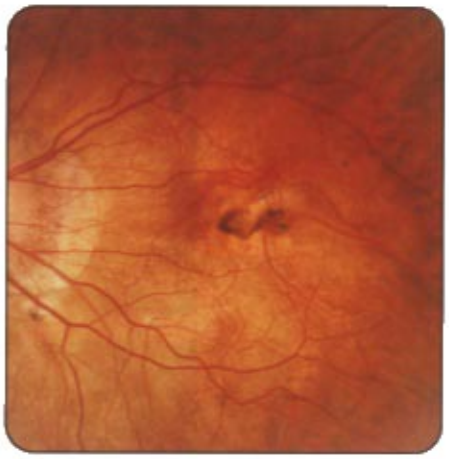

Fig. 12.153

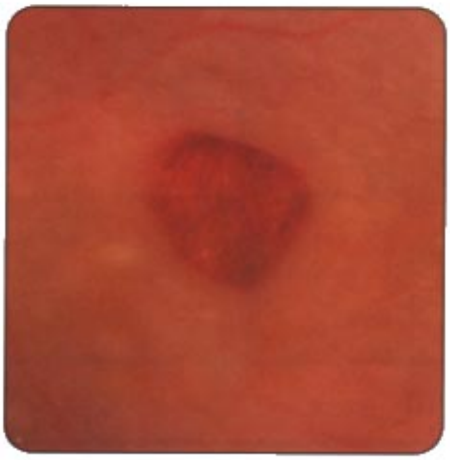

Fig. 12.154

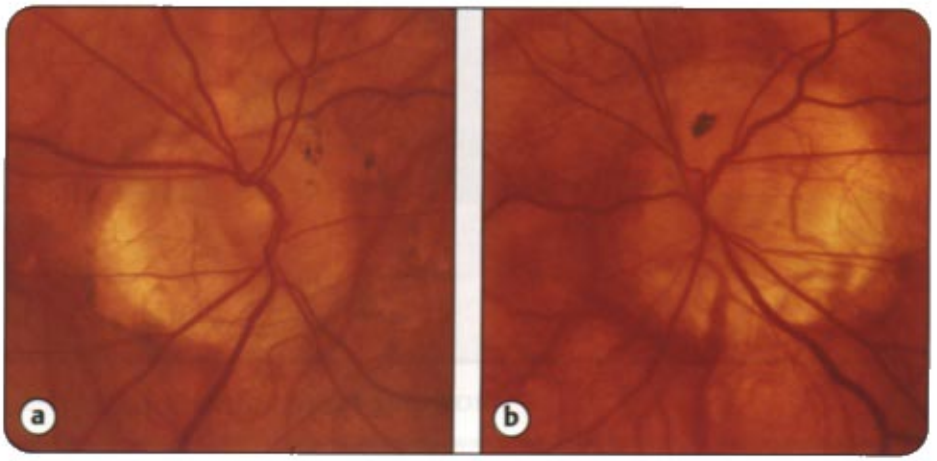

Fig. 12.155

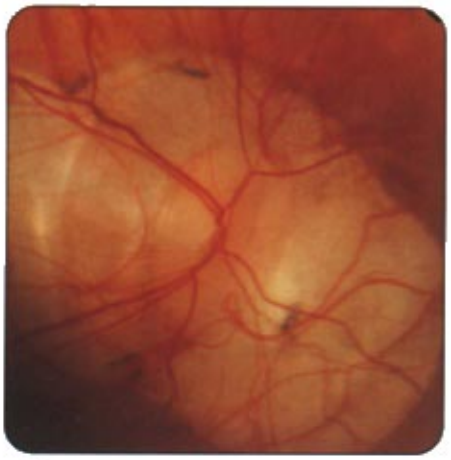

Fig. 12.156

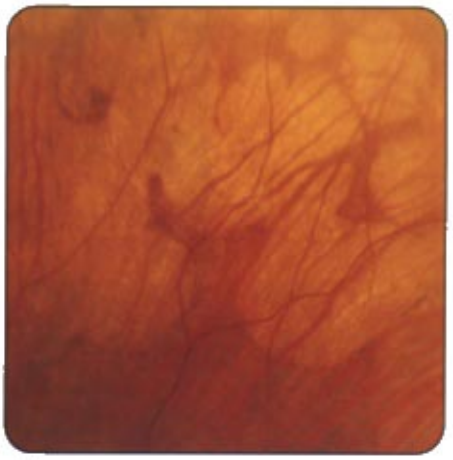

Fig. 12.157

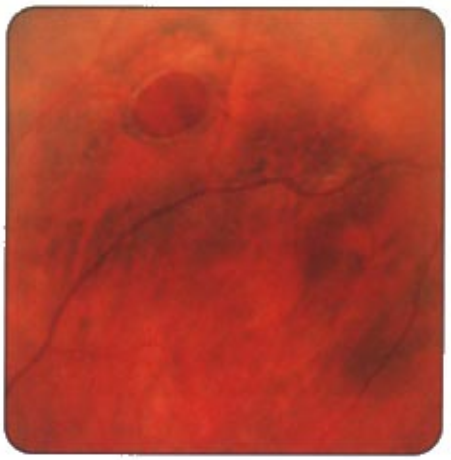

Fig. 12.158

Hereditary

End-stage Stargardt macular dystrophy

Stargardt macular dystrophy is an uncommon, recessively inherited, bilateral condition which starts during the teen years and results in severe visual loss within 5 years.

Signs

Several different appearances at the macula may be encountered:

- Isolated atrophic maculopathy (**Fig. 12.159a and b**).
- Atrophic maculopathy surrounded by fish-tail-shaped flecks (**Fig. 12.160a–d**).
- Atrophic maculopathy associated with diffuse flecks (**Fig. 12.161a and b**).
- Beaten-metal appearance.
- Bull's-eye pattern.

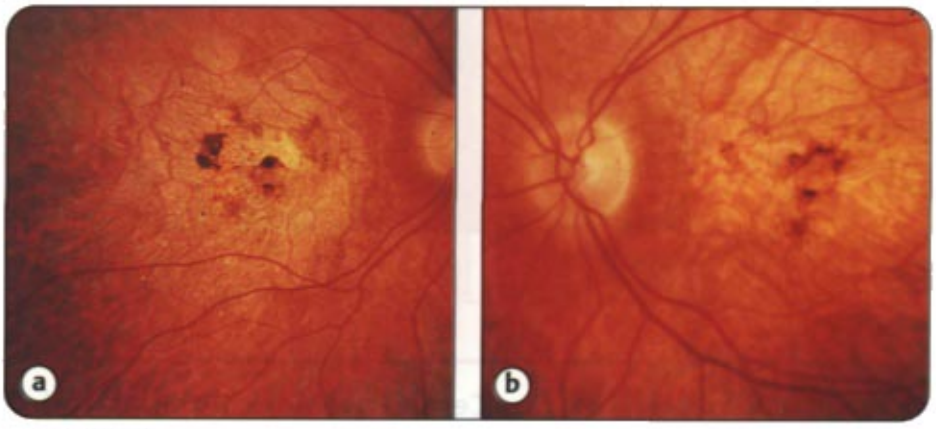

Fig. 12.159

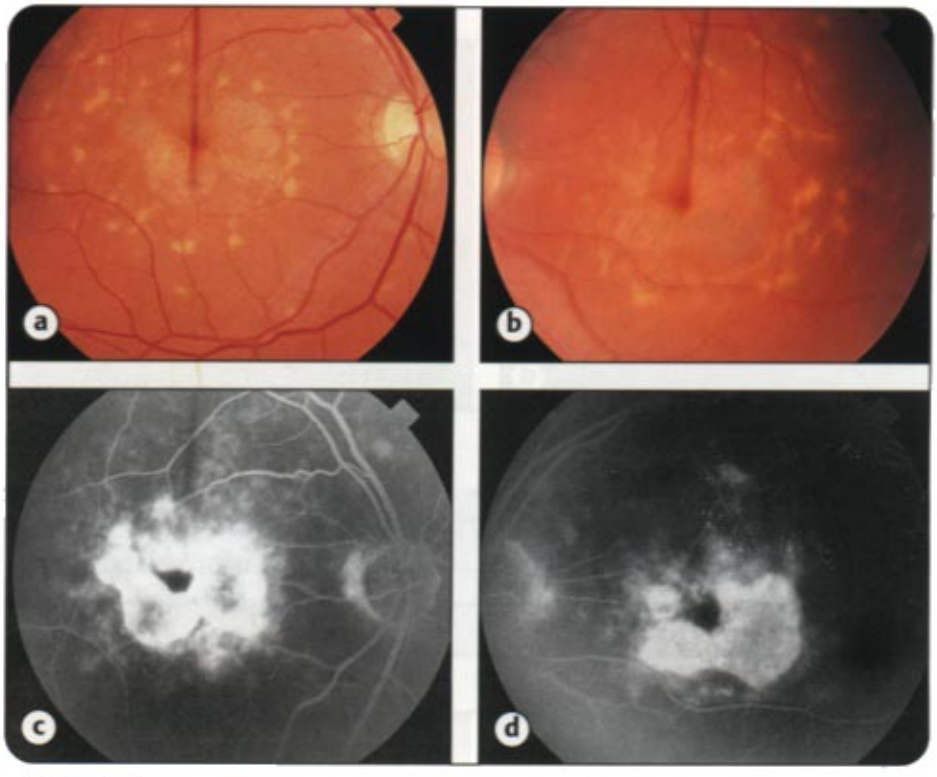

Fig. 12.160

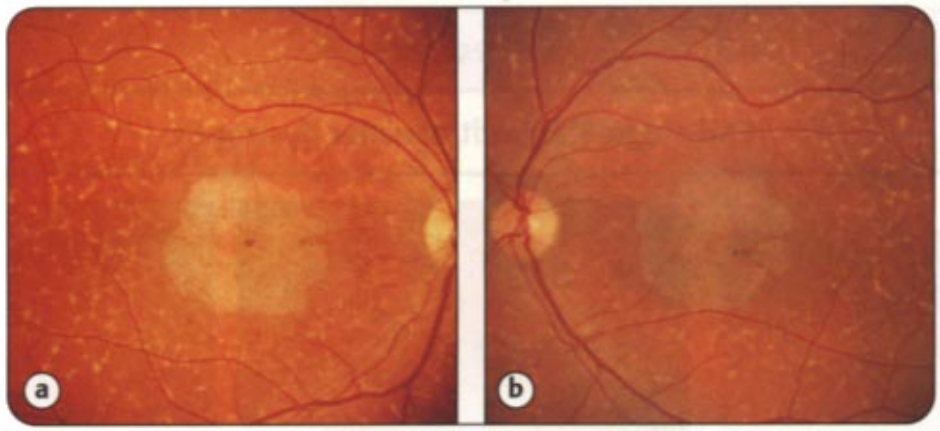

Fig. 12.161

Central areolar choroidal dystrophy

Central areolar choroidal dystrophy is a rare, dominantly inherited, bilateral condition which causes loss of central vision during the fifth decade of life.

Signs

- Circumscribed, atrophic macular lesions between one and three disc diameters in size which are not associated with drusen (**Fig. 12.162a and b**).

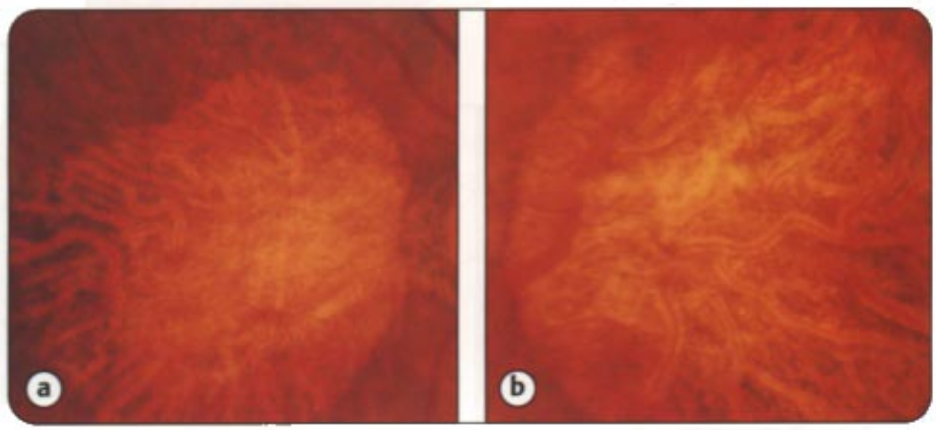

Fig. 12.162

Sorby macular dystrophy

Sorby macular dystrophy is a very rare, dominantly inherited, bilateral condition which causes loss of central vision between the second and fourth decades of life.

Signs

- Yellow–white confluent spots located along the arcades and nasal to the disc, associated with atrophic maculopathy (**Fig. 12.163**).
- Fibrotic macular scar (**Fig. 12.164**) with or without yellow spots.

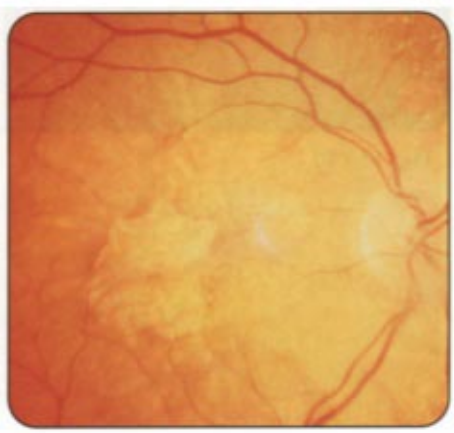

Fig. 12.163

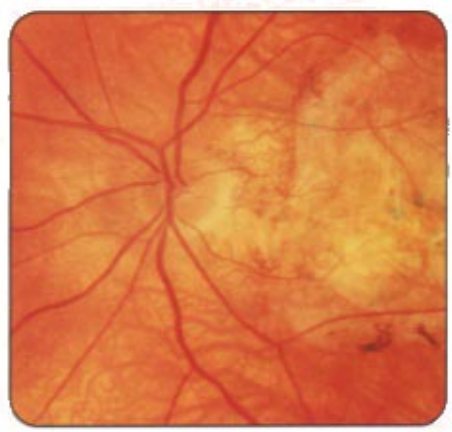

Fig. 12.164

End-stage Best disease

Best disease is a very rare, dominantly inherited condition which starts in early childhood.

Signs

- Atrophic maculopathy which may be asymmetric (**Fig. 12.165a and b**).
- Fibrotic (disciform) macular scar associated with choroidal neovascularization in some cases.

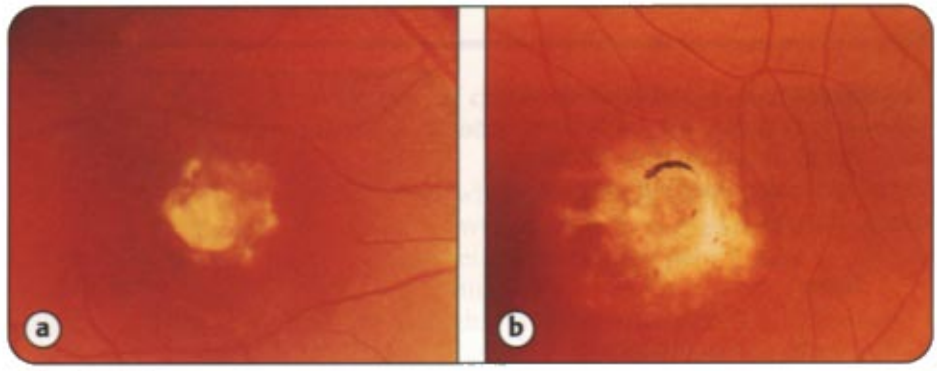

Fig. 12.165

End-stage North Carolina macular dystrophy

North Carolina macular dystrophy is a very rare, dominantly inherited, bilateral, slowly progressive condition which starts during the early teen years.

Signs

- Sharply circumscribed, atrophic macular lesions (**Fig. 12.166a and b**).

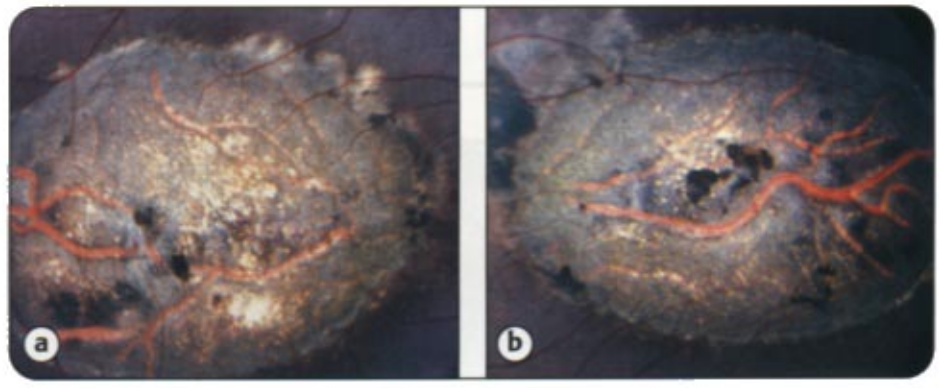

Fig. 12.166

BULL'S-EYE MACULOPATHIES

Signs

- Bull's-eye maculopathy is characterized by a central foveolar hyperpigmentation surrounded by an oval depigmented zone which is itself outlined by a ring of hyperpigmentation (**Fig. 12.167a**).
  On fluorescein angiography, the depigmented zone shows up as an RPE window defect (**Fig. 12.167b**).

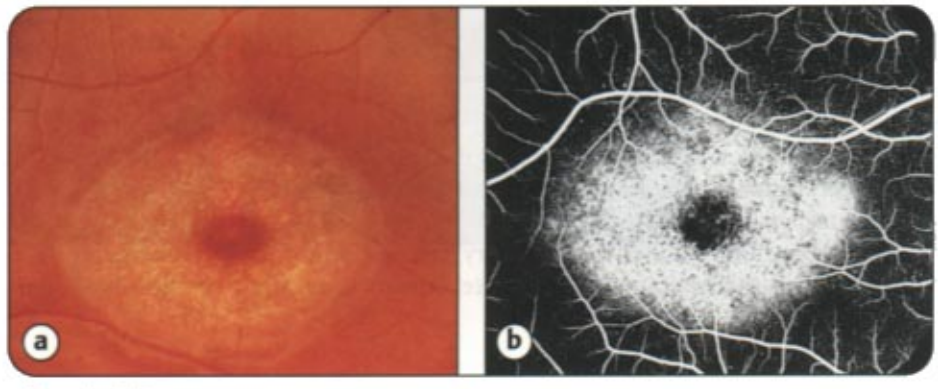

Fig. 12.167

Causes

Antimalarials

Chloroquine, if administered long term in high doses, may occasionally give rise to maculopathy. Hydroxychloroquine very rarely if ever causes retinal toxicity.

Signs

- Bull's-eye maculopathy (**Fig. 12.168a and b**) is a late feature which, in very advanced cases, may be associated with retinal vascular attenuation and peripheral pigmentary changes.

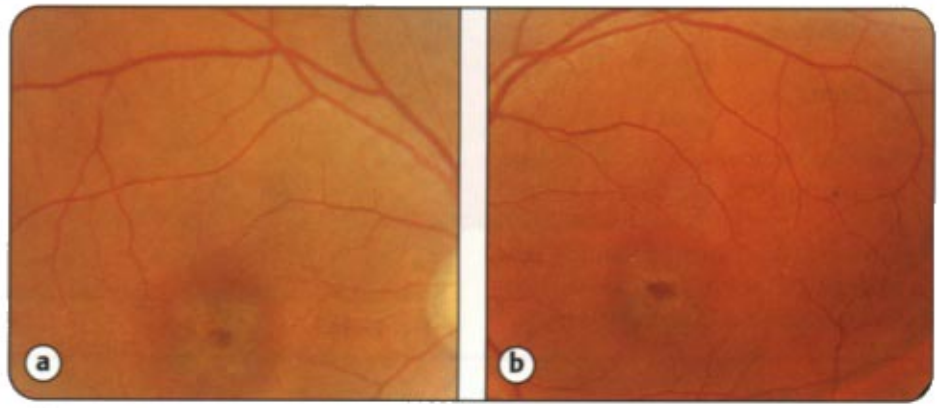

Fig. 12.168

Cone dystrophy

This is a very rare retinal dystrophy which presents between the first and third decades of life. Inheritance is usually either autosomal dominant or X-linked recessive.

Signs

- Bull's-eye maculopathy which may be associated with a golden reflex (**Fig. 12.169a and b**), temporal disc pallor, retinal vascular attenuation, and mild peripheral pigmentary changes.

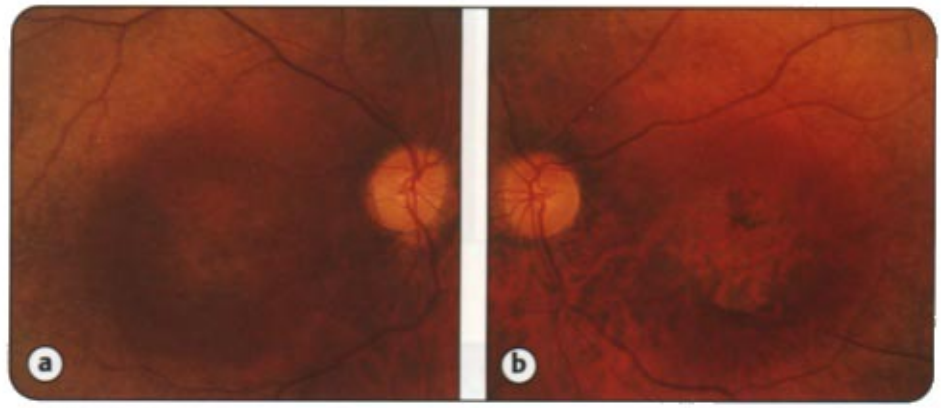

Fig. 12.169

Other causes

- Benign concentric annular macular dystrophy – a very rare, dominantly inherited condition which may cause a mild impairment of central vision.
- Inverse retinitis pigmentosa – a very rare condition in which the pigmentary changes are confined to the posterior pole.
- Fenestrated sheen dystrophy – a very rare, dominantly inherited condition which presents in young adults. Bull's-eye maculopathy is a late feature.
- Stargardt disease.
- Resolved acute idiopathic maculopathy.

EXUDATIVE MACULOPATHIES

Macular edema

Macular edema consists of increased fluid within the neuroretina at the macula and may by cystoid or noncystoid.

Signs of noncystoid macular edema

- Retinal thickening at the posterior pole which is not associated with cystoid spaces at the fovea.
- Fluorescein angiography shows intraretinal leakage during the late phase (*see* **Fig. 12.23d**).

Signs of cystoid macular edema

- Thickening of the fovea associated with microcysts (**Fig. 12.170a**).
- Fluorescein angiography shows a flower-petal pattern of leakage at the fovea during the late phases (**Fig. 12.170c–d**) which may be associated with leakage from the disc.

The definition of clinically significant macular edema in diabetic patients

- Retinal edema within 500 μm of the center of the fovea.
- Hard exudates within 500 μm of the fovea if associated with adjacent retinal thickening (which may be outside the 500-μm limit).
- Retinal edema that is one disc area (1500 μm^2) or larger, any part of which is within one disc diameter of the center of the fovea.

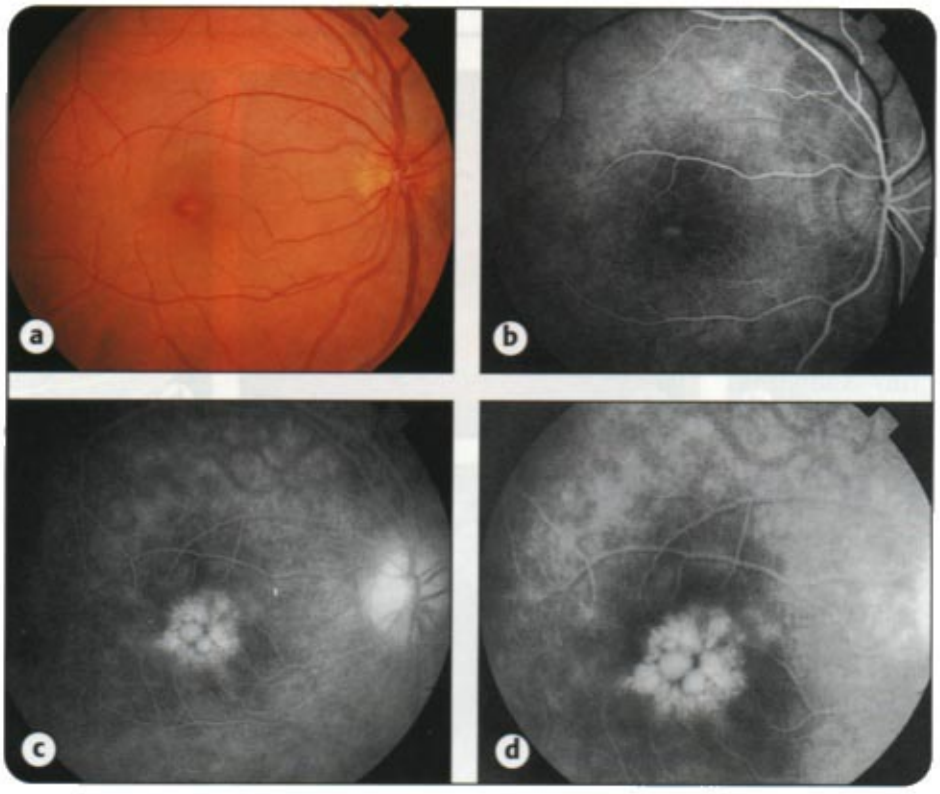

Fig. 12.170

Table 12.5 Causes of macular edema

Iatrogenic	**Retinal vascular disease**
• cataract surgery (Irvine–Gass syndrome)	• background diabetic retinopathy
• YAG laser capsulotomy	• retinal vein occlusion
• retinal detachment surgery	
• retinal laser photocoagulation	**Secondary to other maculopathies**
• retinal cryotherapy	• neovascular
• topical 2% epinephrine in aphakia	• preretinal macular fibrosis
	• vitreomacular traction syndrome
Chronic intraocular inflammation	
• chronic iridocyclitis	**Associated with hereditary dystrophies**
• intermediate uveitis	• retinitis pigmentosa
• birdshot retinochoroidopathy	• gyrate atrophy
• panuveitis	
• infectious endophthalmitis	**Miscellaneous**
	• high doses of nicotinic acid
	• familial dominant

Serous detachment of the neuroretina

In serous detachment of the neuroretina there is a separation of the neuroretina from the RPE by fluid.

Signs

- Unilateral, shallow elevation of the neuroretina at the posterior pole, with indistinct borders.
- In some cases, tiny subretinal precipitates may be seen.

Causes

- Idiopathic central serous retinopathy.

Figure 12.171a shows central serous retinopathy in which fluorescein angiography (**Fig. 12.171b–d**) demonstrates a progressive 'smoke-stack' leak under the neuroretina.

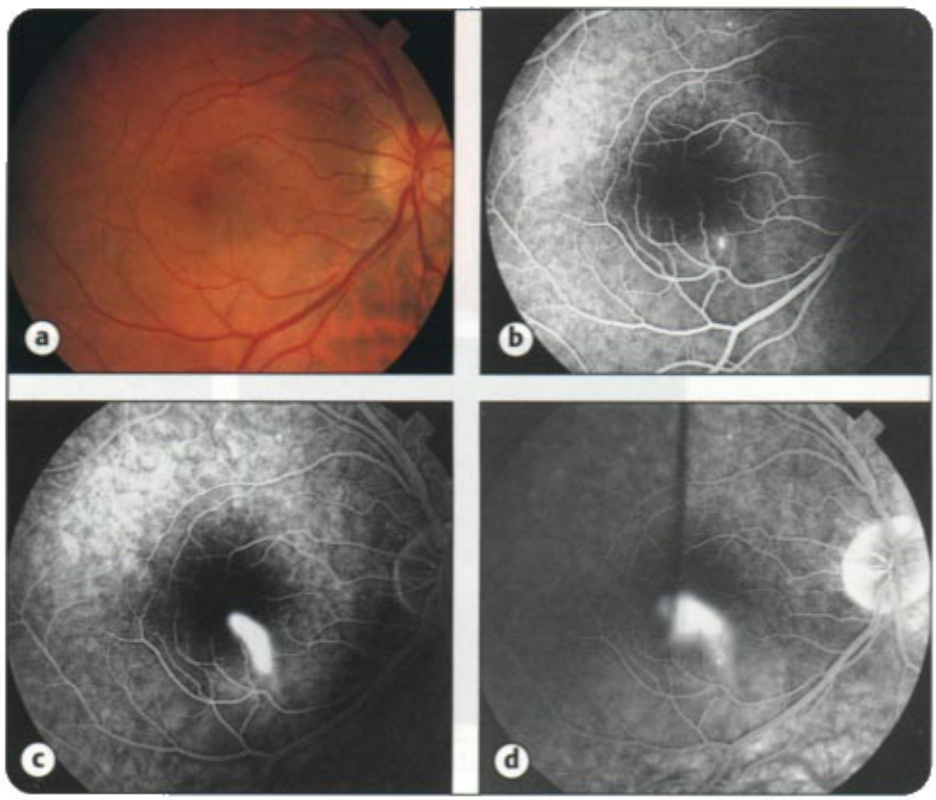

Fig. 12.171

Figure 12.172a shows central serous retinopathy in which fluorescein angiography (**Fig. 12.172b–d**) demonstrates a gradually increasing 'ink-blot' leak under the neuroretina.

- Detachment of the RPE.
- Optic disc pit (**Fig. 12.173a**) in which fluorescein angiography (**Fig. 12.173b–d**) demonstrates hyperfluorescence of the pit and transmitted choroidal fluorescence at the macula due to RPE atrophy secondary to longstanding serous detachment.
- Optic disc coloboma.
- Harada disease (*see* **Fig. 12.140a and b**).

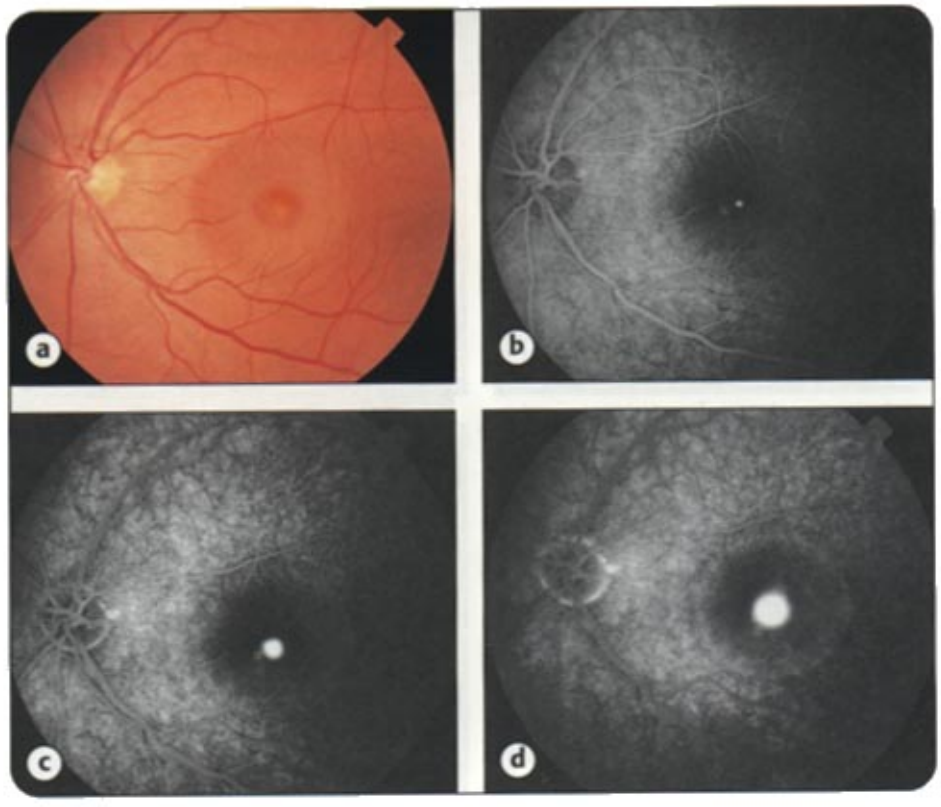

Fig. 12.172

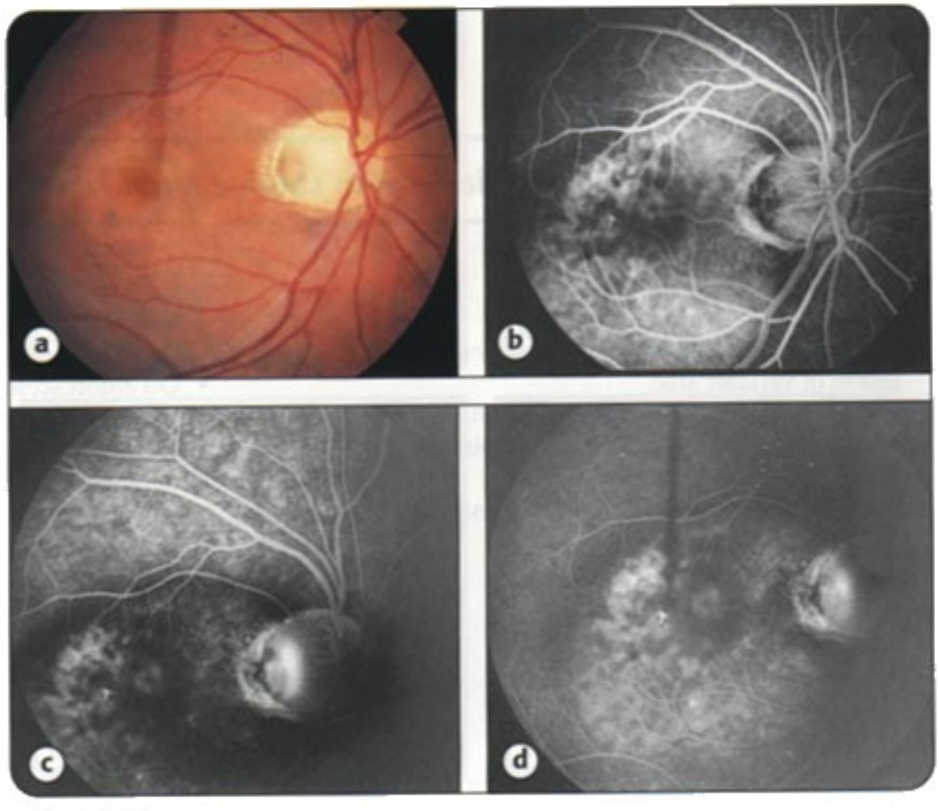

Fig. 12.173

Detachment of the RPE

In detachment of the RPE there is a separation of the RPE from Bruch membrane.

Signs

- Usually unilateral, sharply circumscribed, dome-shaped elevation at the posterior pole of varying size (**Fig. 12.174a**) with clear or cloudy sub-RPE fluid.
- Fluorescein angiography shows an early area of hyperfluorescence which increases in intensity but not in size during the later phases (**Fig. 12.174b–d**). The angiogram also shows extensive drusen and a large choroidal neovascular membrane adjacent to the RPE detachment.

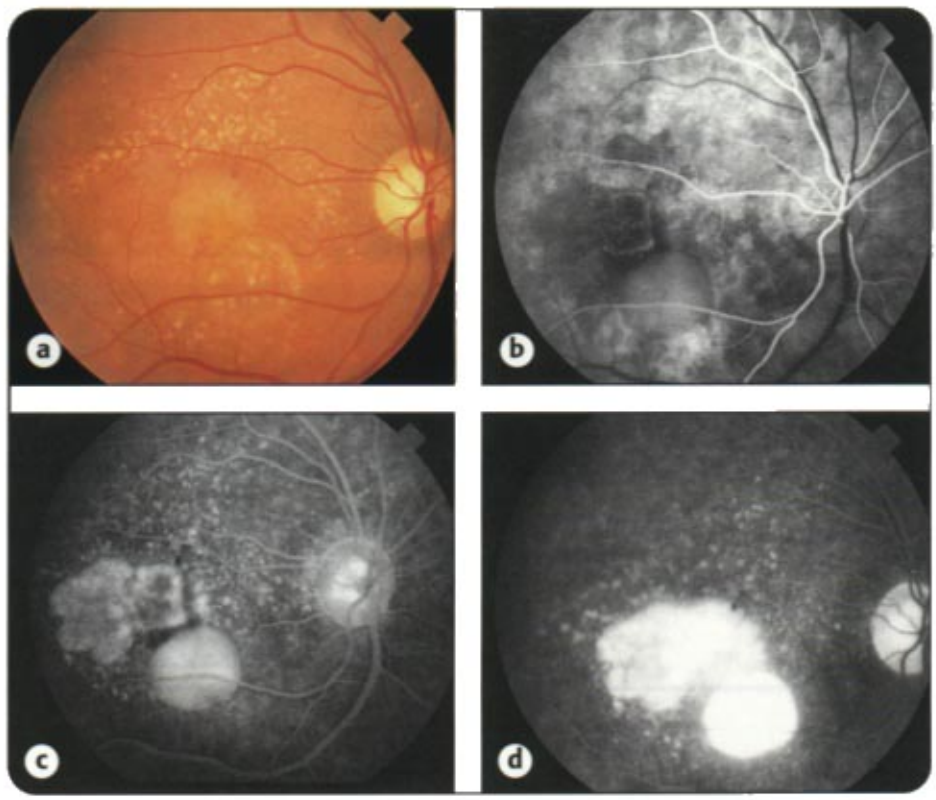

Fig. 12.174

Associated with choroidal neovascularization

Signs

- Gray–green or pinkish-yellow, slightly elevated subretinal lesion of variable size.
- If the lesion has broken into the subretinal space, it usually assumes a translucent, pale-pink or yellow–white appearance.
- Most membranes are difficult to identify clinically until they have caused secondary changes such as hemorrhagic detachment of the RPE or sensory retina, or subretinal exudation.
- On fluorescein angiography a neovascular membrane shows a characteristic lacy-filling pattern during the early phase of the angiogram, followed by an increase in fluorescence and late leakage.

Figure 12.175a–d shows a small, age-related, juxtafoveal choroidal neovascular membrane.

Figure 12.176a–d shows a large, age-related choroidal neovascular membrane.

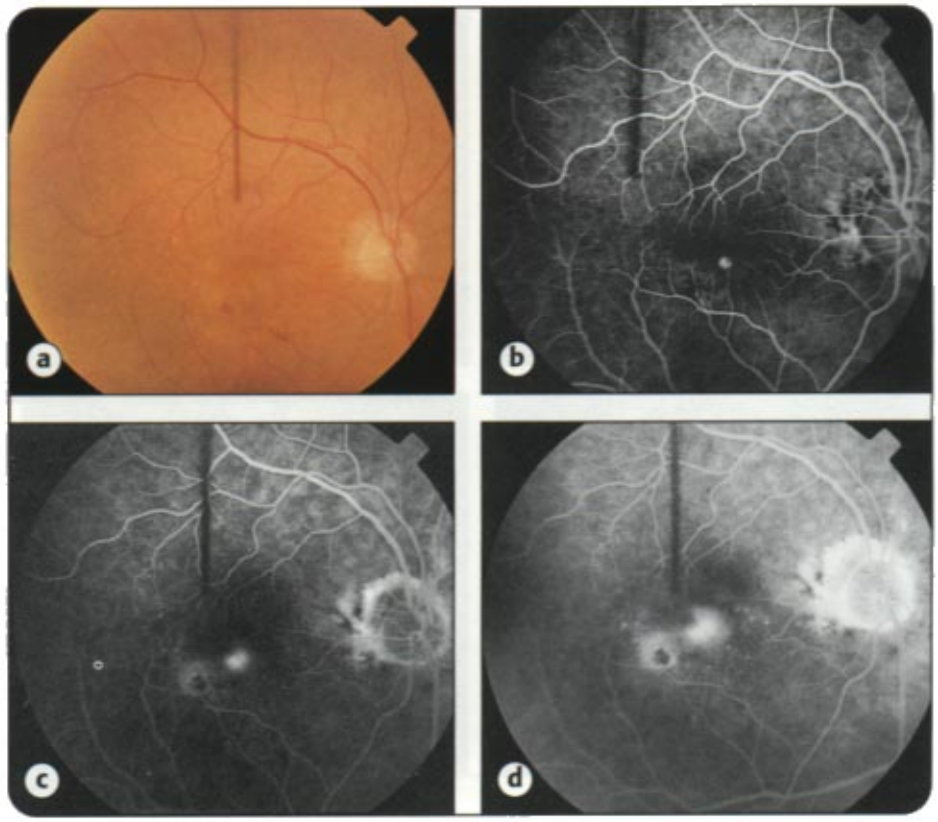

Fig. 12.175

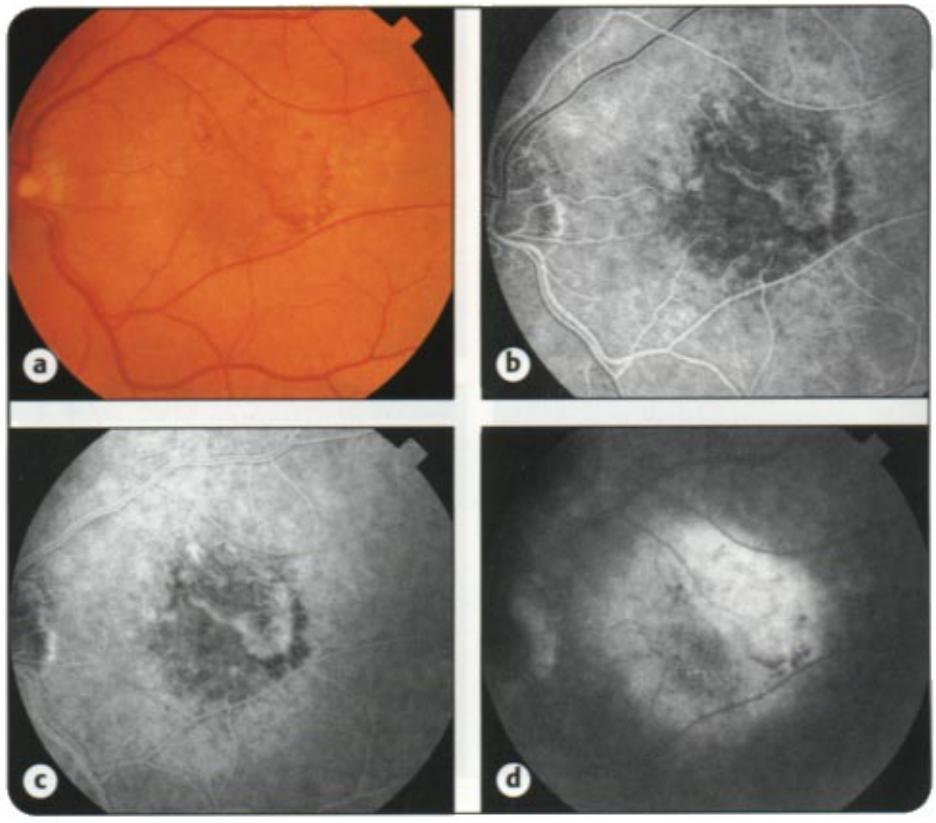

Fig. 12.176

Figure 12.177a–d shows the same eye as in **Figure 12.176** after laser photocoagulation.

Figure 12.178a shows a macular disturbance and a flame-shaped hemorrhage above the disc. **Figure 12.178b–d** shows a large subfoveal neovascular membrane.

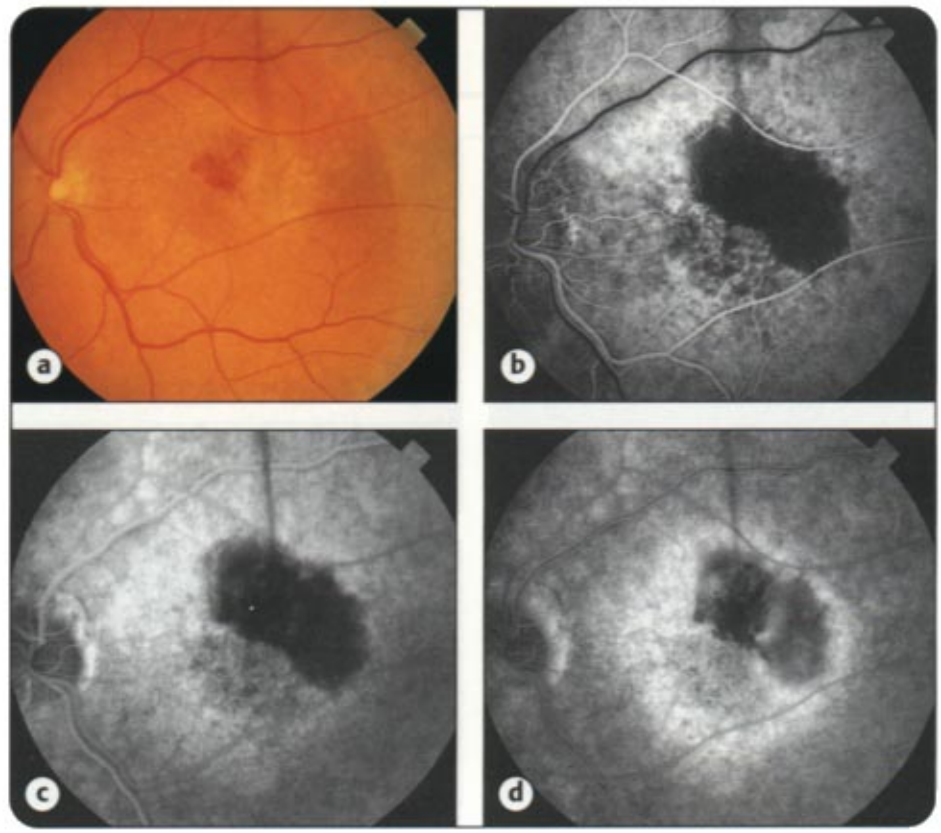

Fig. 12.177

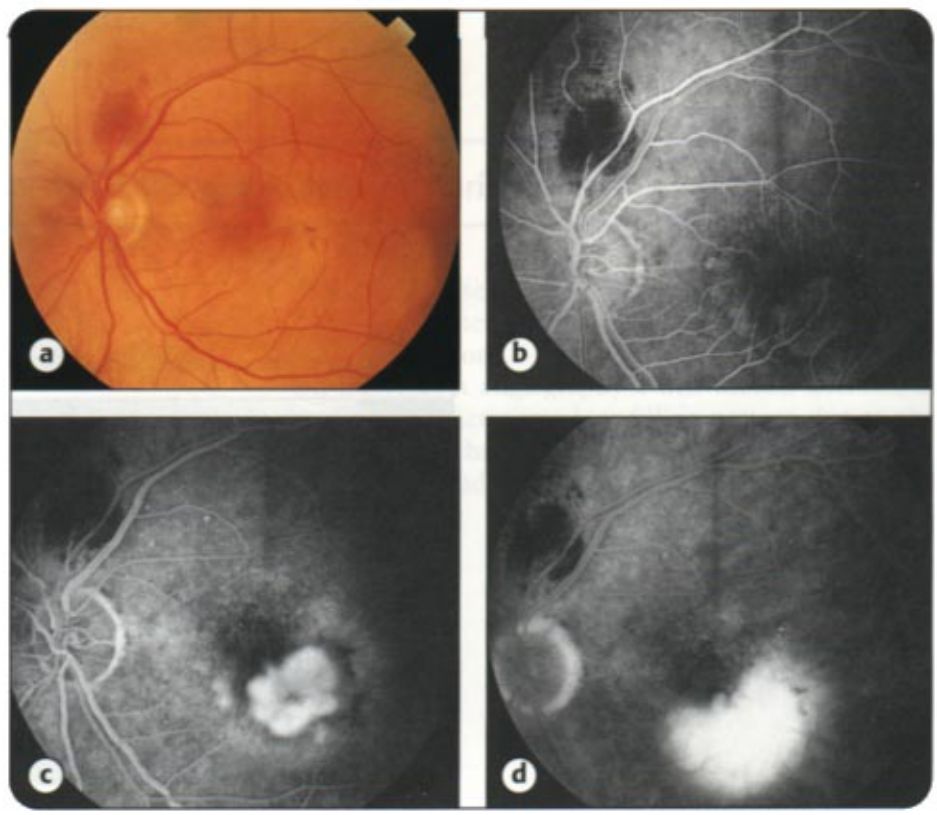

Fig. 12.178

Figure 12.179a–d shows a choroidal neovascular membrane in a highly myopic eye.

Figure 12.180a–d shows a tear (rip) of the RPE which appears as a retracted flap of RPE adjacent to a hypopigmented area of denuded Bruch membrane. This may occur spontaneously or following photocoagulation.

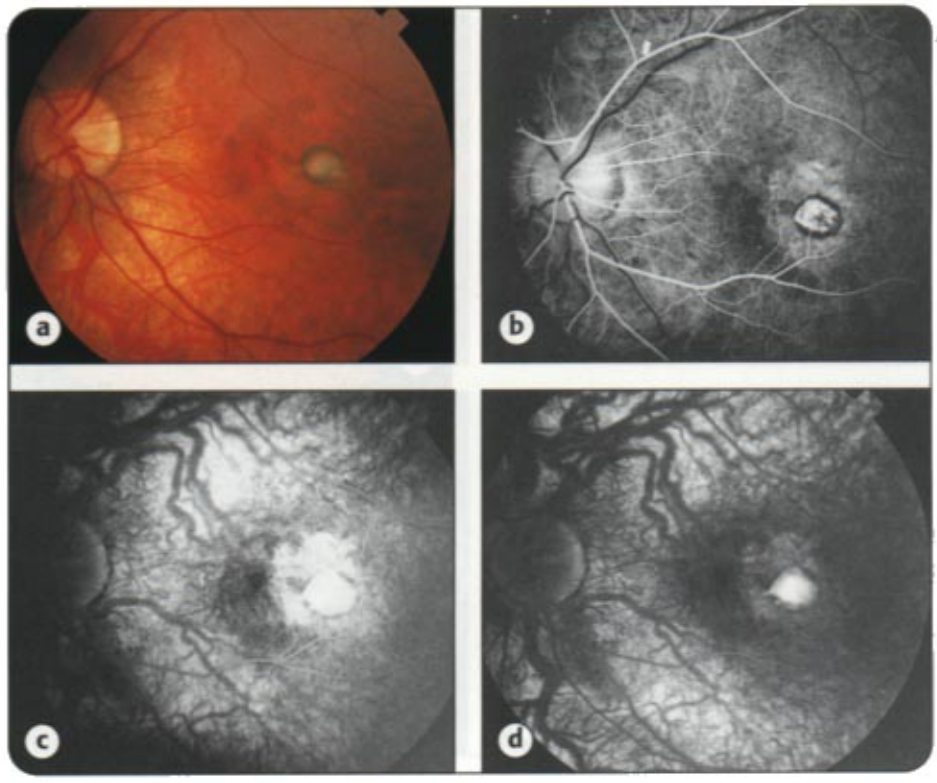

Fig. 12.179

Table 12.6 Causes of macular choroidal neovascularization

Degenerative
• age-related macular degeneration
• high myopia
• angioid streaks
• inappropriate laser photocoagulation
• traumatic choroidal rupture
Inflammatory
• presumed ocular histoplasmosis syndrome
• punctate inner choroidopathy
• serpiginous choroidopathy
• birdshot retinochoroidopathy
• multifocal choroiditis with panuveitis syndrome
• rubella retinopathy
Hereditary
• Sorsby pseudoinflammatory dystrophy
• Best disease
• optic disc drusen
Miscellaneous
• choroidal nevus
• idiopathic

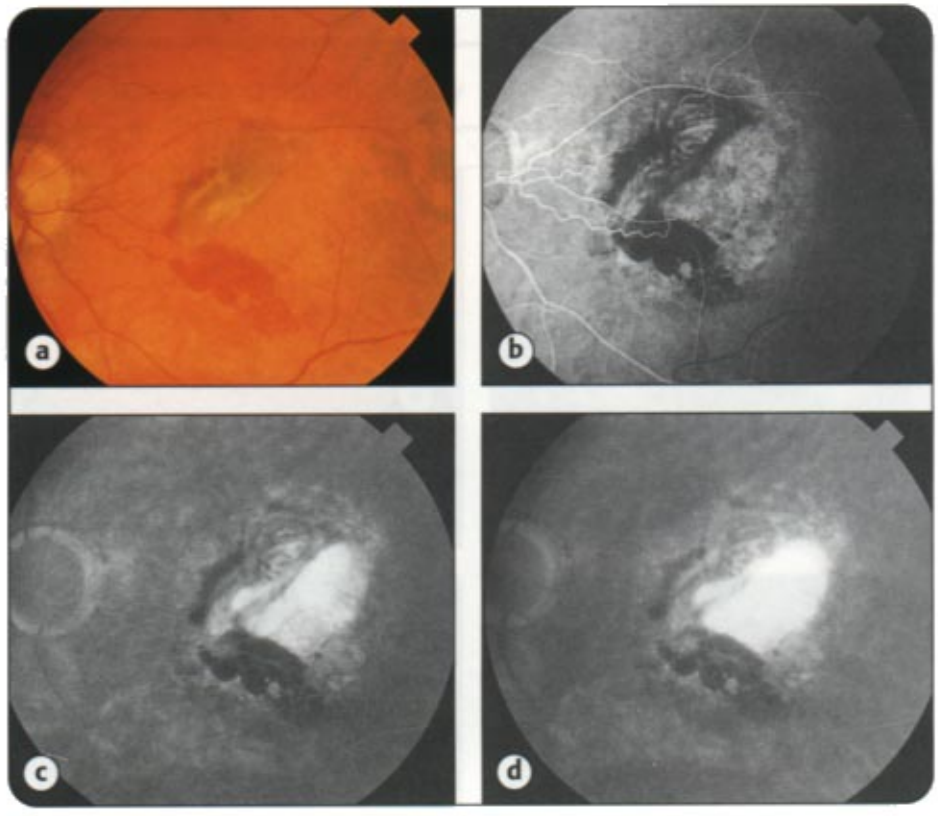

Fig. 12.180

Figure 12.181a–d shows juxtafoveal choroidal neovascularization in punctate inner choroidopathy.
Figure 12.182a–d shows subfoveal choroidal neovascularization in presumed ocular histoplasmosis.

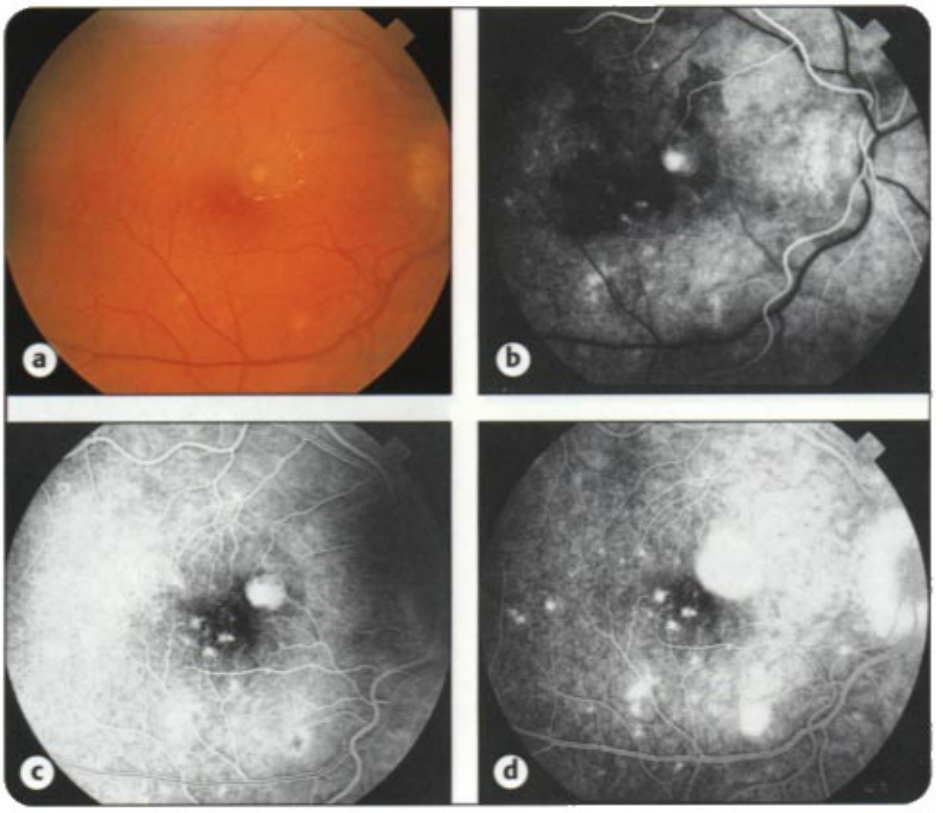

Fig. 12.181

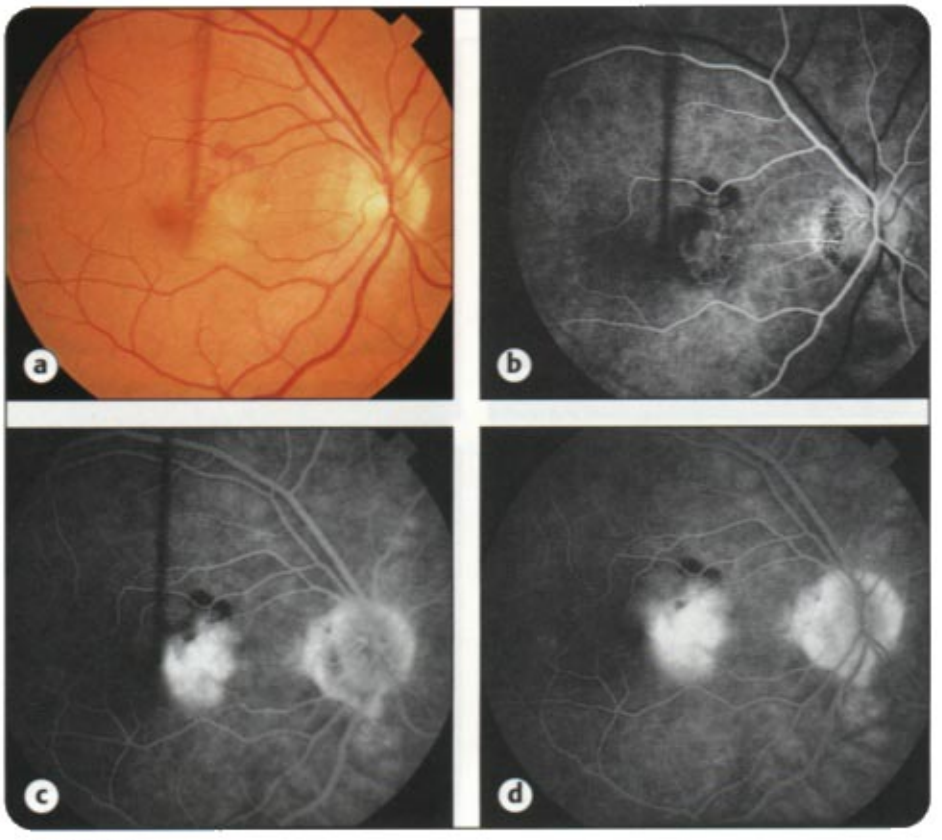

Fig. 12.182

Figure 12.183a–d shows the same eye as in **Figure 12.182** after laser photocoagulation to the choroidal neovascular membrane.

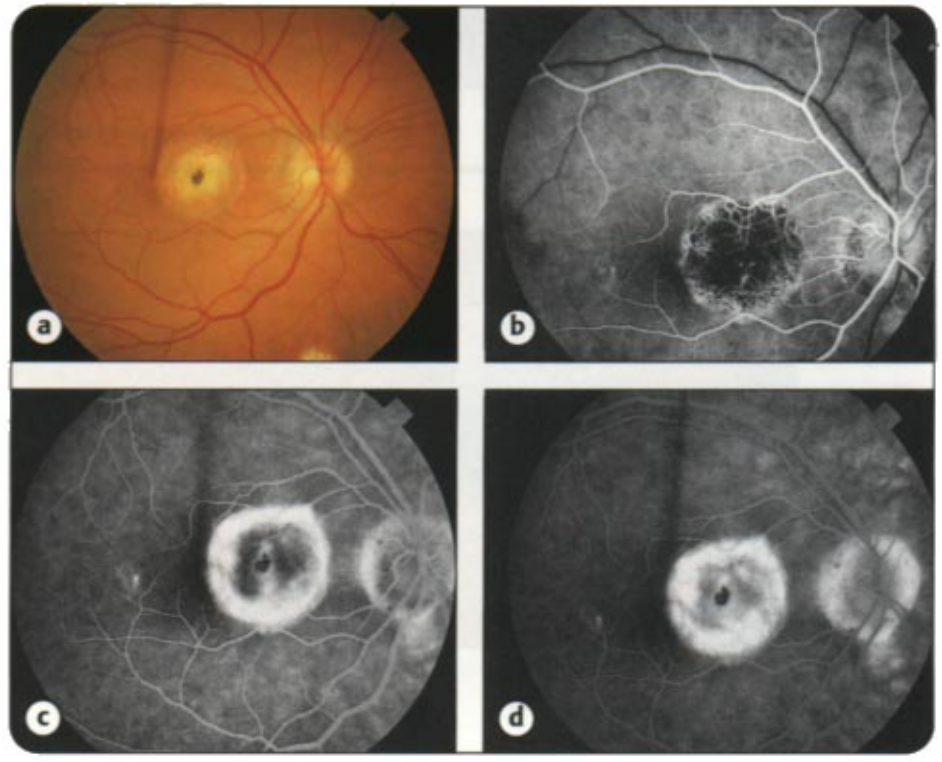

Fig. 12.183

Acute idiopathic maculopathy

Acute idiopathic maculopathy is a very rare, self-limiting condition which affects the young, causing sudden severe visual loss after a flu-like illness.

Signs

- Irregular neuroretinal detachment overlying a yellowish-gray thickening at the level of the RPE (**Fig. 12.184a**).
- A few hemorrhages may overlie the lesion.
- In contrast to central serous retinopathy, there is extensive fluorescein leakage (**Fig. 12.184b–d**).
- After resolution of symptoms, a bull's-eye appearance at the macula persists.

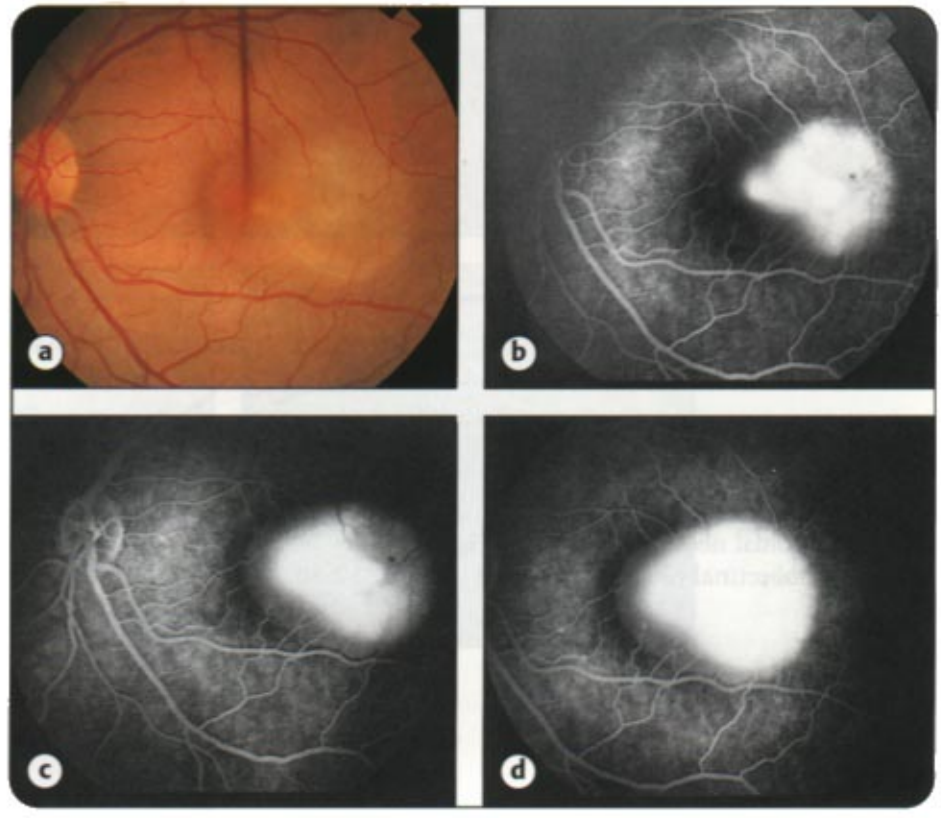

Fig. 12.184

FIBROTIC MACULOPATHIES

Preretinal macular fibrosis

Preretinal macular fibrosis is a common condition which is caused by contraction of an epiretinal membrane over the macula.

Signs (in order of severity)

- Transparent membrane – fine retinal striae and vascular tortuosity (cellophane maculopathy) (**Fig. 12.185a**), which is seen better on fluorescein angiography (**Fig. 12.185b**).
- Opaque membrane obscuring some of the blood vessels or RPE (macular pucker) (**Fig. 12.186**).
- Frequently the wrinkles radiate from a point called the epicenter (**Fig. 12.187**).

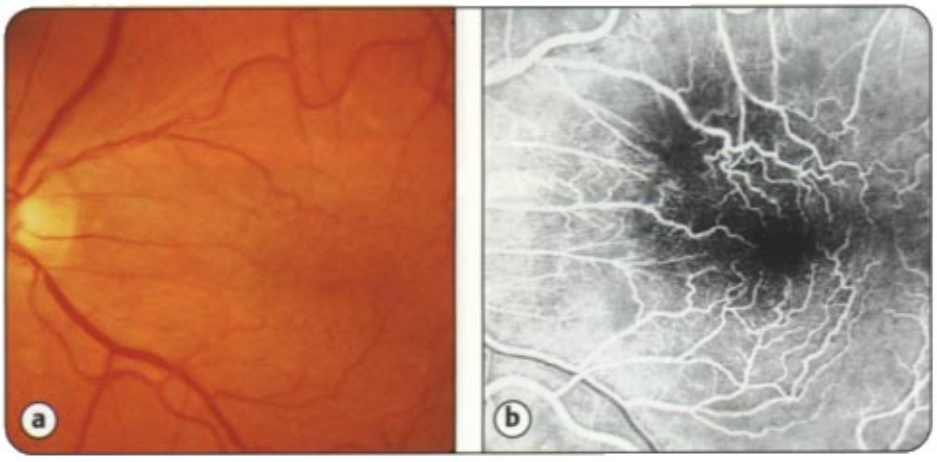

Fig. 12.185

Causes

- Idiopathic.
- Chronic intraocular inflammation.
- Retinal vascular disorders.
- Iatrogenic – scleral buckling procedures, retinal cryotherapy, and retinal photocoagulation.

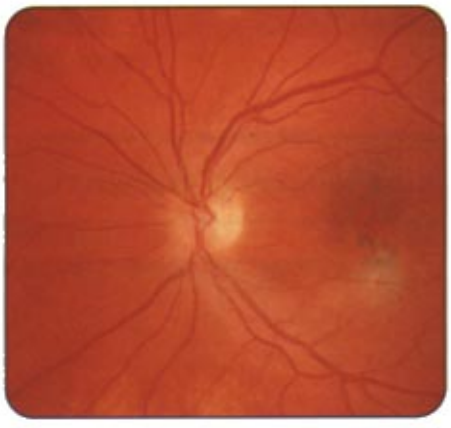

Fig. 12.186

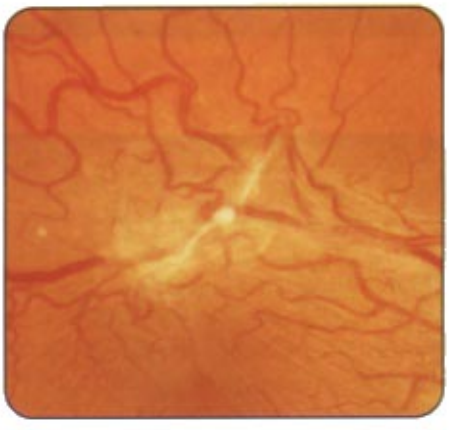

Fig. 12.187

Subretinal macular fibrosis

Causes

- End-stage of exudative macular degeneration associated with choroidal neovascularization (**Fig. 12.188**).
- Diffuse subretinal fibrosis syndrome (*see* **Fig. 12.86**).

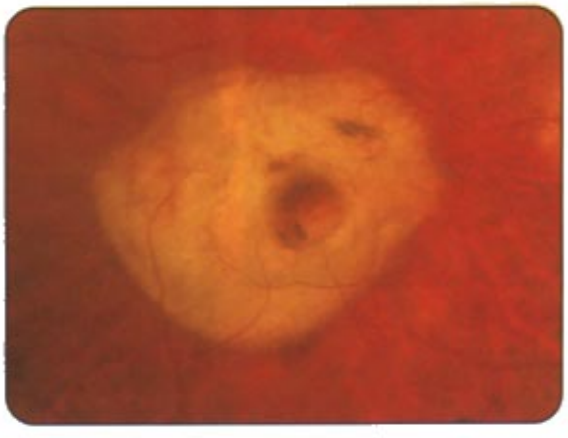

Fig. 12.188

MACULAR HOLE

Idiopathic macular hole

Idiopathic macular hole is a common, age-related condition which is eventually bilateral in about 15% of cases.

Signs

- Stage 1 – yellow foveolar spot or perifoveal ring, associated with loss of the foveolar depression.
- Stage 2 – central round foveal defect with a rim of elevated retina.
- Stage 3 – central round foveal defect with a central operculum that is smaller than the hole.
- Stage 4 – central foveal defect with complete separation of the operculum.
- Multiple yellow deposits at the level of the RPE are seen within the hole (**Figs 12.189, 12.190a**).
- Fluorescein angiography shows a corresponding circular zone of hyperfluorescence due to thinning of the RPE (**Fig. 12.190b–d**).

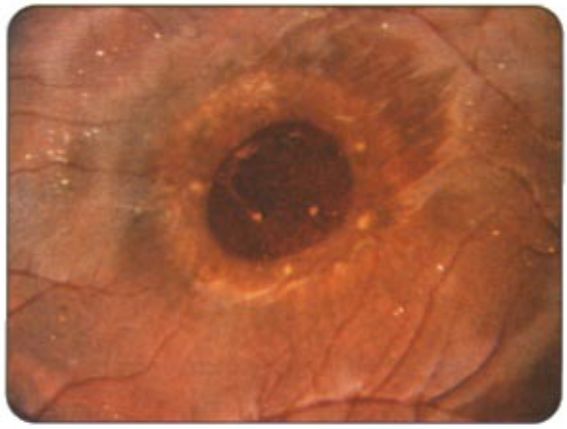

Fig. 12.189

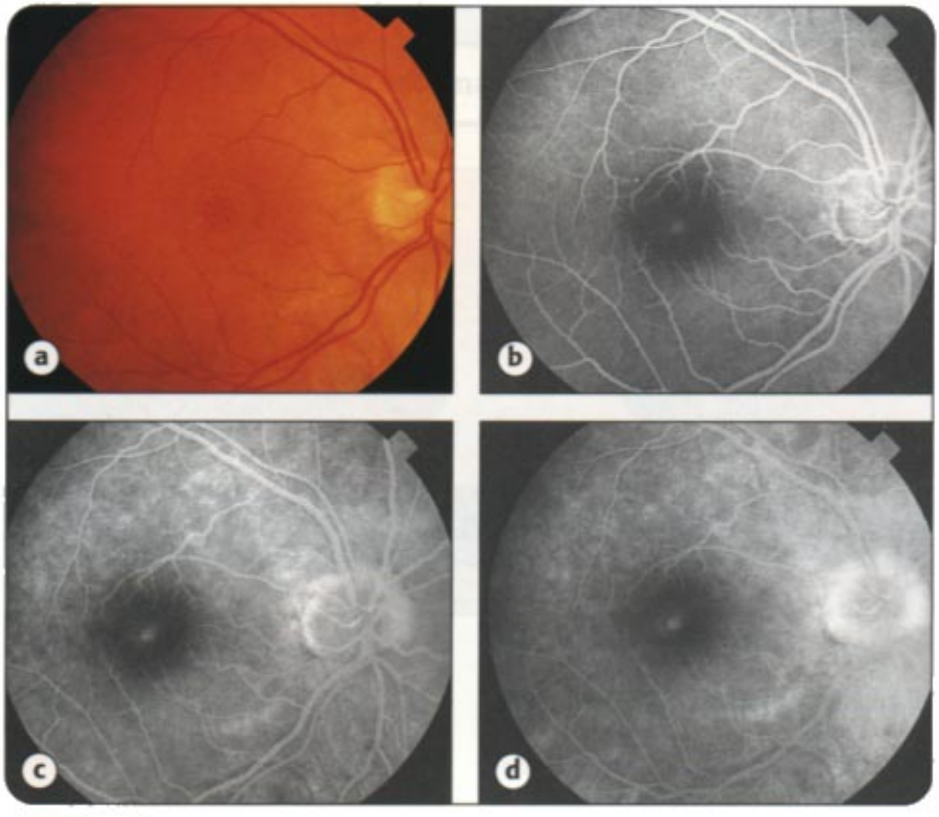

Fig. 12.190

Other macular holes

- Severe myopia may be associated with a full-thickness macular hole. This may give rise to retinal detachment which is mainly confined to the posterior pole (*see* **Fig. 12.154**).
- Severe blunt ocular trauma may give rise to Berlin edema (commotio retinae) which may subsequently result in the formation of either an outer lamellar hole or a full-thickness macular hole (**Fig. 12.191**).
- An inner lamellar hole is caused by chronic cystoid macular edema.

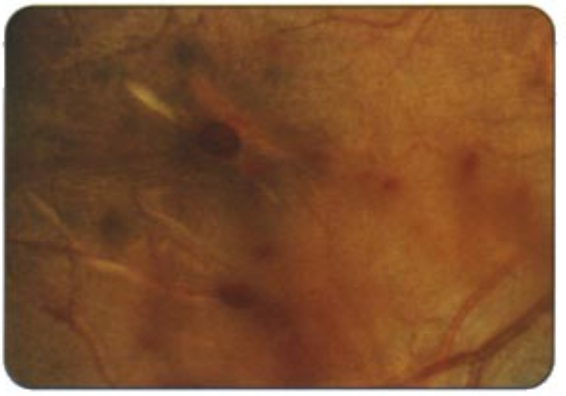

Fig. 12.191

Macular pseudohole

Causes

- A round discontinuity in an epiretinal membrane overlying the macula (**Fig. 12.192**).
- In eyes with rhegmatogenous detachment involving the posterior pole, a macular pseudohole (**Fig. 12.193**) may be seen because of the thinness of the retina at the fovea.

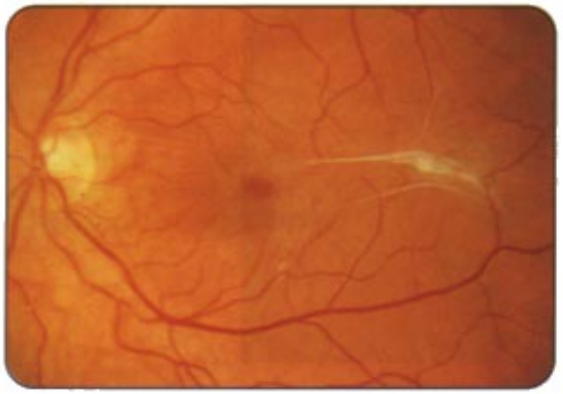

Fig. 12.192

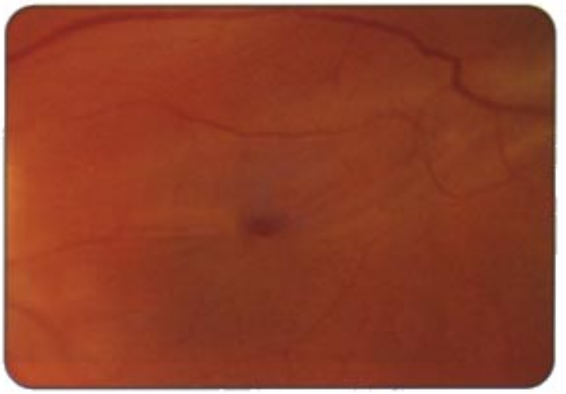

Fig. 12.193

CRYSTALLINE MACULOPATHIES

Signs

- Multitude of tiny glistening crystals at the posterior pole.
- In some cases the crystals have a perifoveal distribution.
- Patients with cystinosis and Bietti retinal dystrophy may also show corneal crystals.

Figure 12.194 shows tamoxifen maculopathy.
Figure 12.195 shows perifoveal distribution of canthaxanthin deposits.
Figure 12.196 shows a more diffuse distribution of crystals in Bietti retinal dystrophy.

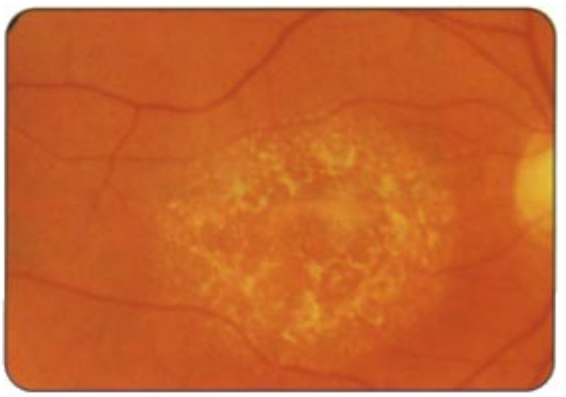

Fig. 12.194

Table 12.7 Causes of crystalline maculopathy

Drug-induced
• tamoxifen
• canthaxanthin
• talc
• methoxyflurane
Metabolic disorders
• cystinosis
• primary oxalosis type I
Eponymous conditions
• Bietti retinal dystrophy
• Sjögren–Larsson syndrome

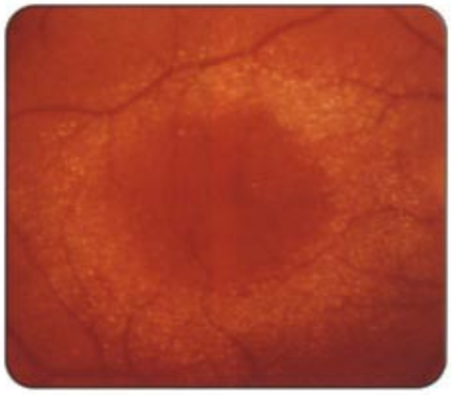

Fig. 12.195

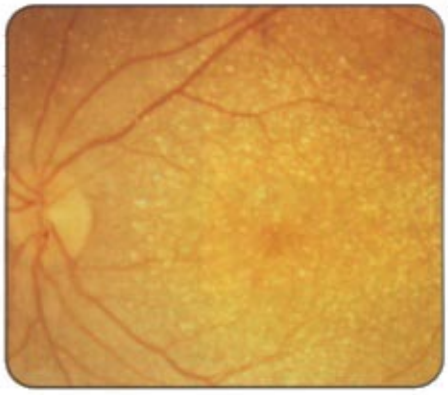

Fig. 12.196

COLORED MACULAR LESIONS

Egg-yolk lesions

Best vitelliform macular dystrophy

Best vitelliform macular dystrophy is a very rare, dominantly inherited condition which starts in childhood. The lesions may be single or multiple, unilateral or bilateral, and, if bilateral, they may be asymmetric.

Stages (in chronological order)

- Previtelliform – normal fundus but abnormal EOG.
- Vitelliform – egg-yolk macular lesion (**Fig. 12.197a and b**).
- Pseudohypopyon – partial absorption of lesion (**Fig. 12.198a and b**).
- Vitelliruptive – scrambled-egg appearance (**Fig. 12.199a and b**).
- End stage (*see* **Fig. 12.165a and b**).

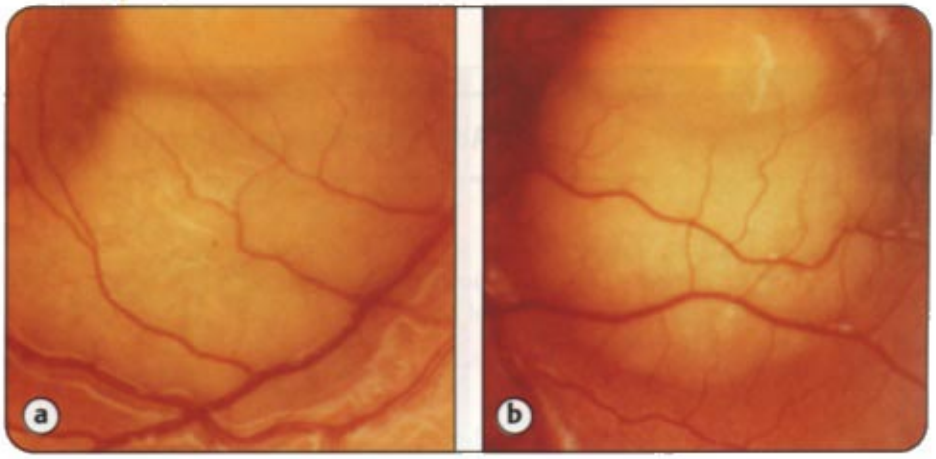

Fig. 12.197

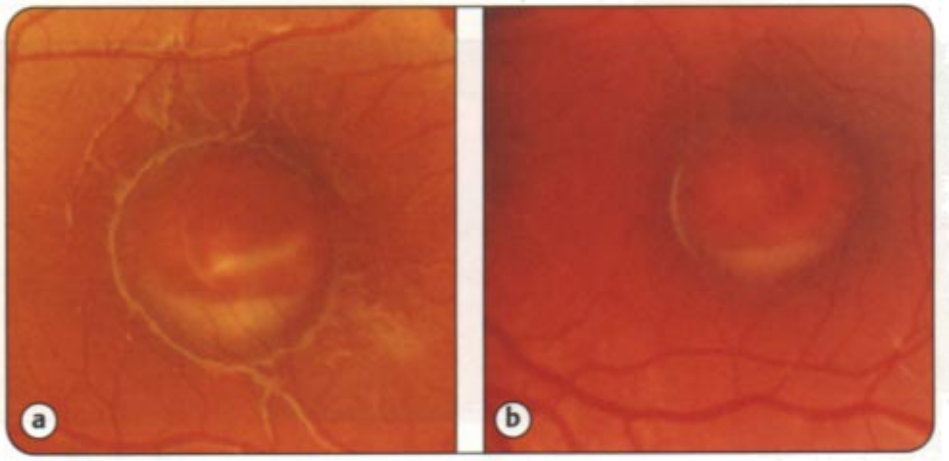

Fig. 12.198

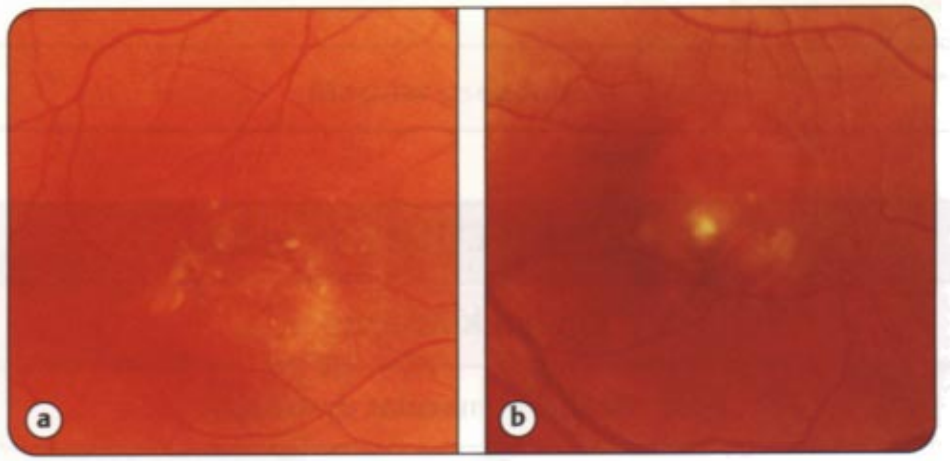

Fig. 12.199

Adult vitelliform macular dystrophy

Adult vitelliform macular dystrophy is an uncommon, bilateral, relatively innocuous condition which presents between the fourth and fifth decades of life.

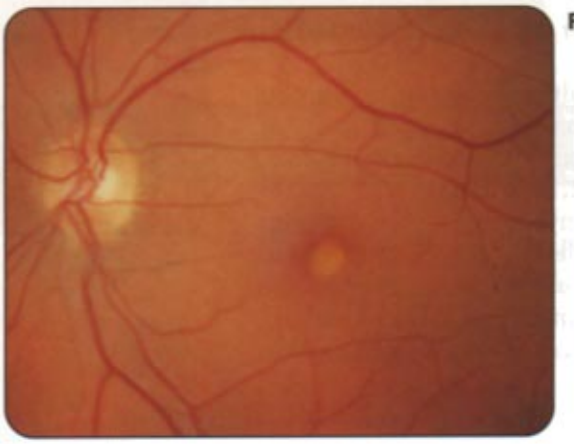

Fig. 12.200

Signs

- Symmetric, small, round, slightly elevated, yellow subretinal lesions at the fovea (**Fig. 12.200**).

Differential diagnosis

- Large drusen.
- Small RPE detachment.

Cherry-red spot lesions

Signs

- Round red foveal area surrounded by a larger concentric area of retinal pallor.

Figure 12.201 shows a central retinal artery occlusion with an intact cilioretinal artery.
Figure 12.202 shows a cherry-red spot in Tay–Sachs disease.

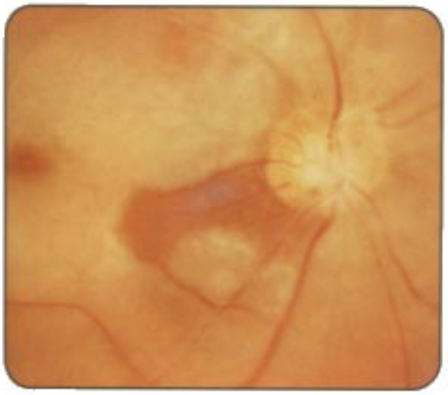

Fig. 12.201

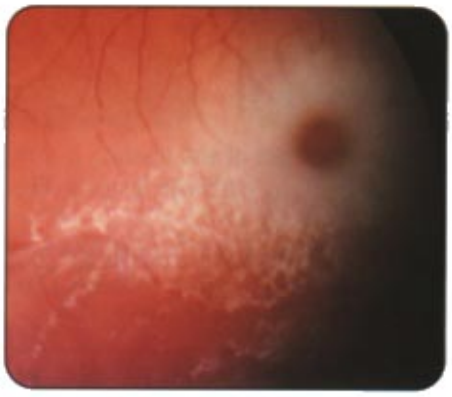

Fig. 12.202

Table 12.8 Causes of cherry-red spot at macula

- central retinal artery occlusion
- Tay–Sachs disease
- Sandhoff disease
- Niemann–Pick disease
- generalized gangliosidosis
- sialidosis types I and II

Dark lesions

Dark macular lesions may be unilateral (**Fig. 12.203**) or bilateral (**Fig. 12.204a and b**) and are most frequently caused by congenital toxoplasmosis.

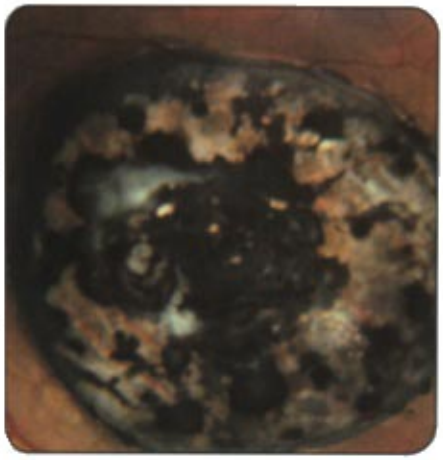

Fig. 12.203

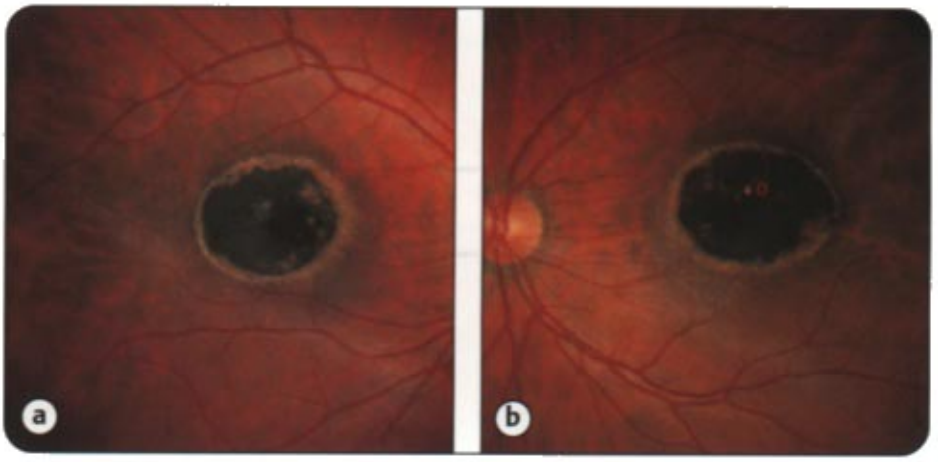

Fig. 12.204

SOLITARY FUNDUS LESIONS

Dark lesions

Congenital RPE hypertrophy

Signs

- Circular, dark-gray or black, placoid lesion, one to two disc diameters in size, with smooth (**Fig. 12.205**) or scalloped borders.
- A nonpigmented halo may be present near the margin of the lesion.
- Occasionally, hypopigmented lacunae may be present with the lesions (**Figs 12.206, 12.207**).
- Occasionally, most of the lesion itself is not pigmented but is outlined by a thin rim of pigment and may resemble a focus of old chorioretinitis (**Fig. 12.208**).

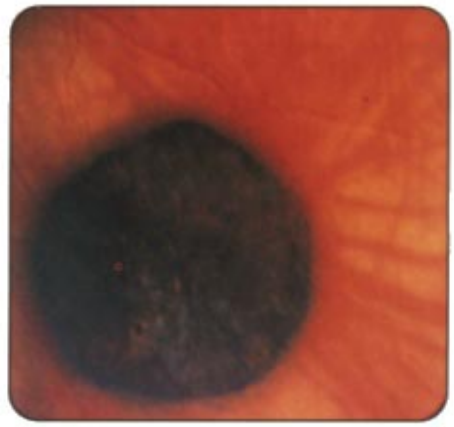

Fig. 12.205

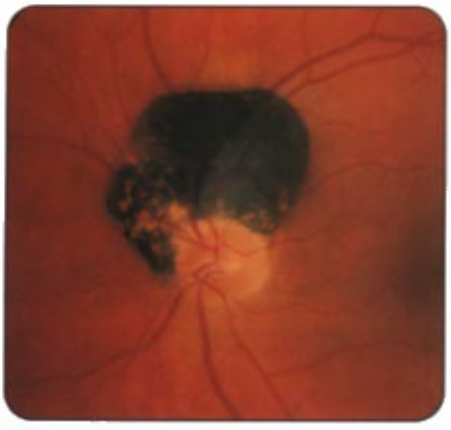

Fig. 12.206

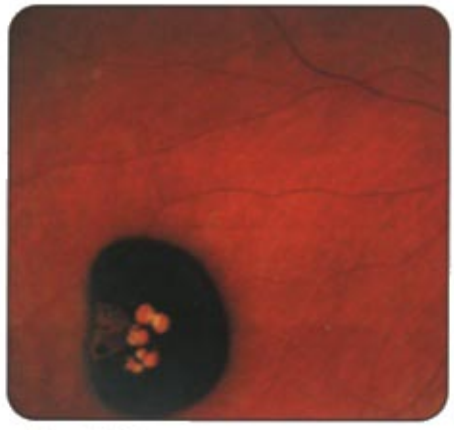

Fig. 12.207

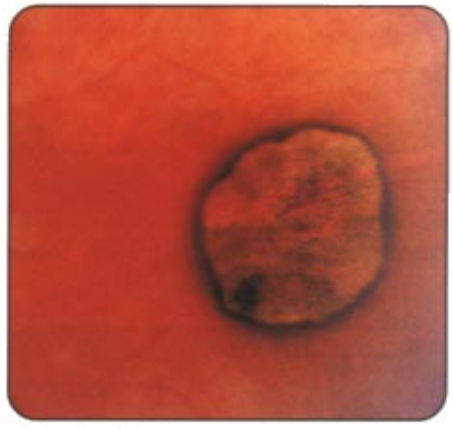

Fig. 12.208

Choroidal nevus

Signs

- Flat or minimally elevated, circular, slate-gray lesion less than 5 mm in diameter (**Figs 12.209, 12.210**).
- Surface drusen is a a common finding in longstanding lesions (**Figs 12.211, 12.212**).
- Secondary choroidal neovascularization may occur occasionally.

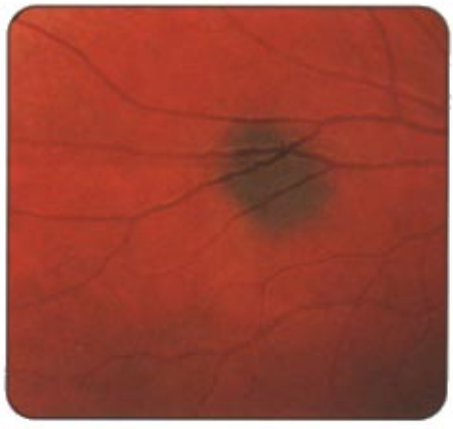

Fig. 12.209

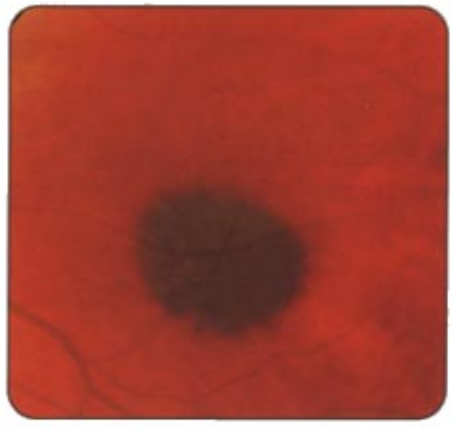

Fig. 12.210

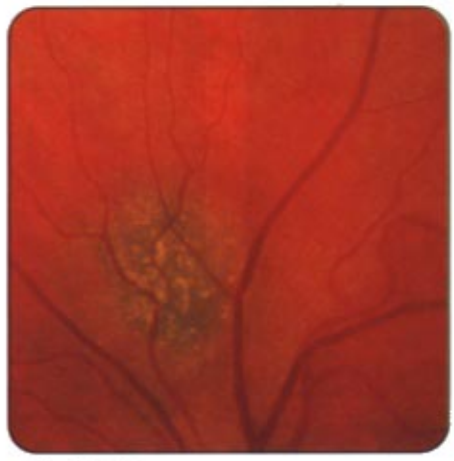

Fig. 12.211

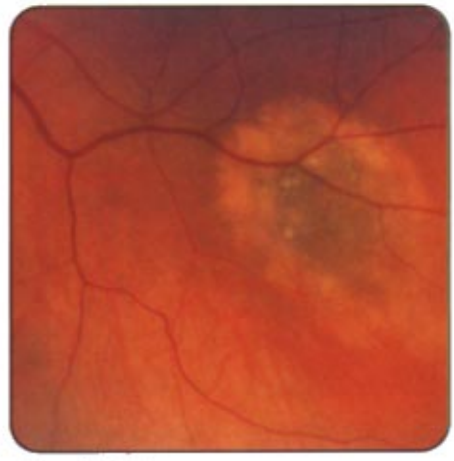

Fig. 12.212

Choroidal melanoma

Signs

- A typical melanoma is a pigmented, elevated, oval-shaped mass (**Figs 12.213–12.215**).
- In some cases the tumor is nonpigmented (**Figs 12.216, 12.217a**).
- Fluorescein angiography (**Fig. 12.217b and c**) shows the normal retinal blood vessels overlying the tumor as well as the vessels within the tumor (double circulation). The late phase of the angiogram shows leakage of dye from the tumor vessels (**Fig. 12.217d**).
- Secondary exudative retinal detachment is frequent (**Fig. 12.218**).
- Other variable features include orange (lipofuscin) pigment on the surface of the tumor and choroidal folds.

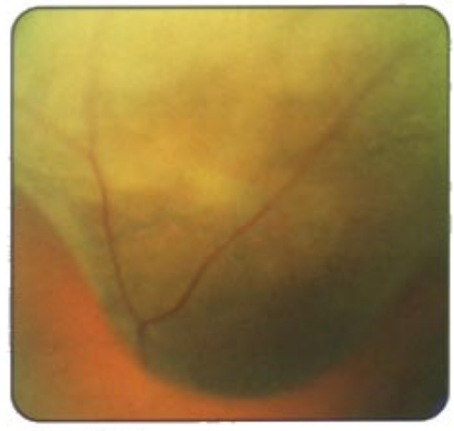

Fig. 12.213

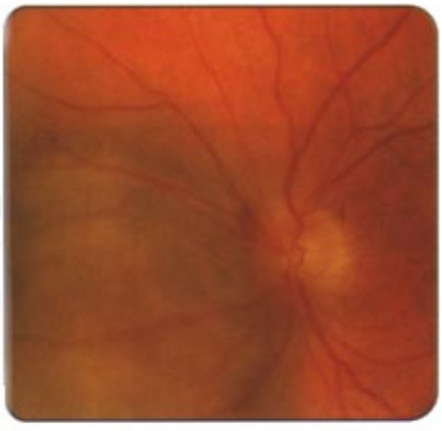

Fig. 12.214

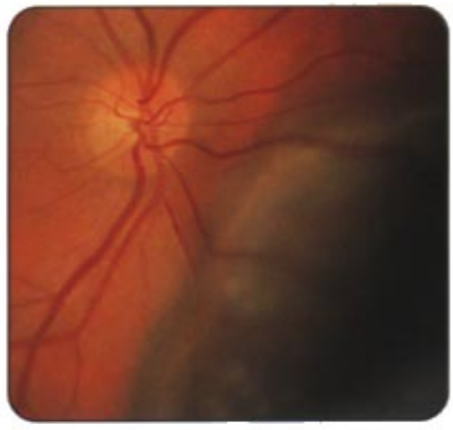

Fig. 12.215

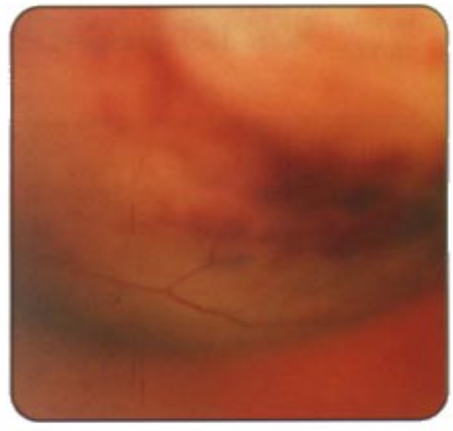

Fig. 12.216

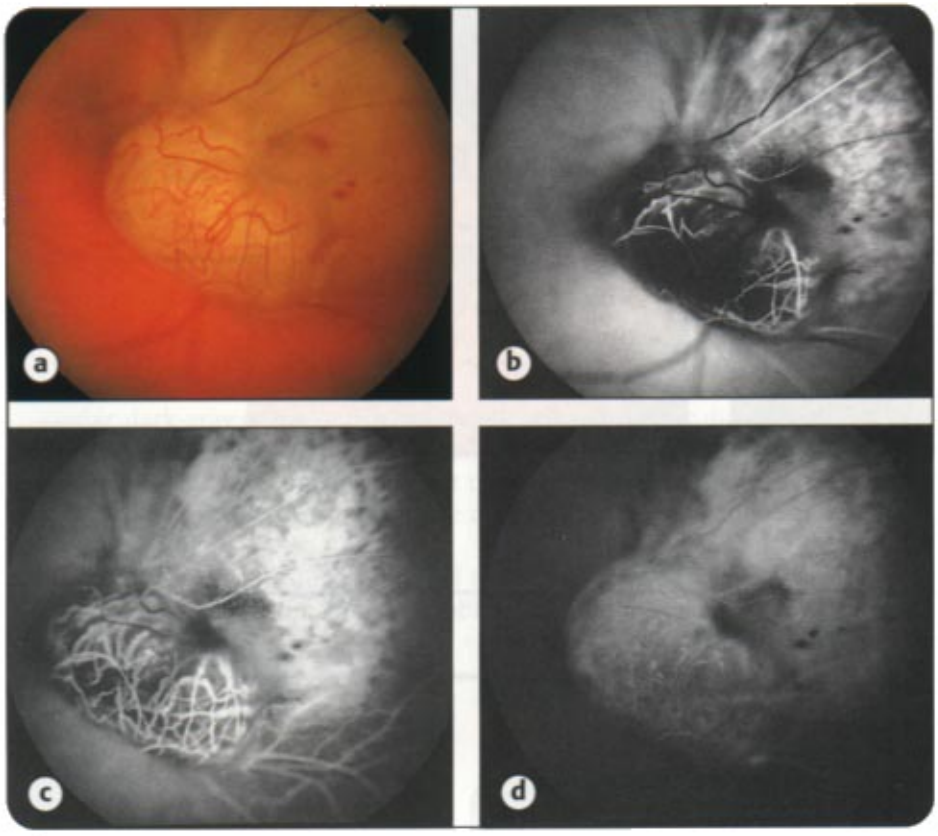

Fig. 12.217

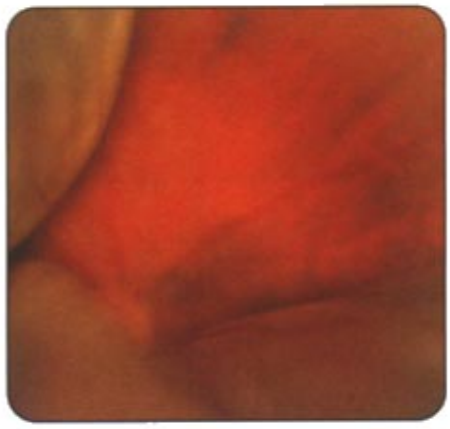

Fig. 12.218

RPE hamartoma

Signs

- Nodular, jet-black lesion which involves the full thickness of the retina and tends to spill onto the inner retinal surface.
- The lesion is usually between a half to one disc diameter in size and most frequently occurs in the macula (**Fig. 12.219**) but may occur elsewhere (**Fig. 12.220**).

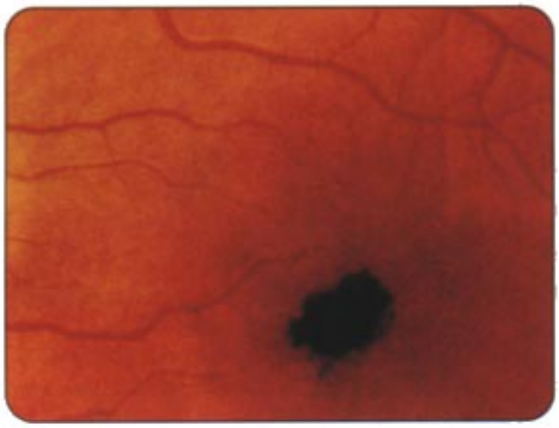

Fig. 12.219

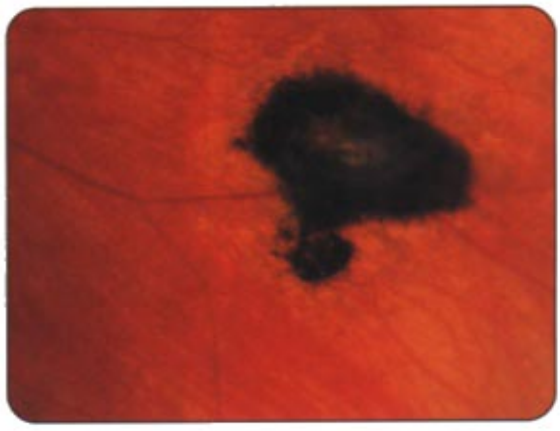

Fig. 12.220

Black 'sunburst' spots in nonproliferative sickle cell retinopathy

Signs

- Asymptomatic peripheral hyperpigmented lesions (**Fig. 12.221**) which are the result of resorbed subretinal hemorrhages.

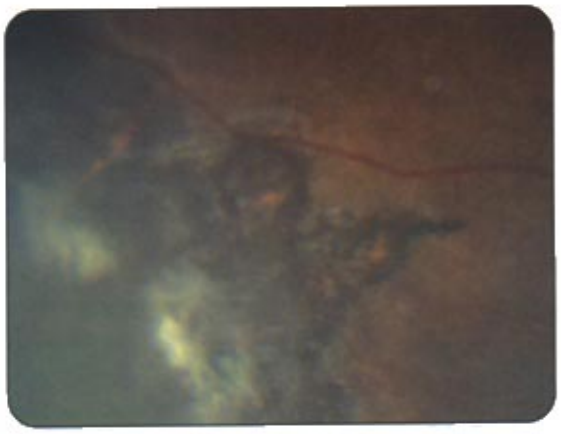

Fig. 12.221

Pale lesions

Old retinochoroiditis

Signs

- Round or oval atrophic lesion with visible choroidal vessels and variable surrounding pigmentation (**Fig. 12.222**).

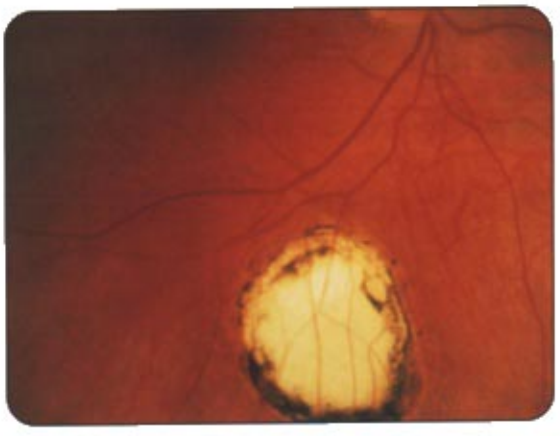

Fig. 12.222

Idiopathic chorioretinal atrophy

Signs

- Similar to old choroiditis but usually with less surrounding pigmentation (**Fig. 12.223**).
- Multiple small lesions in the peripheral fundus are referred to as 'paving-stone' degeneration.

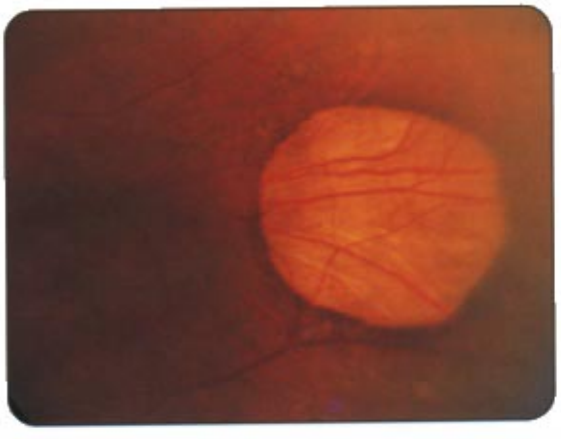

Fig. 12.223

Coloboma of retina and choroid

Signs

- Circular or oval area located inferior to the disc (**Fig. 12.224**).
- It may be associated with colobomas of the lens, ciliary body, and optic disc.

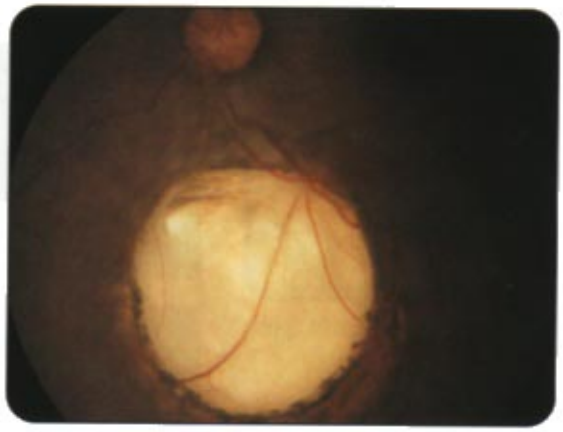

Fig. 12.224

Congenital albinotic spots

Signs

- Small pale circular lesions without pigmented outlines which usually occur in the peripheral fundus (**Fig. 12.225**) and rarely at the macula (**Fig. 12.226a and b**).
- Similar grouped spots are referred to as 'polar-bear tracks'.

Fig. 12.225

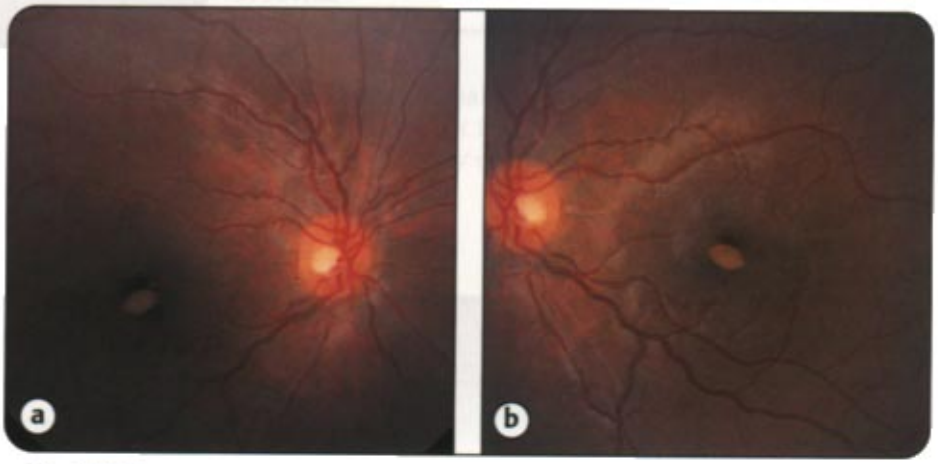

Fig. 12.226

Choroidal metastatic tumor

The most common primary sites for choroidal metastatic tumors are the bronchus in males and the breast in females.

Signs

- Cream–white, placoid, oval lesions, most frequently located at the posterior pole (**Fig. 12.227**).
- In some cases the tumor is more elevated (**Fig. 12.228**) and may resemble an amelanotic melanoma.
- Secondary exudative retinal detachment is common.
- Multiple deposits in the same eye, as well as bilateral involvement, are common.

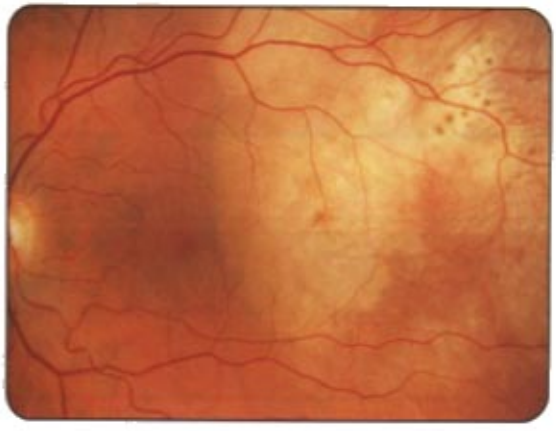

Fig. 12.227

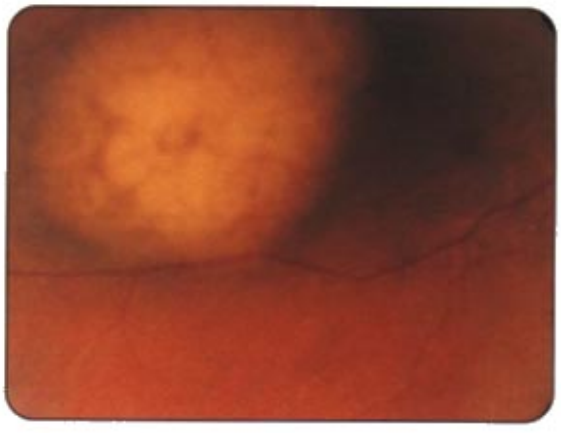

Fig. 12.228

Retinal astrocytoma

Retinal astrocytoma is an uncommon, benign tumor which is present in 50% of patients with tuberous sclerosis.

Signs

- White, mulberry-like lesion (**Figs 12.229, 12.230**).
- Multiple lesions in the same eye are common.
- Both eyes are involved in 15% of cases.

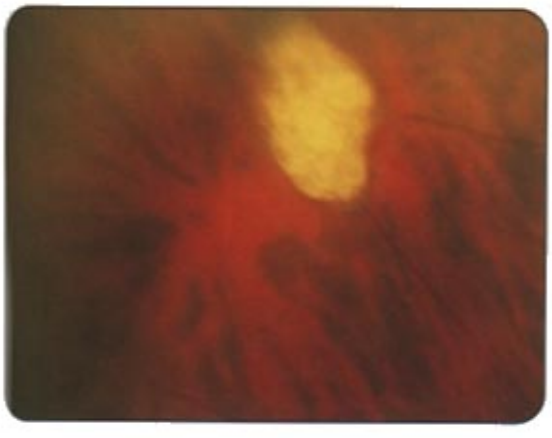

Fig. 12.229

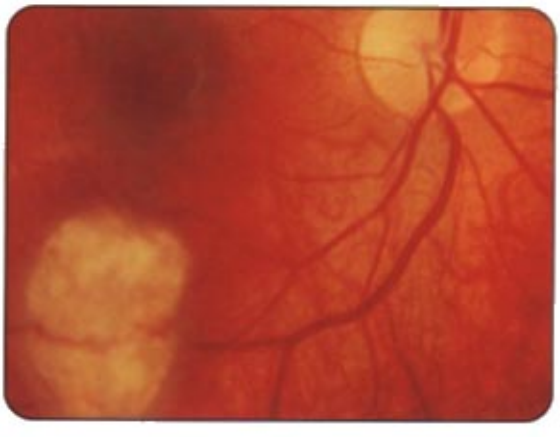

Fig. 12.230

Early retinoblastoma

Signs

- Slightly elevated pale nodule (**Fig. 12.231**).

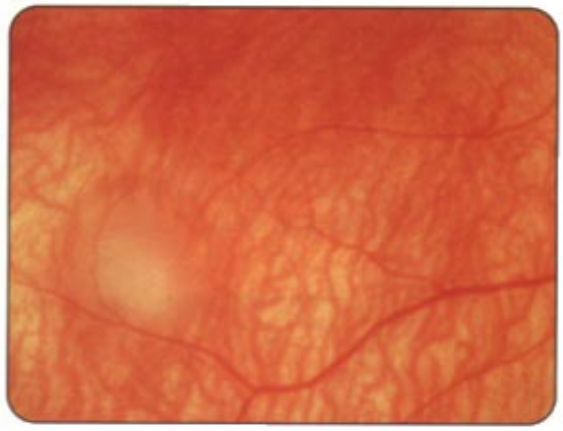

Fig. 12.231

MULTIPLE YELLOW–WHITE FLECKS

Nonhereditary

Hard drusen

Hard drusen are very common age-related changes which are not usually associated with the subsequent development of macular degeneration.

Signs

- Multiple, small, round, discrete, well-defined lesions at the level of the RPE (**Fig. 12.232**) which are usually symmetric (**Fig. 12.233a and b**).
- When they involve the peripheral retina, the lesions are associated with mild pigmentary changes (**Fig. 12.234a and b**).

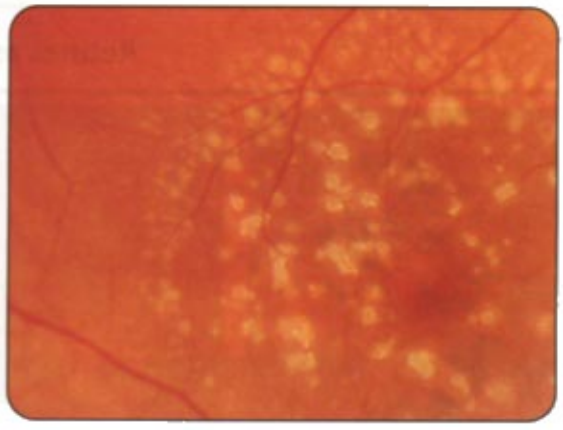

Fig. 12.232

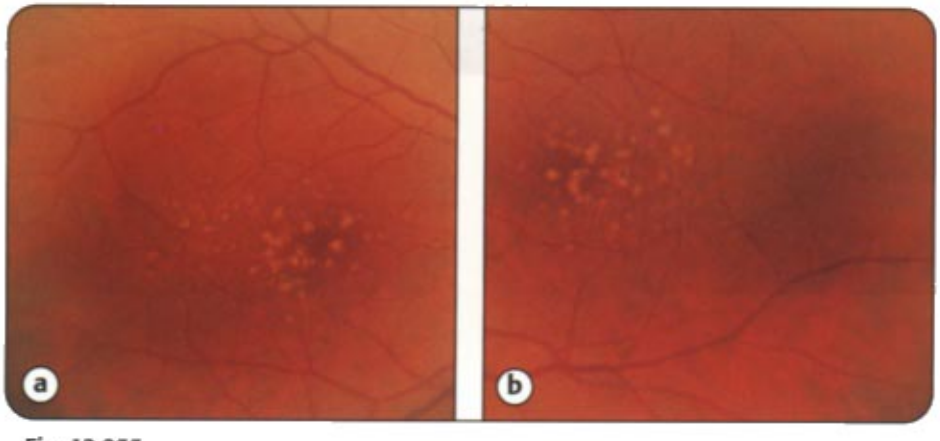

Fig. 12.233

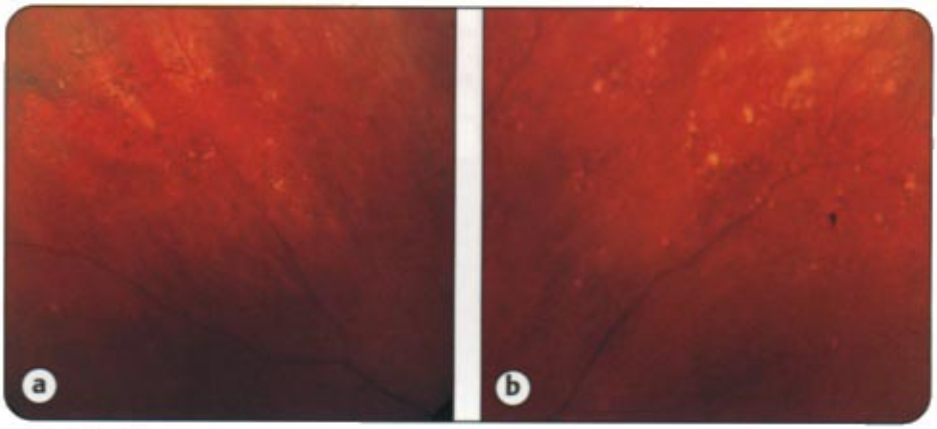

Fig. 12.234

Soft drusen

Soft drusen are common age-related changes which are associated with an increased risk of age-related macular degeneration.

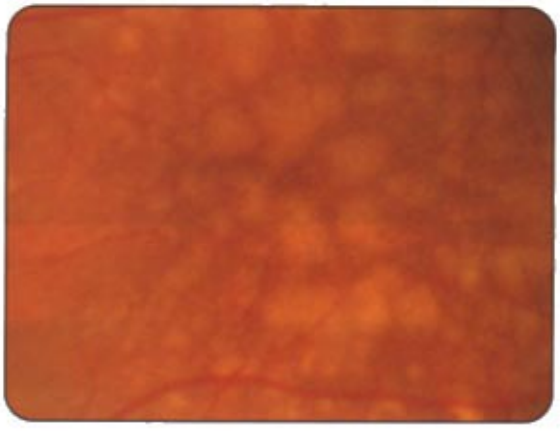

Fig. 12.235

Signs

- The lesions are larger than hard drusen and have poorly delineated soft margins (**Figs 12.235, 12.236a and b**).
- With time the lesions become confluent.
- In some cases the lesions are asymmetrically distributed in the two eyes (**Fig. 12.237a and b**).
- Secondary changes in the RPE are frequently seen in association with soft drusen (**Fig. 12.238a and b**).
- Fluorescein angiography shows hyperfluorescence (**Fig. 12.239b–d**).

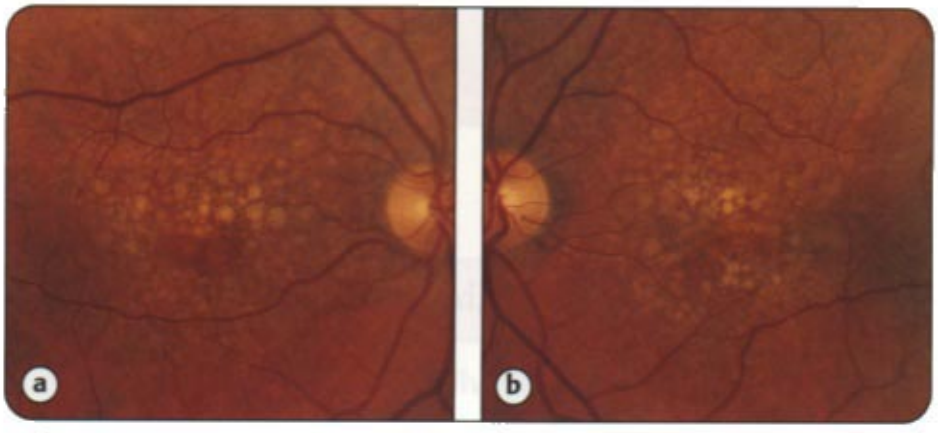

Fig. 12.236

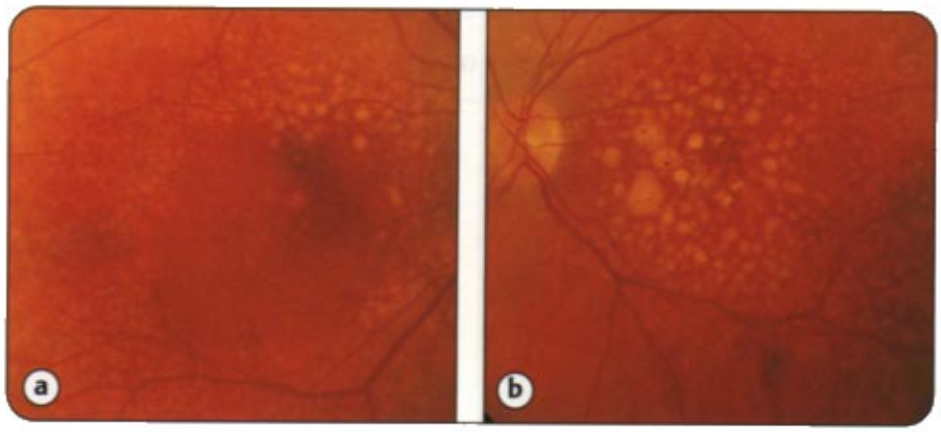

Fig. 12.237

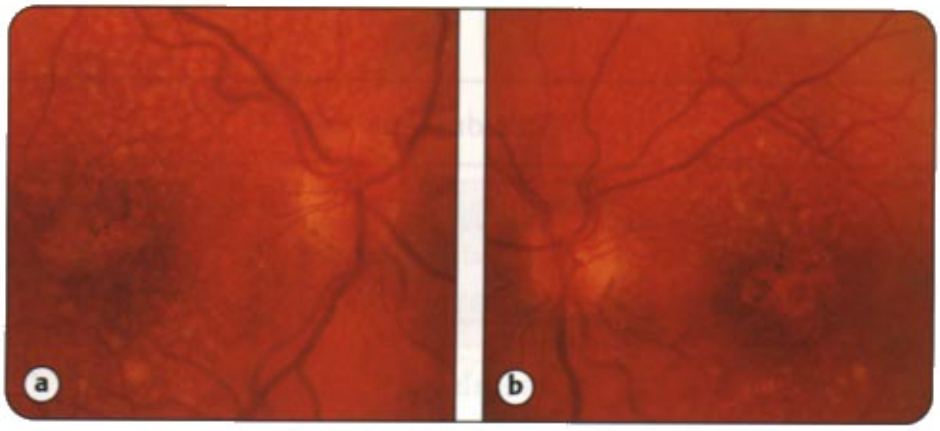

Fig. 12.238

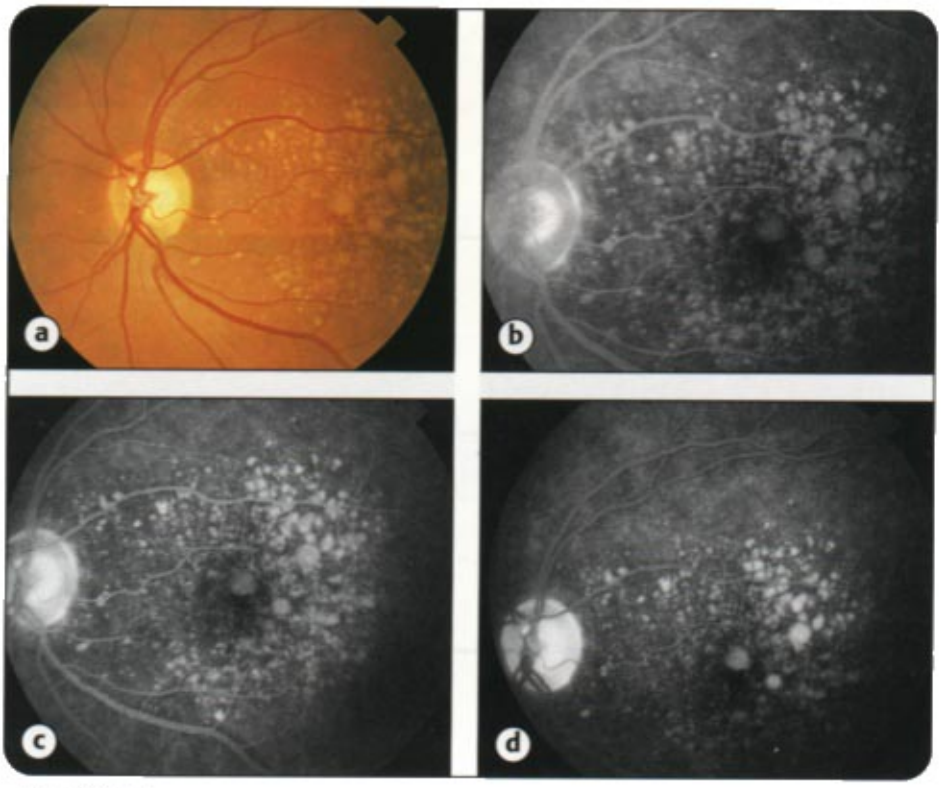

Fig. 12.239

Cuticular (basal laminar) drusen

Cuticular drusen are uncommon changes which occur in middle-aged individuals.

Signs

- Innumerable, small, uniformly sized, discrete, round, slightly raised, yellow subretinal lesions (**Fig. 12.240a**).
- On fluorescein angiography they give rise to a so-called starry-night or Milky-Way appearance (**Fig. 12.240b**).
- The lesions may be associated with pseudovitelliform macular degeneration.

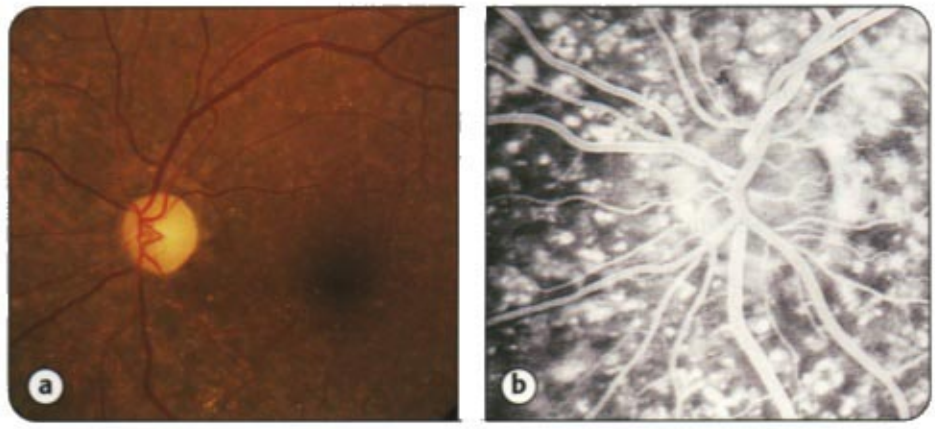

Fig. 12.240

Type II mesoangiocapillary membranoproliferative glomerulonephritis

Type II mesoangiocapillary membranoproliferative glomerulonephritis is a rare disease characterized by hematuria, proteinuria, and renal failure.

Signs

Bilateral, diffuse symmetric, yellow, drusen-like lesions at the posterior pole (**Fig. 12.241a and b**).

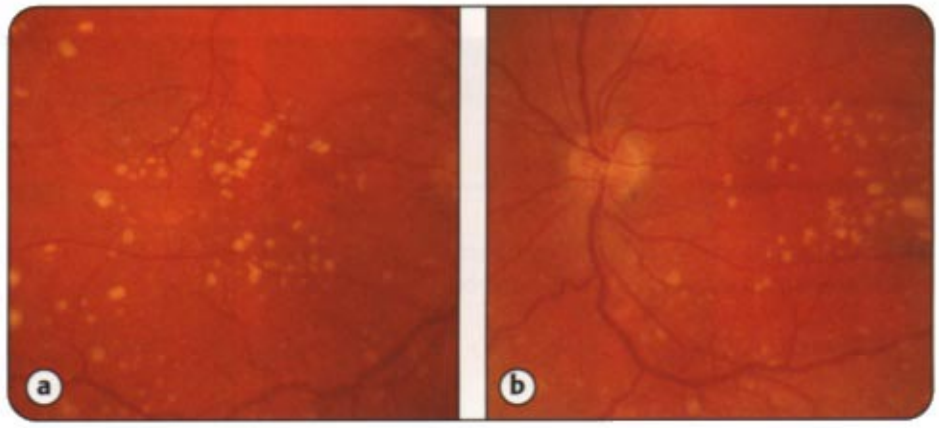

Fig. 12.241

Hereditary

Familial dominant drusen in young individuals (Doyne honeycomb dystrophy)

Doyne honeycomb dystrophy is an uncommon condition which appears during the late teens or early twenties. The long-term prognosis is good and only a small minority of patients subsequently develop macular degeneration.

Signs

- Large, discrete, nodular drusen, often in a very symmetric distribution such as temporal to the fovea (**Fig. 12.242a and b**) or nasal to the disc.

- With time, the drusen merge together and eventually become confluent, giving rise to a honeycomb appearance (**Fig. 12.243**).
- In very longstanding cases, pigmentary changes develop (**Fig. 12.244**) and visual acuity becomes compromised.

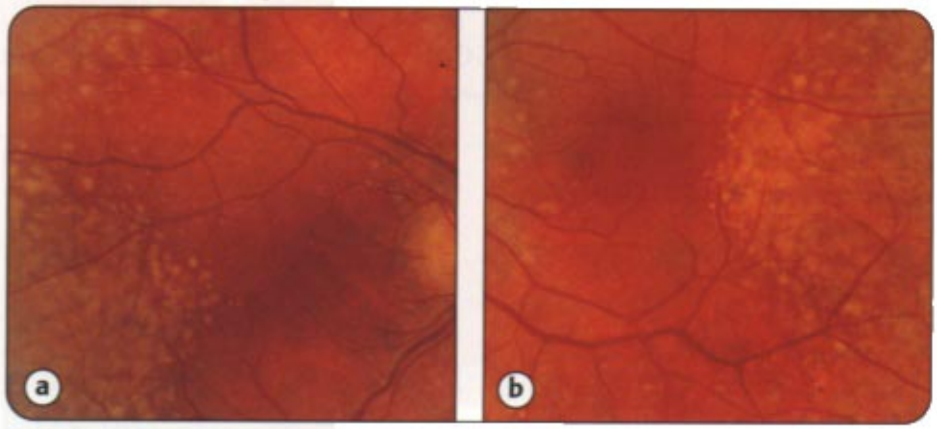

Fig. 12.242

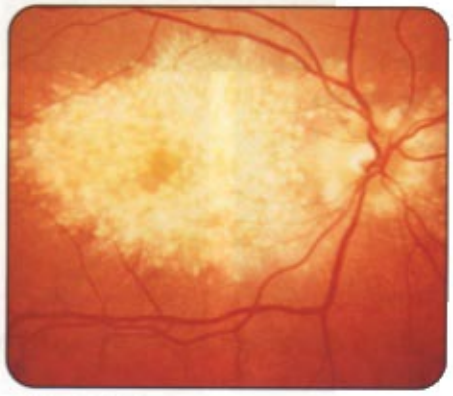

Fig. 12.243

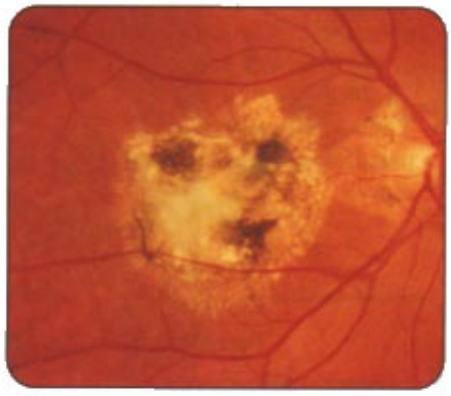

Fig. 12.244

Malattia levantinese

Malattia levantinese is a very rare, dominantly inherited condition which presents during the second decade of life.

Signs

- Bilateral, symmetric, diffuse, yellowish, round spots of variable size in the perifoveal region.
- Later these spots tend to merge, forming a white circle with indistinct edges (**Fig. 12.245**).

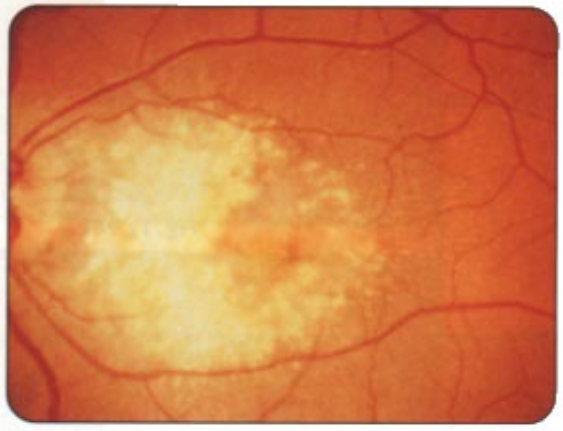

Fig. 12.245

Fundus flavimaculatus

Fundus flavimaculatas is a rare, recessively inherited condition.

Signs

- Yellow–white, fish-tail-shaped flecks at the posterior pole and midperiphery of both eyes (**Figs 12.246, 12.247a and b**).
- The flecks may occur in isolation or in association with maculopathy (Stargardt macular dystrophy – *see* **Fig. 12.161a and b**).
- Fluorescein angiography in longstanding cases shows hyperfluorescence due to atrophic changes in the RPE (**Fig. 12.247c and d**). A dark choroid characterized by absence of background choroidal fluorescence is seen in 85% of cases.

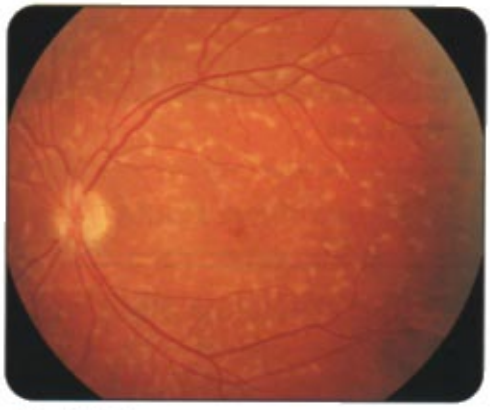

Fig. 12.246

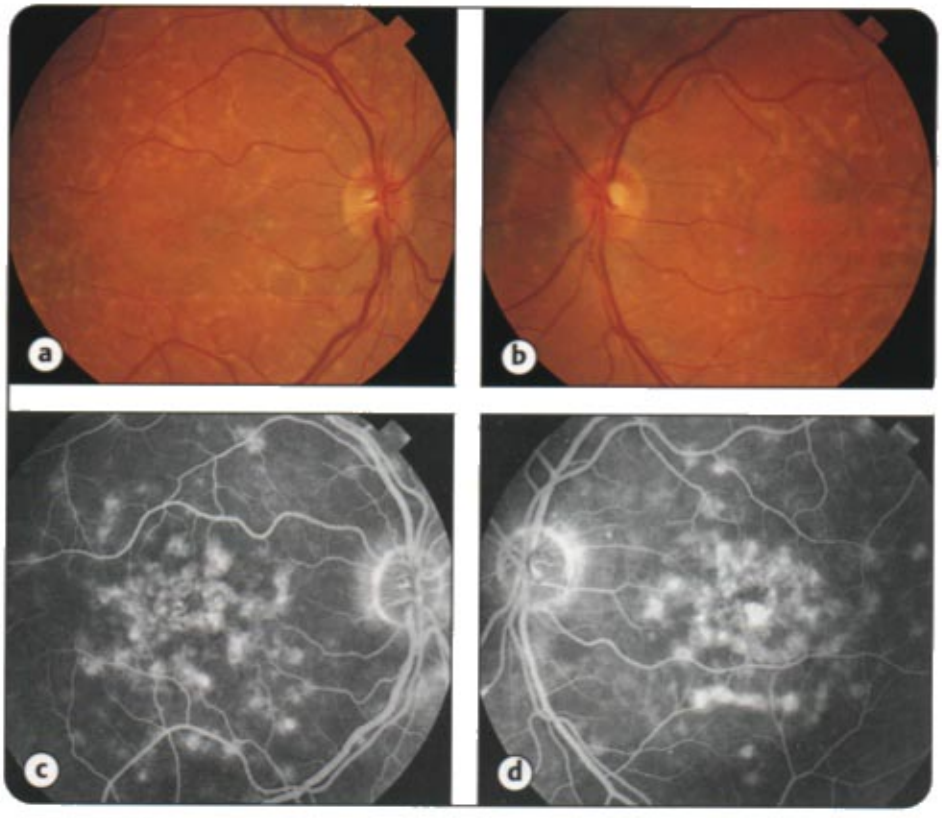

Fig. 12.247

Fundus albipunctatus

Fundus albipunctatus is a rare, recessively inherited condition characterized by congenital nonprogressive night-blindness.

Signs

- A multitude of tiny yellow–white spots extending from the posterior pole (where they are most numerous) to the periphery (**Fig. 12.248a and b**).
- The macula is spared and visual acuity is unaffected, although there is congenital stationary night-blindness.

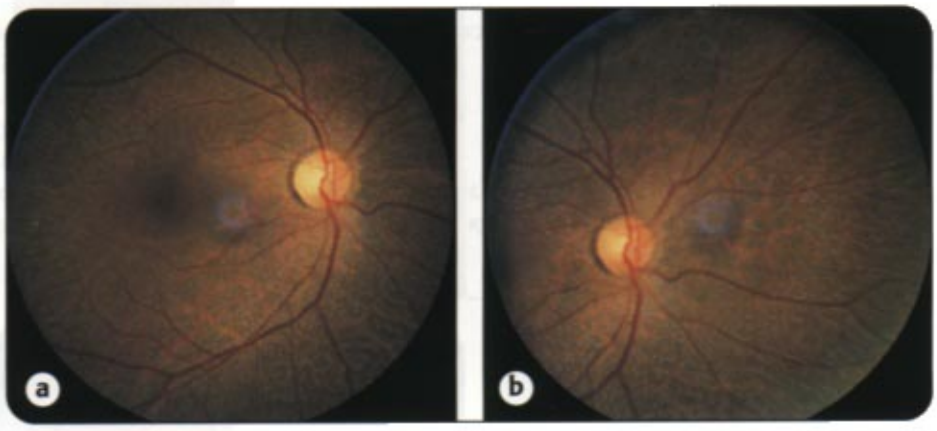

Fig. 12.248

Retinitis punctata albescens

Retinitis punctata albescens is a subtype of retinitis pigmentosa and is characterized by progressive night-blindness.

Signs

- Multiple discrete white dots around the retinal mid-periphery, similar to fundus albipunctatus, associated with arteriolar attenuation (**Fig. 12.249**).

Systemic associations

- Bassen–Kornzweig syndrome.

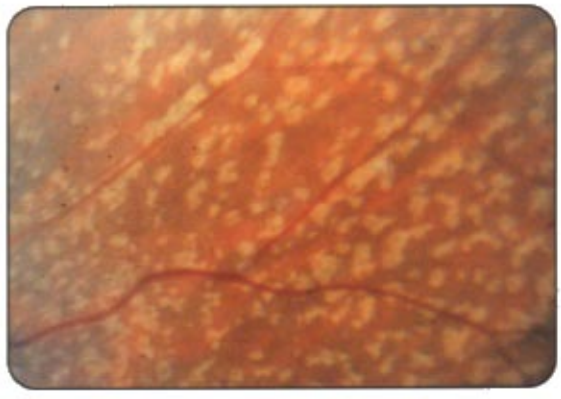

Fig. 12.249

Pattern dystrophy

Pattern dystrophy is an uncommon, dominantly inherited condition which is slowly progressive but has a relatively good long-term prognosis.

Signs

- Bilateral, symmetric lesions with various geometric shapes (**Fig. 12.250**) such as that of a butterfly.

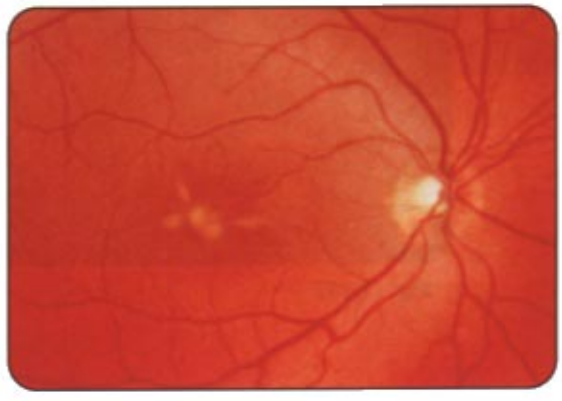

Fig. 12.250

Benign flecked retina syndrome

Benign flecked retina syndrome is very rare and has an excellent long-term prognosis.

Signs

- Flecks scattered throughout the fundus, similar to fundus flavimaculatus but without macular involvement (**Fig. 12.251a and b**).

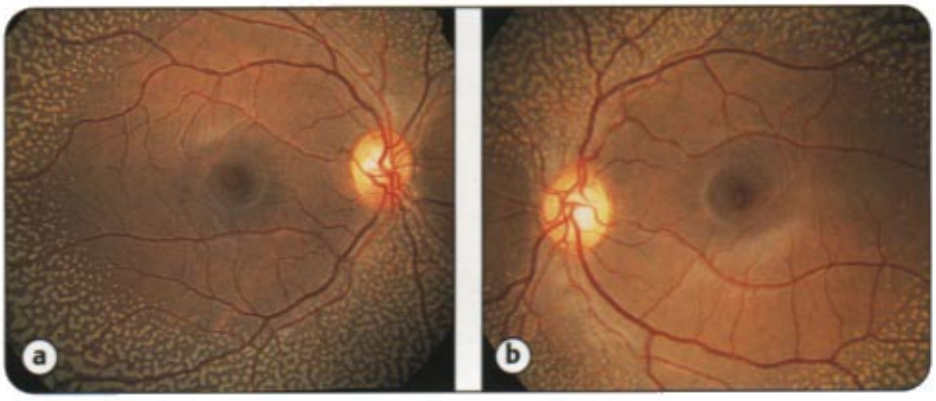

Fig. 12.251

Alport syndrome

Alport syndrome is a rare, X-linked disorder characterized by progressive renal disease and deafness.

Signs

- Pale punctate yellow flecks at the posterior pole (**Fig. 12.252**).
- Confluent and clustered yellow flecks in the midperiphery (**Fig. 12.253**).

Look for

- Anterior lenticonus.
- Microspherophakia.

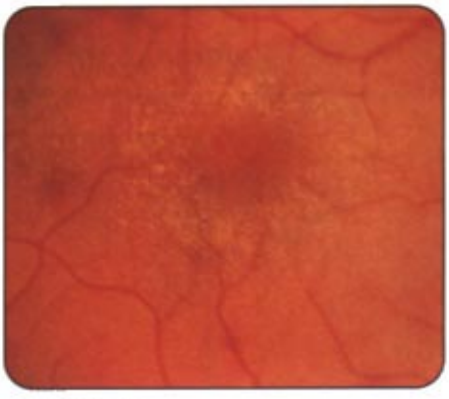

Fig. 12.252

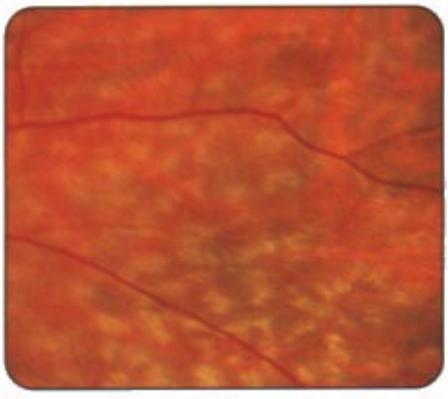

Fig. 12.253

Early North Carolina macular dystrophy

North Carolina macular dystrophy is a very rare, dominantly inherited condition.

Signs

- Yellow–white drusen-like deposits in the periphery (**Fig. 12.254**) and at the macula (**Fig. 12.255**) which develop during childhood in eyes with normal visual acuity.

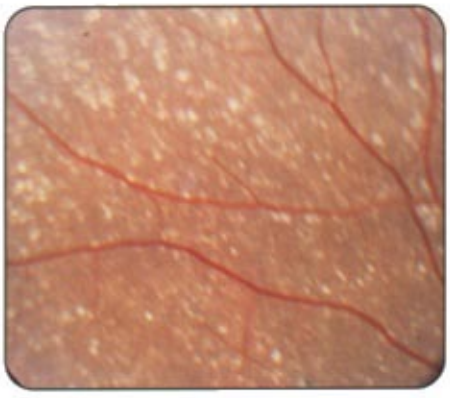

Fig. 12.254

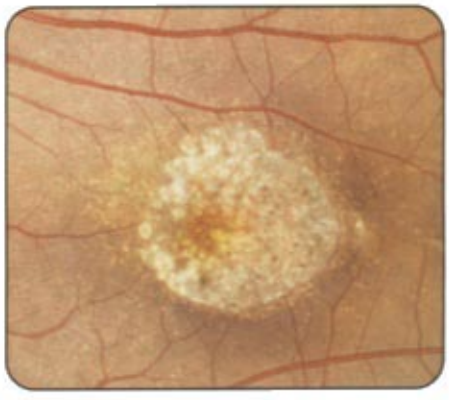

Fig. 12.255

DIFFUSE PIGMENT CLUMPING

Nonhereditary

Age-related honeycomb (reticular) degeneration

Signs

- Bilateral, fine network of midperipheral perivascular pigmentation (**Fig. 12.256**).

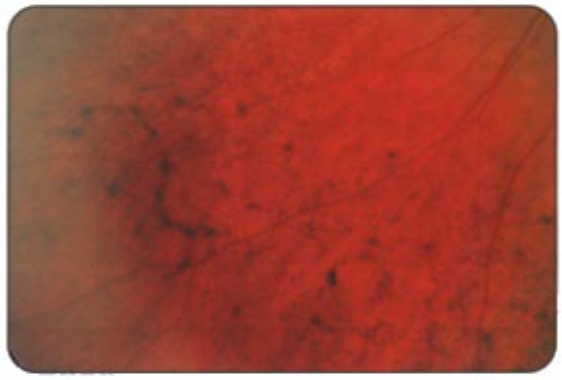

Fig. 12.256

Advanced chloroquine toxicity

Advanced retinotoxicity is very rare and only occurs when large doses of chloroquine have been administered for a long time.

Signs

- Pigment clumping which may be perivascular (**Fig. 12.257**).

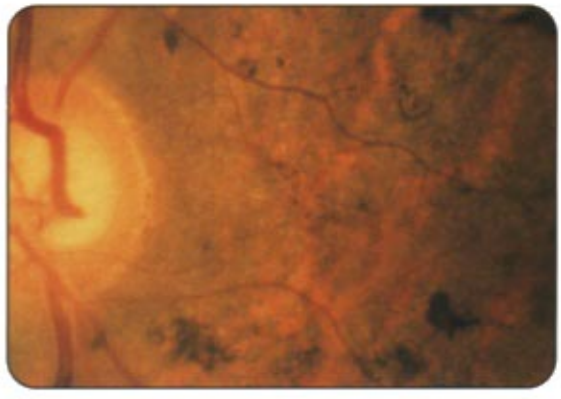

Fig. 12.257

Related to retinal detachment

- Following resolution of exudative retinal detachment (leopard spots) (**Fig. 12.258**).
- Following collapse of retinoschisis or old retinal detchment which is usually inferior (**Fig. 12.259**).

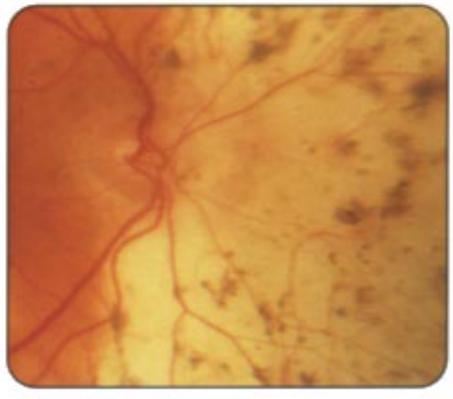

Fig. 12.258

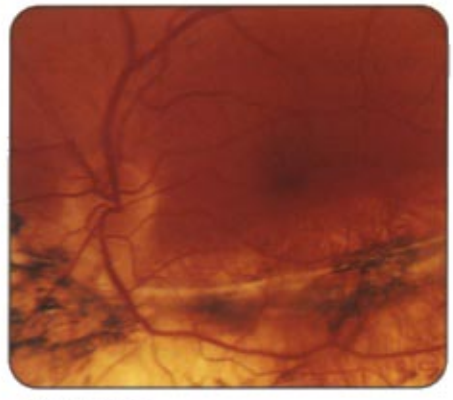

Fig. 12.259

Grouped congenital hypertrophy of the RPE

Signs

- Small, circumscribed, oval black spots, often organized in a pattern simulating animal footprints (**Fig. 12.260**).

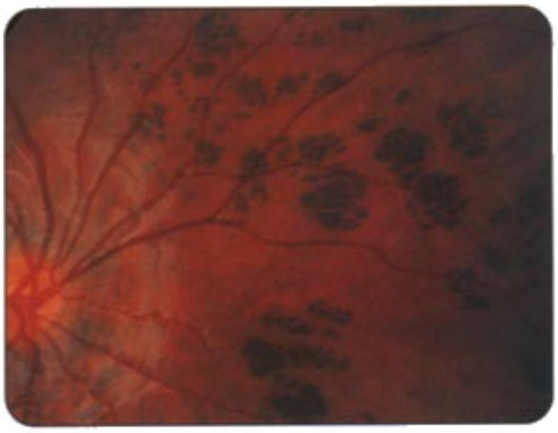

Fig. 12.260

Rubella retinopathy

Retinopathy is a common manifestation of congenital rubella in which visual acuity is usually normal.

Signs

- Generalized mottled pigmentary changes, most marked at the posterior pole (**Fig. 12.261**).
- Subretinal neovascularization develops in a few cases.

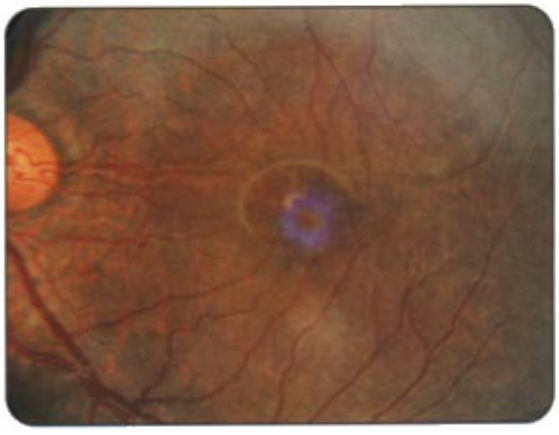

Fig. 12.261

Congenital syphilis

Signs

- Salt and pepper fundus (**Fig. 12.262**) with normal vision in mild cases.
- Pigmentary retinopathy resembling retinitis pigmentosa in severe cases.

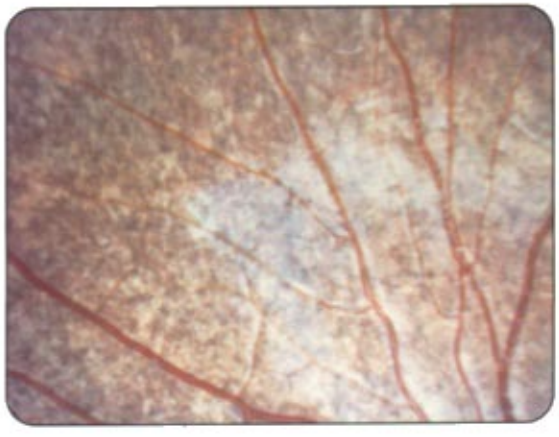

Fig. 12.262

Hereditary

Typical retinitis pigmentosa

Typical retinitis pigmentosa is an uncommon, bilateral, progressive retinal degeneration which usually presents during the second decade of life and leads to severe visual impairment in later life. Inheritance may be dominant, recessive, or X-linked.

Signs

- Bone-spicule perivascular pigmentation (**Fig. 12.263a and b**).
- Arteriolar attenuation which is well demonstrated on fluorescein angiography (**Fig. 12.263c and d**).
- Waxy pallor of optic disc.
- Unmasking of large choroidal vessels (**Fig. 12.264a and b**) is a late feature.
- Maculopathy – atrophic (**Fig. 12.265**), cellophane, or cystoid macular edema.

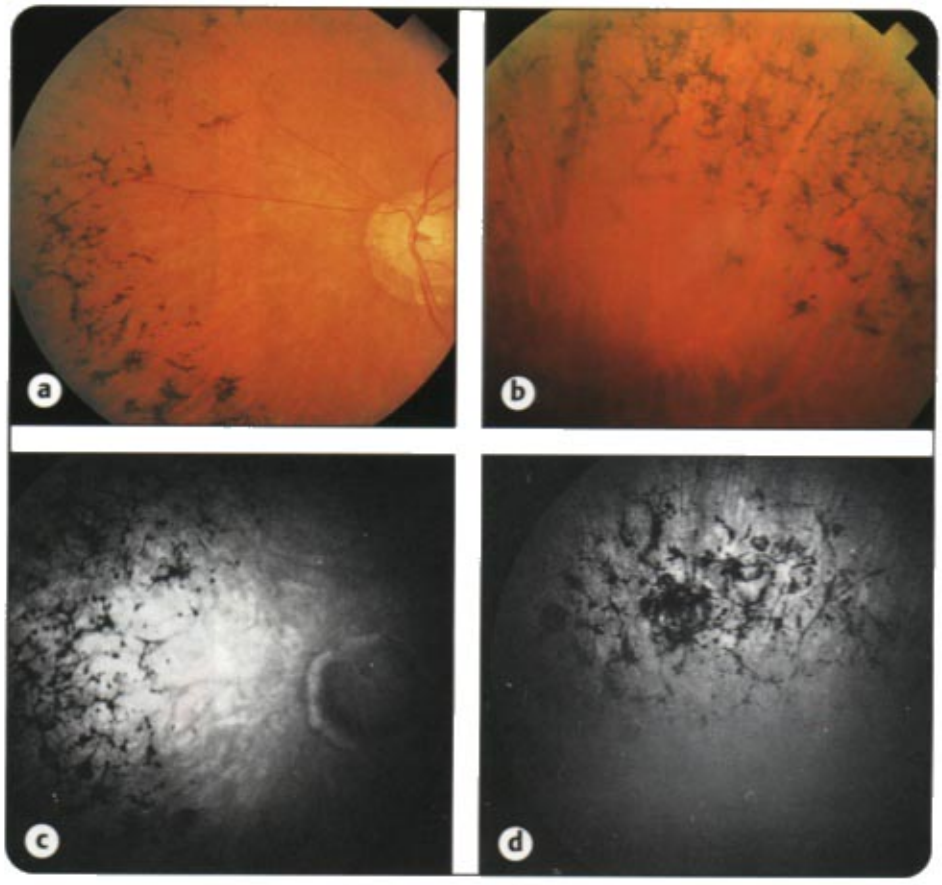

Fig. 12.263

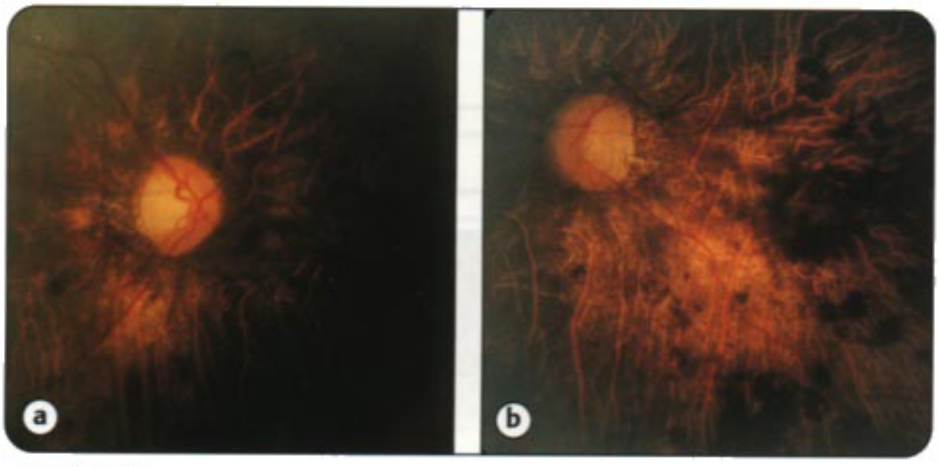

Fig. 12.264

Fig. 12.265

Atypical retinitis pigmentosa

Signs

- Sectorial retinitis pigmentosa – only one quadrant (**Fig. 12.266**) or one half (**Fig. 12.267**) is involved.
- Pericentric retinitis pigmentosa – changes confined to the posterior pole (**Fig. 12.268**).
- Unilateral.
- Paravenous.
- With relatively mild, nonspecific pigmentary changes (**Fig. 12.269**) or no pigmentary changes (**Fig. 12.270**).

Systemic associations of pigmentary retinopathy

- Refsum disease.
- Bassen–Kornzweig syndrome.
- Usher syndrome (types I and II).
- Laurence–Moon syndrome.
- Bardet–Bield syndrome.
- Kearns–Sayre syndrome.
- Cockayne syndrome.
- Alström syndrome.
- Olivopontocerebellar atrophy.
- Batten–Vogt disease.
- Hallervorden–Spatz syndrome.
- Mucopolysaccharidosis (types I and II).
- Mucolipidosis type IV.
- Jeune syndrome.
- Sly syndrome.

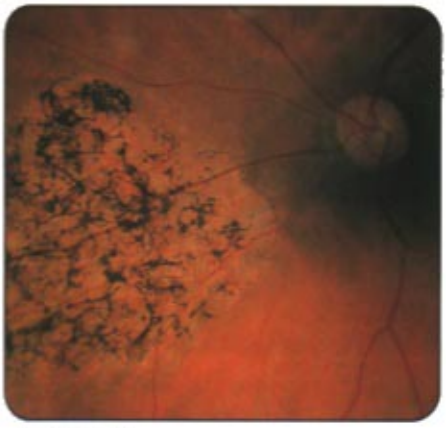

Fig. 12.266

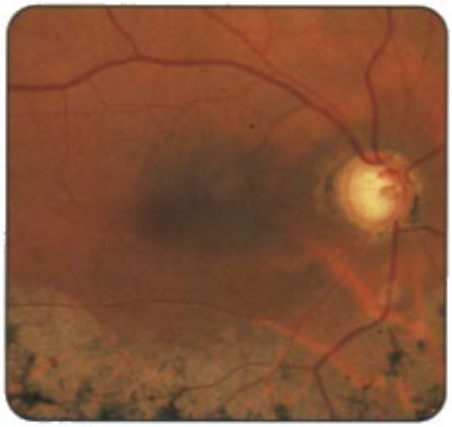

Fig. 12.267

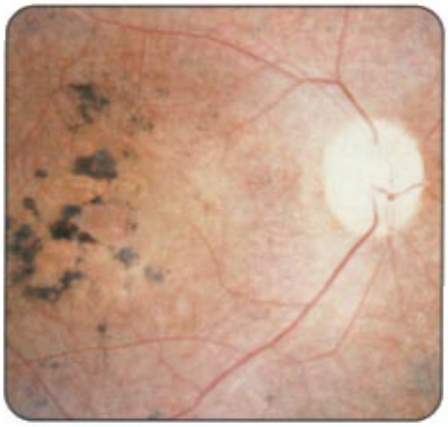

Fig. 12.268

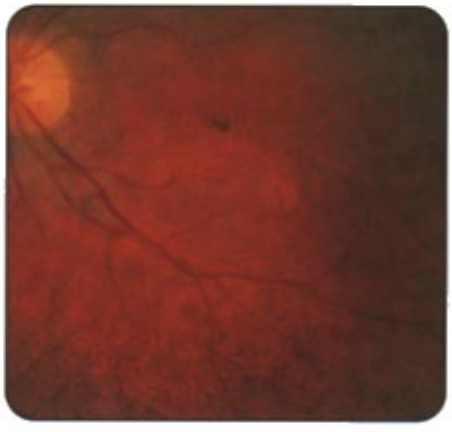

Fig. 12.269

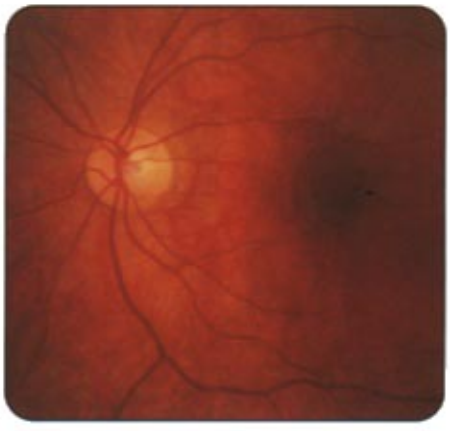

Fig. 12.270

Female carriers of choroideremia

Signs

- Peripheral pigmentary granularity or clumping (**Fig. 12.271**).

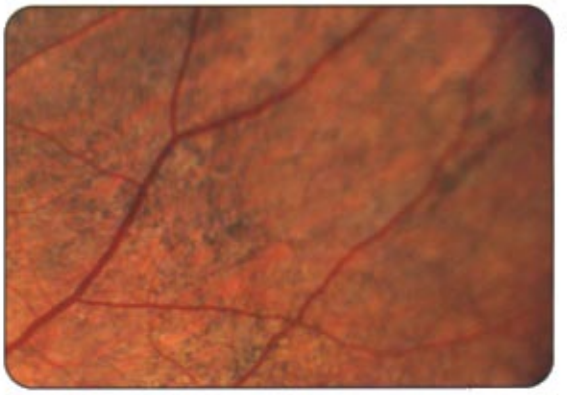

Fig. 12.271

In eyes with angioid streaks

Signs

- Peau-d'orange pigmentary mottling (**Fig. 12.272**) and reticular-like peripheral pigmentary clumping.

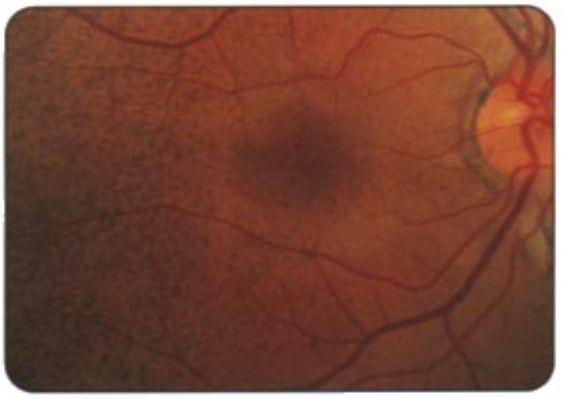

Fig. 12.272

GENERALIZED WHITE FUNDUS

Nonhereditary

Extensive retinal nerve fiber myelination

Signs

- Extensive white lesions following the distribution of the retinal nerve fiber layers (**Fig. 12.273**).

Fig. 12.273

High myopia

Signs

- Diffuse chorioretinal atrophy with visualization of large choroidal vessels (**Fig. 12.274**).

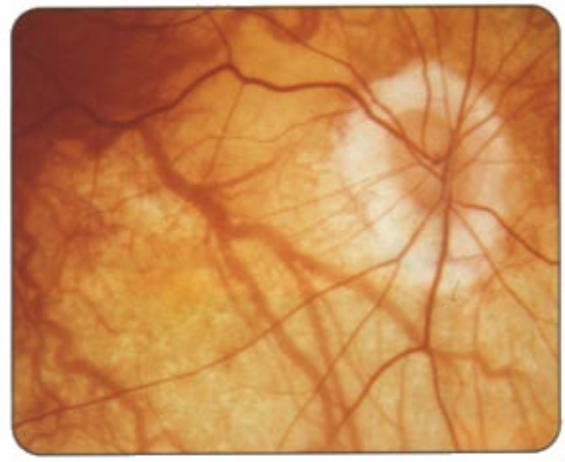

Fig. 12.274

Acute retinal ischemia

Signs

- Branch retinal artery occlusion – pallor is confined to the area supplied by the occluded vessel (**Fig. 12.275a**) and fluorescein angiography shows initial nonfilling (**Fig. 12.275b–d**) of the retinal circulation.
- Central retinal artery occlusion – pallor at the posterior pole with a cherry-red spot at the macula (**Fig. 12.276a**). The angiogram (**Fig. 12.276b–d**) shows extensive nonfilling of the retinal circulation.
- Ophthalmic artery occlusion is characterized by choroidal and retinal ischemia and nonperfusion (**Fig. 12.277a–d**).

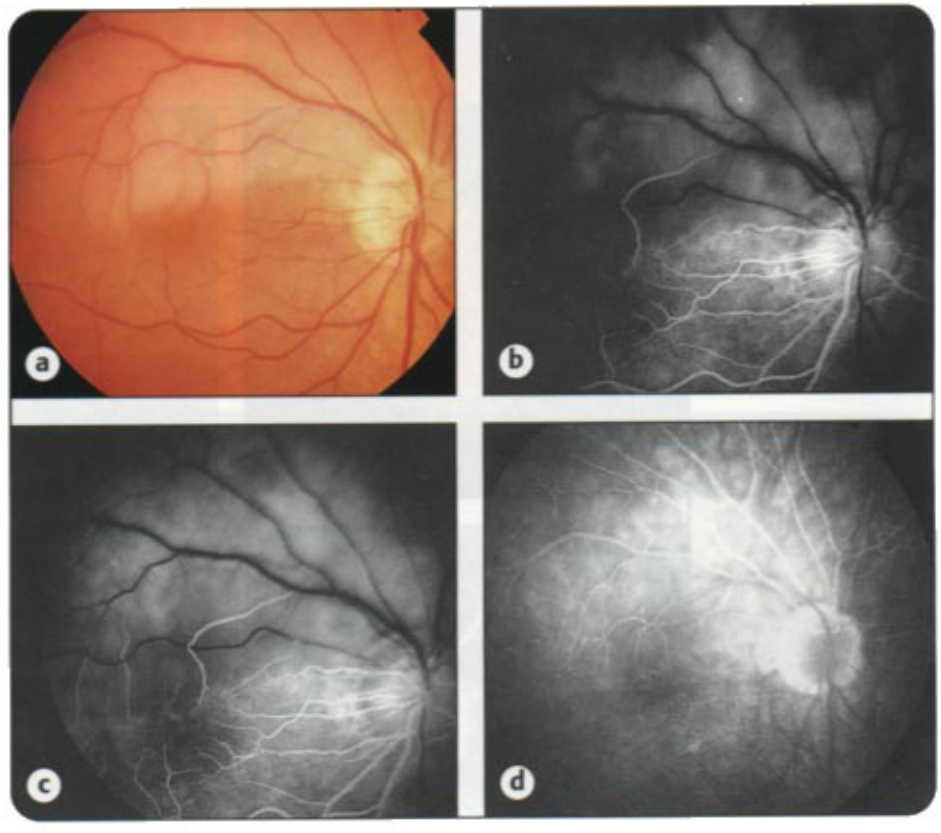

Fig. 12.275

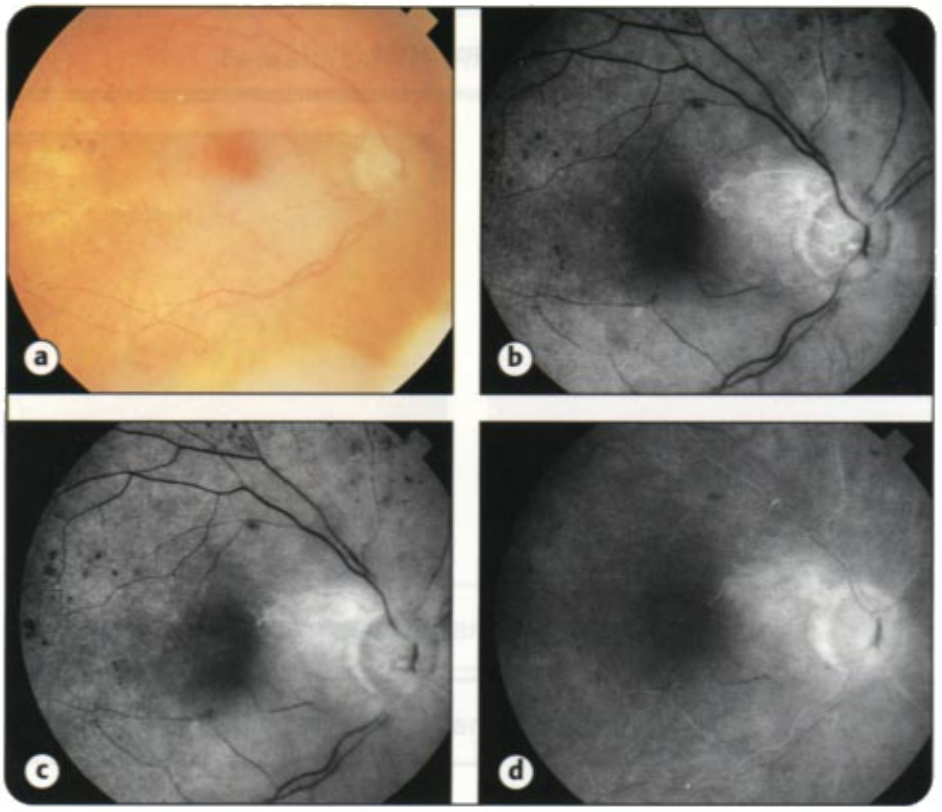

Fig. 12.276

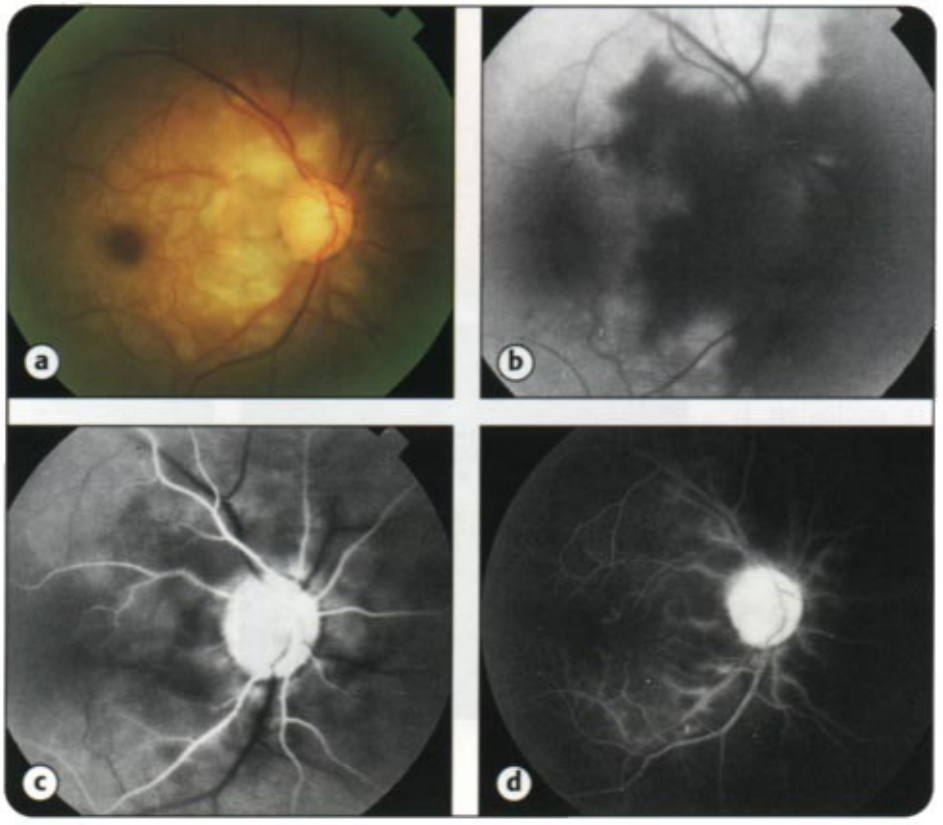

Fig. 12.277

Commotio retinae

Signs

- Localized pallor (**Fig. 12.278**) caused by blunt ocular trauma.

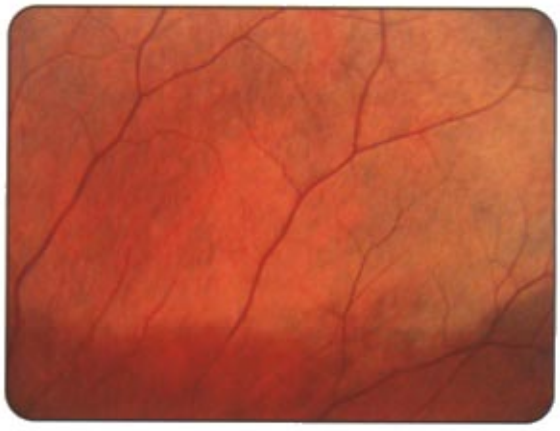

Fig. 12.278

Large coloboma of retina and choroid

Signs

- Large, oval, sharply defined white area below (**Fig. 12.279**) and sometimes including the disc.

Look for

- Associated ocular and systemic malformations.

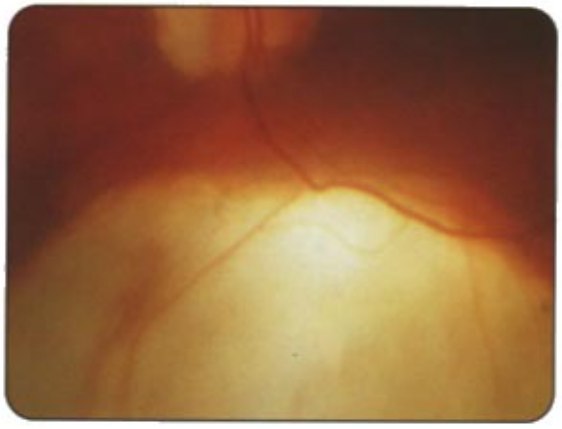

Fig. 12.279

Hereditary

Albinism

Signs

- Generalized fundus pallor with prominent choroidal vasculature (**Fig. 12.280**).

Look for

- Iris transillumination (**Fig. 12.281**).

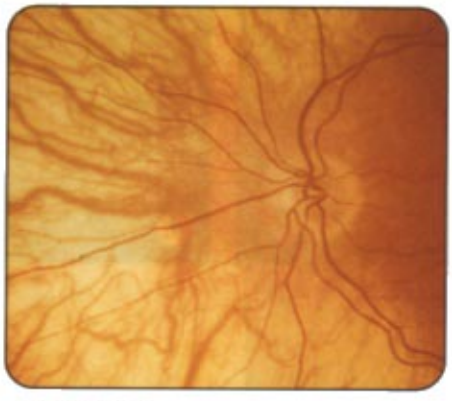

Fig. 12.280

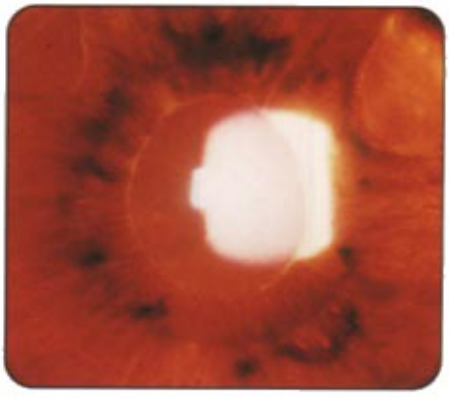

Fig. 12.281

Choroideremia

Choroideremia is a very rare condition which is inherited in an X-linked recessive manner. It presents during the first decade of life and results in blindness by the seventh.

Signs

- Late stages show extensive RPE and choroidal atrophy with sparing of the fovea (**Fig. 12.282a and b**). The angiogram (**Fig. 12.282c–f**) shows normal retinal vessels and large choroidal vessels. The center of the macula shows hypofluorescence surrounded by hyperfluorescence.

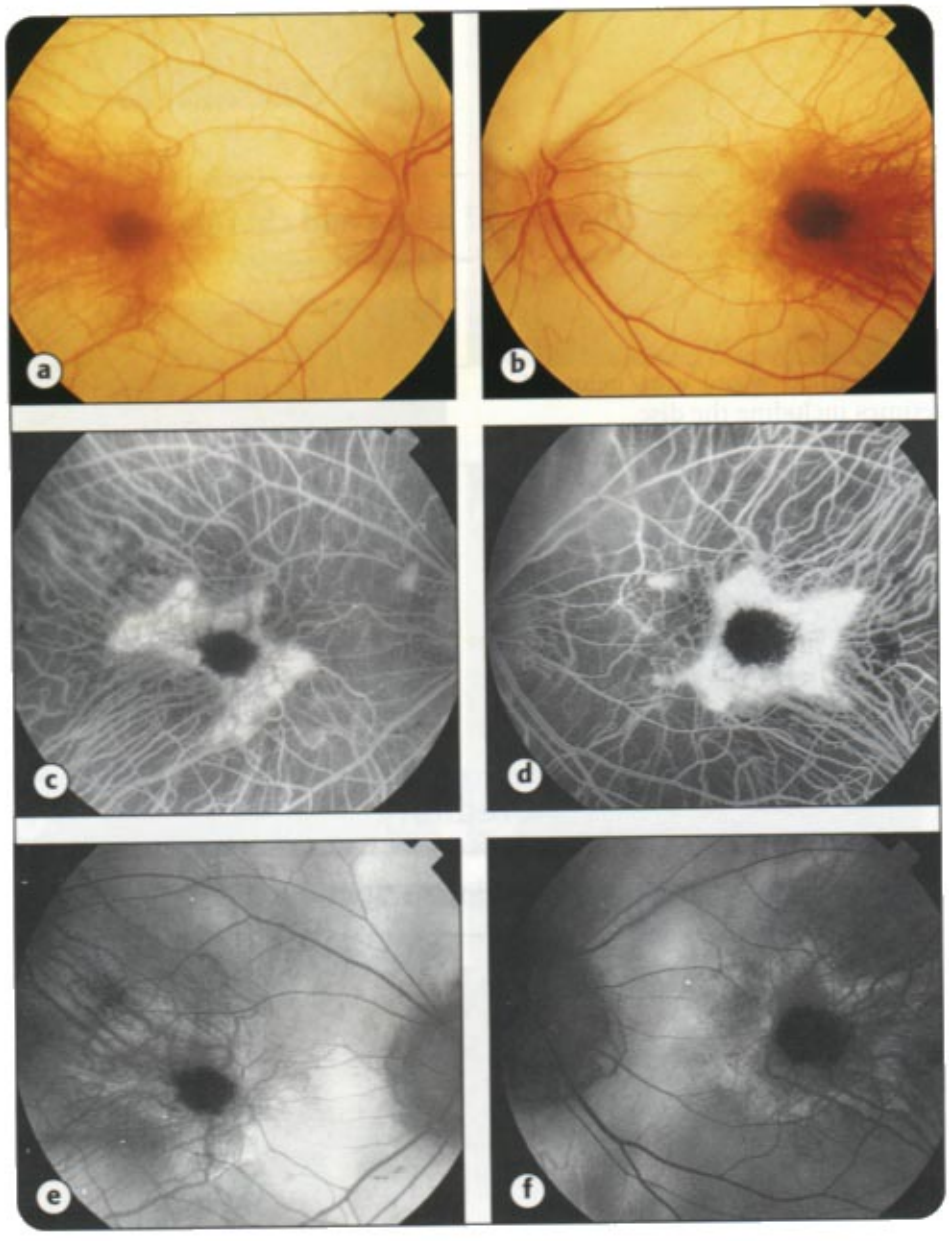

Fig. 12.282

Gyrate atrophy

Gyrate atrophy is a very rare, recessively inherited condition which usually presents during the first decade of life and causes severe visual loss by the fourth or fifth decades. Some patients have associated hyperornithemia.

Signs

- Peripheral scalloped areas of chorioretinal atrophy (**Fig. 12.283a and b**) associated with vitreous degeneration.

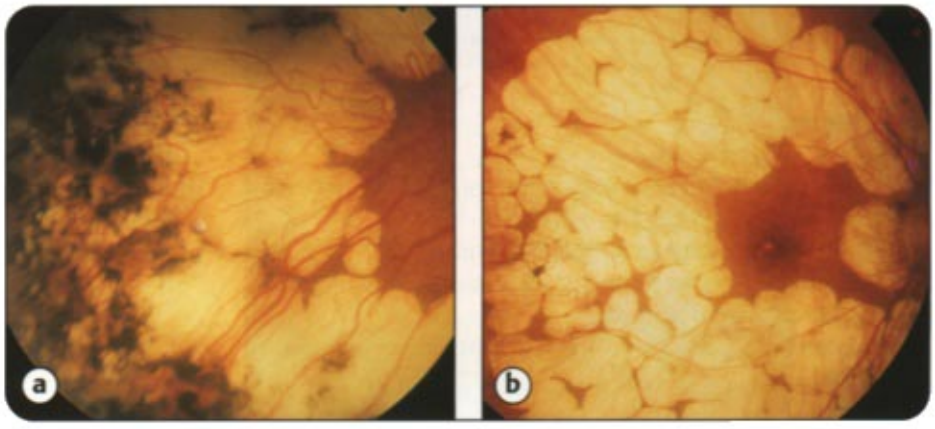

Fig. 12.283

Diffuse choroidal atrophy

Diffuse choroidal atrophy is a very rare, dominantly inherited condition which presents between the first and fourth decades of life.

Signs

- Diffuse atrophy of the RPE and choriocapillaris with prominence of the larger choroidal vessels (**Fig. 12.284a and b**). The retinal blood vessels are normal or slightly constricted.

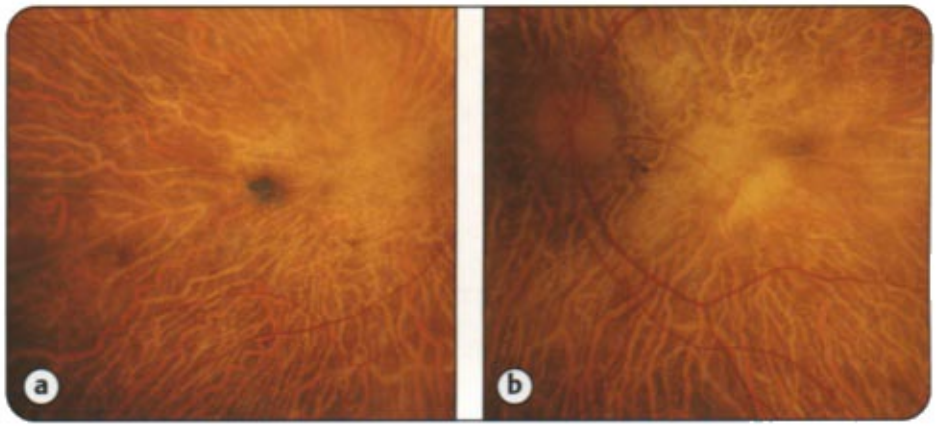

Fig. 12.284

Progressive bifocal chorioretinal atrophy

Progressive bifocal chorioretinal atrophy is a very rare, dominantly inherited condition of early onset.

Signs

- Large atrophic macular and nasal retinal lesions (**Fig. 12.285**).

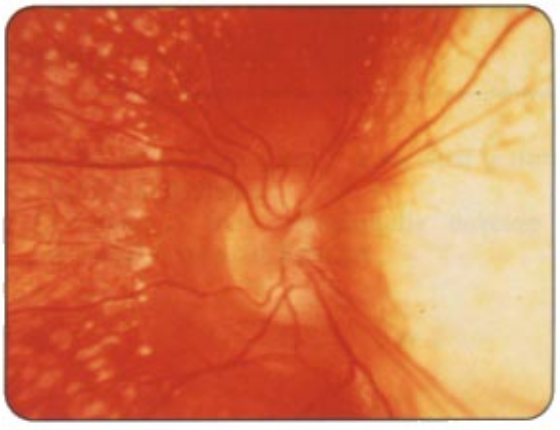

Fig. 12.285

FOLDS AND LINEAR STREAKS

Chorioretinal folds

Signs

- Roughly parallel striae, which are most frequently seen at the posterior pole (**Fig. 12.286a**).
- The folds may vary in length and width.
- The folds are most frequently horizontal but they may also be vertical or oblique.
- Occasionally they are associated with circumpapillary (Paton) lines.
- Fluorescein angiography (**Fig. 12.286b–d**) shows hyperfluorescent and hypofluorescent streaks in which the former correspond to the crests and the latter to the troughs.

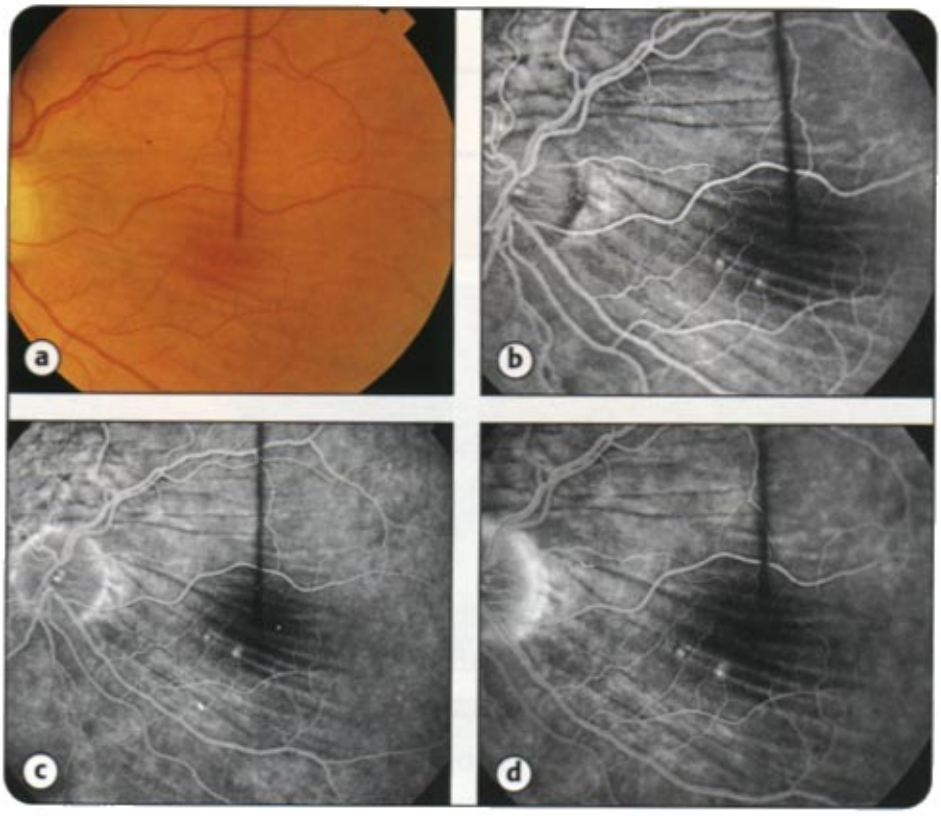

Fig. 12.286

Table 12.9 Causes of chorioretinal folds

Ocular
• hypotony
• disc edema
• choroidal tumors
• hypermetropia (idiopathic)
• scleral buckling
Orbital
• thyroid ophthalmopathy
• pseudotumor
• neoplasm

Lacquer cracks

Lacquer cracks are the result of breaks in Bruch membrane and occur in about 4% of highly myopic eyes.

Signs

- Fine, yellow branching lines at the posterior pole in highly myopic eyes (**Fig. 12.287**).
- Secondary choroidal neovascularization may develop in some cases.

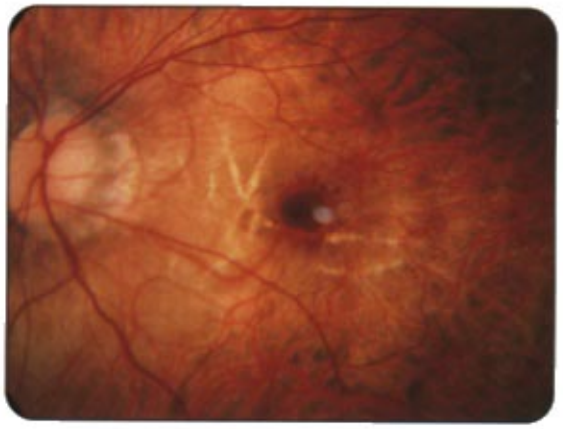

Fig. 12.287

Pigment demarcation lines in retinal detachment

These lines are caused by an attempt to wall-off an inferior retinal detachment.

Signs

- Subretinal demarcation line at the junction of attached and detached retina.
- Initially the line is pigmented, but with time it tends to lose its pigment (**Fig. 12.288**).
- The line is convex with respect to the ora serrata and in some cases several lines may be present (**Fig. 12.289**).

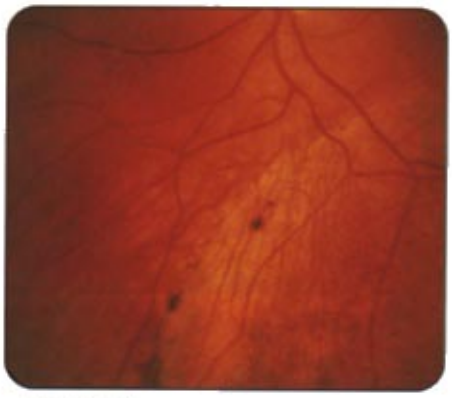

Fig. 12.288

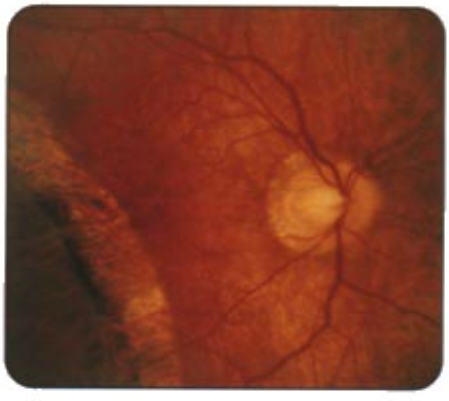

Fig. 12.289

Choroidal rupture

Choroidal rupture is caused by blunt ocular trauma.

Signs

- Crescentic vertical lines, most frequently located temporally and passing through the macula (**Fig. 12.290**).
- In the acute stage there is associated hemorrhage (**Fig. 12.291**) and edema.
- Old lesions may occasionally develop secondary choroidal neovascularization.

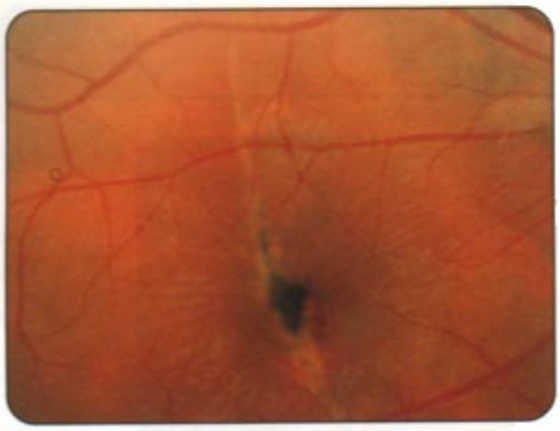

Fig. 12.290

Fig. 12.291

Angioid streaks

Angioid streaks are uncommon lesions which may be associated with a wide variety of systemic disorders.

Signs

- Gray or dark-red streaks with serrated edges, lying beneath normal blood vessels (**Fig. 12.292a and b**).
- The streaks intercommunicate around the disc and then radiate outward toward the periphery.

Associated features

- Peau-d'orange mottling at the posterior pole (**Fig. 12.293a and b**).
- Peripapillary chorioretinal atrophy.
- Optic disc drusen – occasionally.
- Focal peripheral chorioretinal scars (salmon spots).
- Reticular-like peripheral pigmentary clumping.

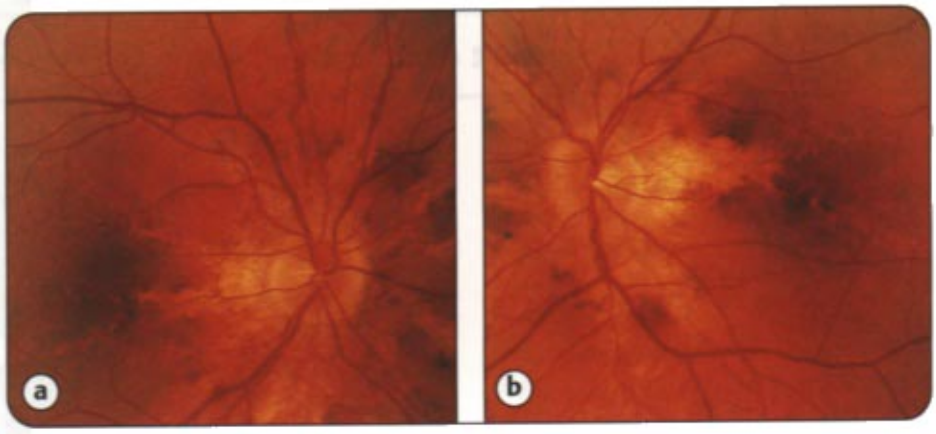

Fig. 12.292

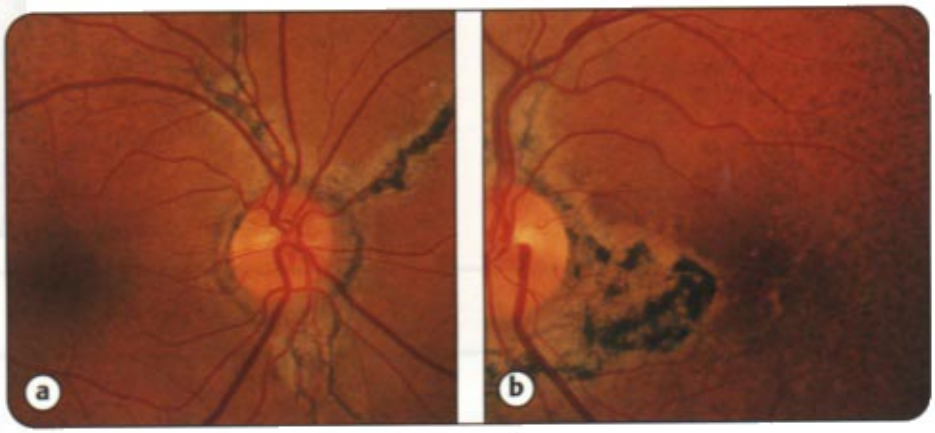

Fig. 12.293

Complications

- Choroidal neovascularization. In **Figure 12.294**, the right eye (**Fig. 12.294a and c**) shows three choroidal neovascular membranes; the left eye (**Fig. 12.294b and d**) has only angioid streaks.
- Choroidal rupture and hemorrhage caused by relatively trivial blunt ocular trauma.

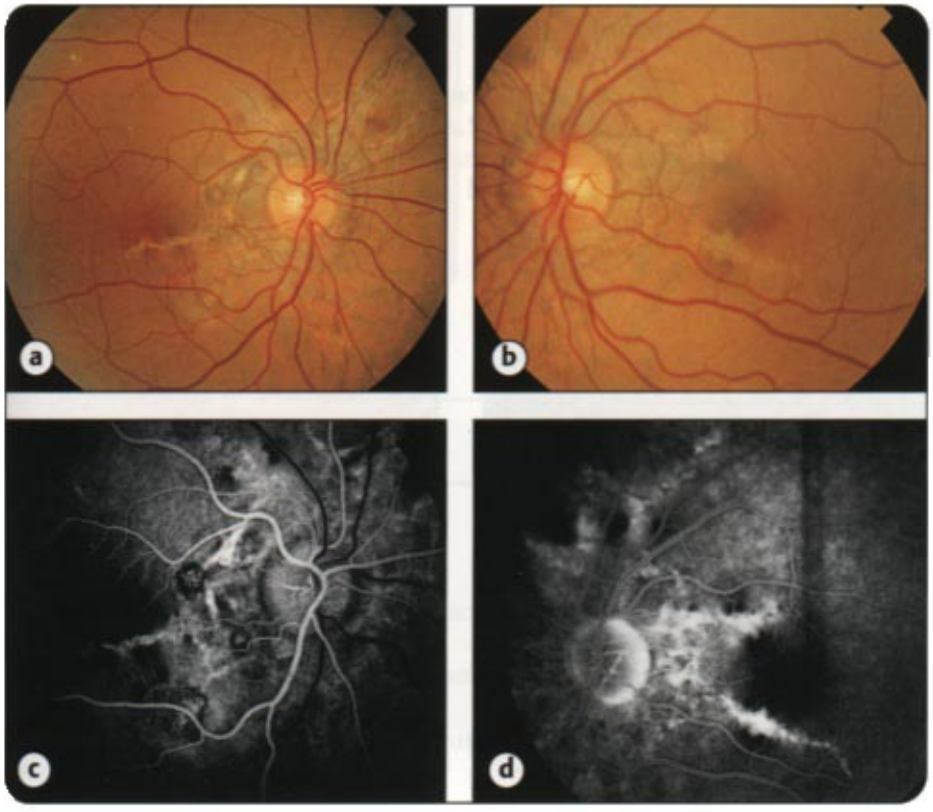

Fig. 12.294

Table 12.10 Systemic associations of angioid streaks

Skin
• pseudoxanthoma elasticum
• Ehlers–Danlos syndrome
• senile elastosis
Bone
• Paget disease
• juvenile Paget disease
• acromegaly
• calcinosis
Blood
• sickle cell anemia
• thalassemia
• abetalipoproteinemia
• hereditary spherocytosis

RETINAL DETACHMENT

Rhegmatogenous retinal detachment

The signs depend on the duration of the detachment and the presence or absence of proliferative vitreoretinopathy (PVR).

Signs of retinal detachment

- Convex configuration of detached retina which has a slightly opaque and corrugated appearance (**Fig. 12.295**).
- Blood vessels appear darker than in attached retina, hence the color contrast between veins and arteries is less apparent.
- Loss of underlying choroidal pattern.

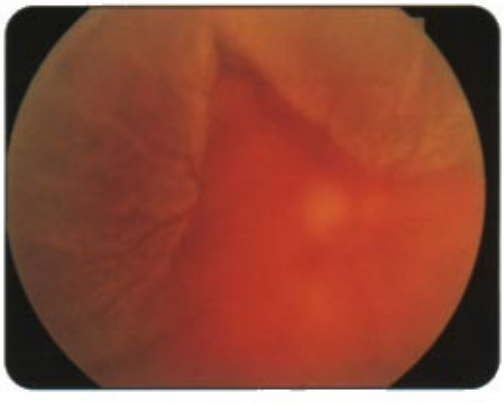

Fig. 12.295

- The area of detachment extends to the ora serrata except in the rare cases caused by a macular hole in which the detachment may remain confined to the posterior pole.
- The detached retina undulates freely with eye movements in the absence of significant PVR.
- An old retinal detachment may be very thin and associated with demarcation lines (**Fig. 12.296**) and secondary intraretinal cysts (**Fig. 12.297**).

Signs of PVR

- Grade A (minimal) – diffuse vitreous haze and pigment (tobacco-dust) cells.
- Grade B (moderate) – retinal stiffness and decreased mobility of vitreous gel.
- Grade C (marked) – rigid retinal folds which may be anterior or posterior. The severity is expressed by the number of clockhours involved by the proliferation, which may be epiretinal (**Figs. 12.298, 12.299**) or subretinal (**Fig. 12.300**) or both.

Retinal breaks

- Retinal breaks appear as discontinuities of the retinal surface (**Figs 12.301, 12.302**).
- A pseudohole may be seen at the macula if the posterior pole is detached (*see* **Fig. 12.193**).

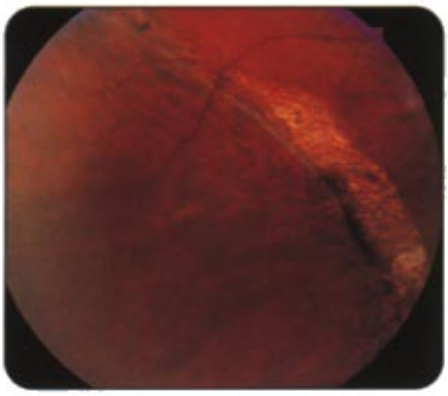

Fig. 12.296

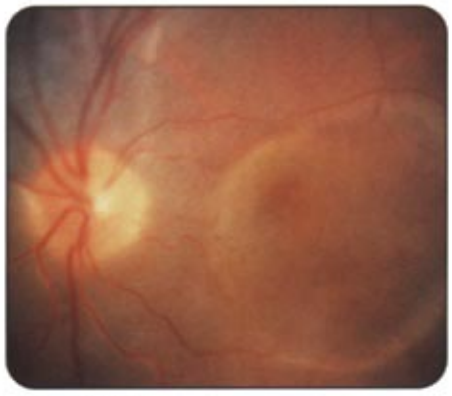

Fig. 12.297

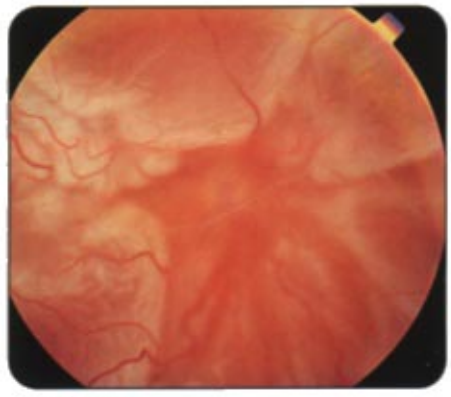

Fig. 12.298

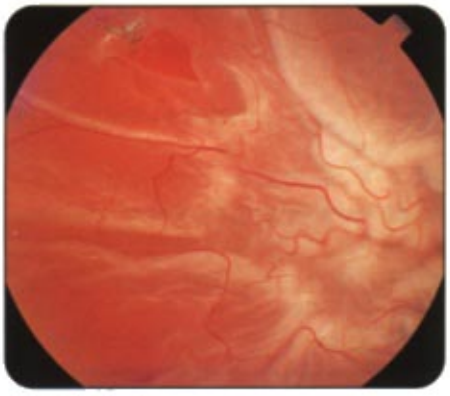

Fig. 12.299

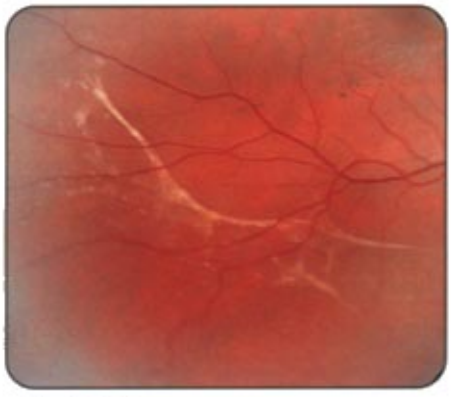

Fig. 12.300

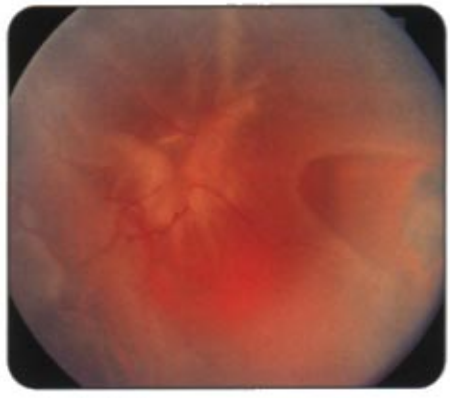

Fig. 12.301

Fig. 12.302

Predisposing vitreoretinal degenerations

- Lattice degeneration – peripheral, sharply demarcated, spindle-shaped areas with an arborizing network of white lines (**Fig. 12.303**) and variable pigmentation (**Fig. 12.304**).
- Snailtrack degeneration – peripheral, sharply demarcated, white, frost-like bands (**Fig. 12.305**).

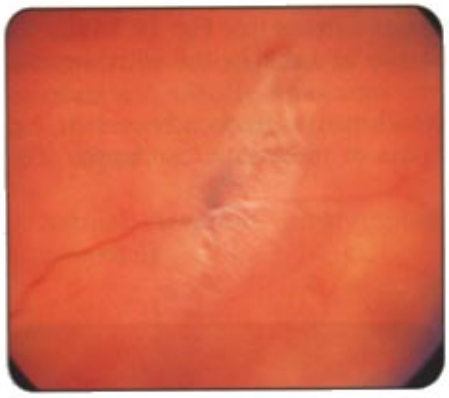

Fig. 12.303

Fig. 12.304

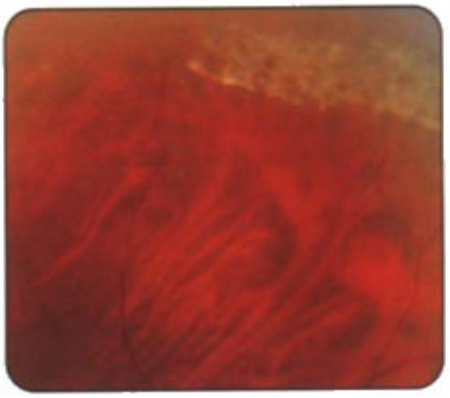

Fig. 12.305

Tractional retinal detachment

Signs

- Concave configuration of detached retina (**Figs 12.306–12.309**).
- The detached retina is shallow and seldom extends to the equator.
- The highest elevation occurs at sites of vitreoretinal traction.
- Retinal mobility is severely reduced.
- Absence of retinal breaks.

Causes

- Proliferative retinopathies (*see* **Figs 12.306, 12.307**).
- Penetrating ocular trauma (*see* **Figs 12.308, 12.309**).
- Severe intraocular inflammation.

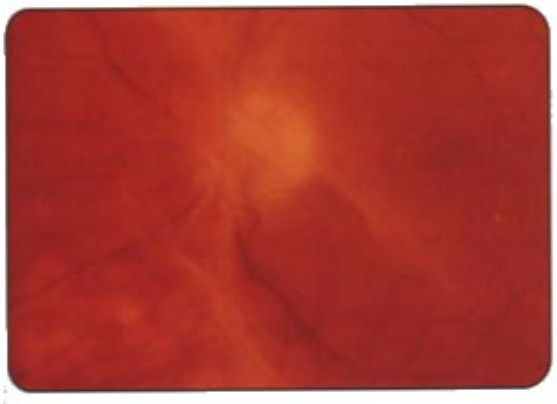

Fig. 12.306

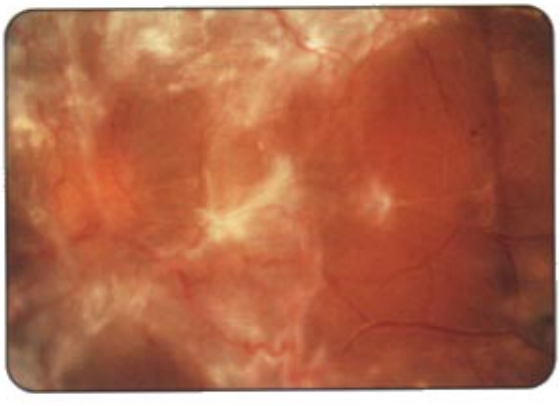

Fig. 12.307

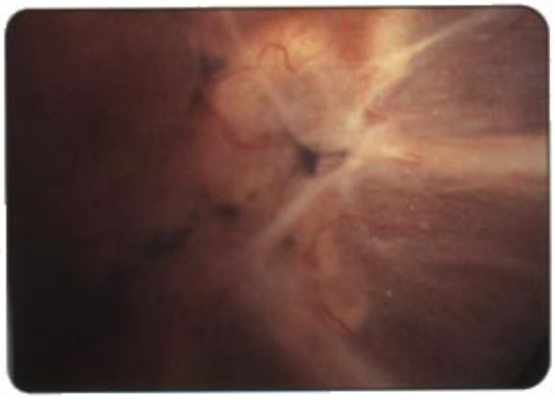

Fig. 12.308

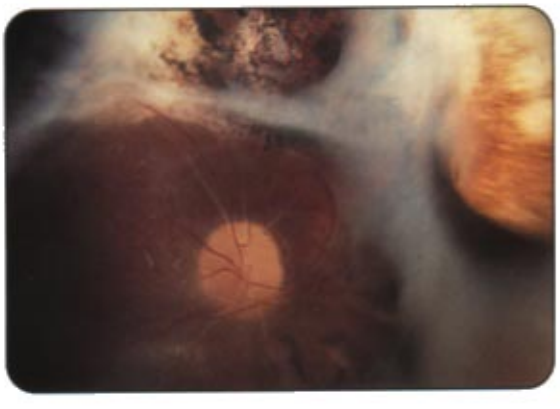

Fig. 12.309

Exudative retinal detachment

Signs

- Convex configuration of detached retina.
- The detached retina is deep (**Fig. 12.310**) and may even touch the back of the lens (**Fig. 12.311**).
- Retinal mobility is marked.
- The subretinal fluid responds to the forces of gravity, so detaching the area of retina under which it accumulates (shifting fluid).
- Scattered subretinal pigment clumps (leopard spots) may be seen when the retina flattens (*see* **Fig. 12.258**).
- The subretinal fluid may be turbid in eyes with posterior scleritis.
- The cause of the detachment is frequently evident, e.g. a choroidal melanoma or metastatic carcinoma (**Fig. 12.312**).

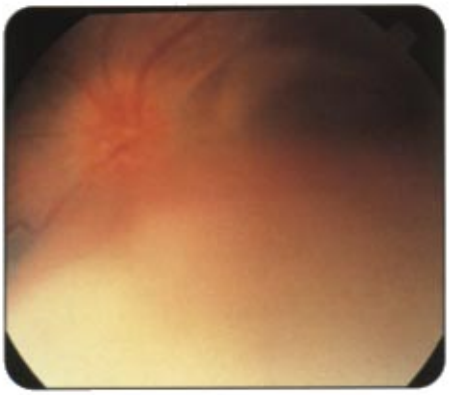

Fig. 12.310

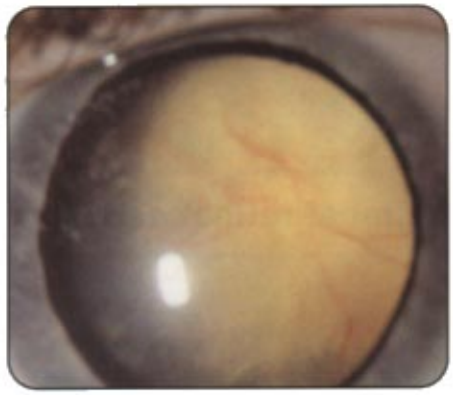

Fig. 12.311

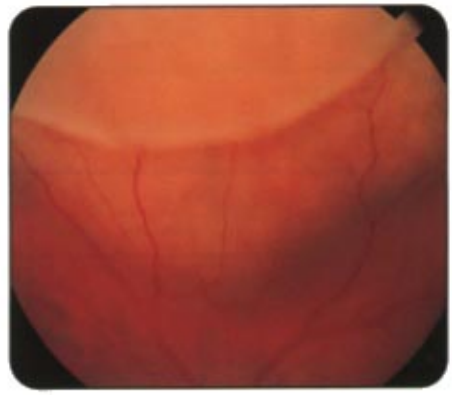

Fig. 12.312

Table 12.11 Causes of exudative retinal detachment

Choroidal tumors	**Systemic**
• primary • metastatic	• hypertension • toxemia of pregnancy • hypoproteinemic states
Intraocular inflammation	
• Harada disease • posterior scleritis • necrotizing retinitis	**Iatrogenic** • retinal detachment surgery • extensive retinal photocoagulation
Leaking subretinal vessels	**Miscellaneous**
• choroidal neovascularization • retinal telangiectasia	• idiopathic bullous central serous retinopathy • uveal effusion syndrome

Differential diagnosis of retinal detachment

Degenerative retinoschisis

Degenerative retinoschisis is a very common, frequently bilateral and, usually, innocuous condition.

Signs

- Retinal elevation, most frequently located infero-temporally.
- Elevation is smooth, thin (**Fig. 12.313**), and relatively immobile, unlike the opaque and corrugated appearance of a rhegmatogenous retinal detachment.
- The inner layer may show 'snowflakes' and sheathing of blood vessels (**Fig. 12.314**)
- Breaks may be present in one (**Fig. 12.315**) or both layers.

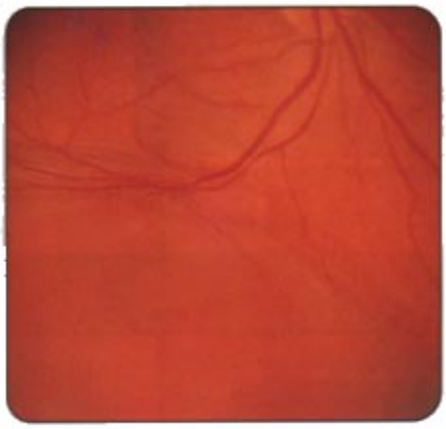

Fig. 12.313

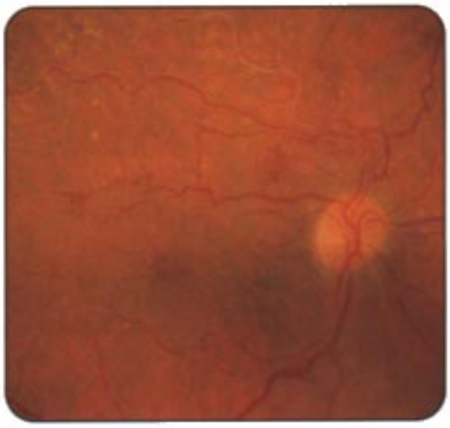

Fig. 12.314

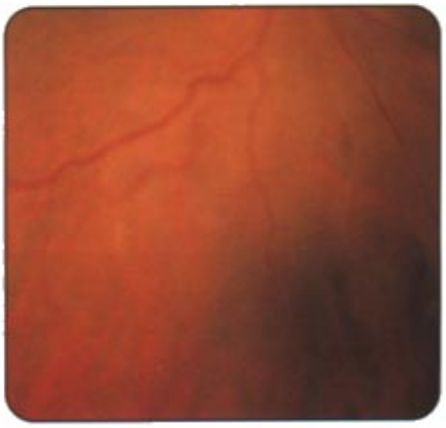

Fig. 12.315

Choroidal detachment

Choroidal detachment is a common condition which typically occurs in eyes with low intraocular pressure.

Signs

- Brown, convex, smooth immobile elevations in the peripheral fundus (**Fig. 12.316**).
- The temporal and nasal elevations are usually most prominent.
- The peripheral fundus and ora serrata my be visualized with ease in severe cases (**Fig. 12.317**).
- Absence of retinal breaks.

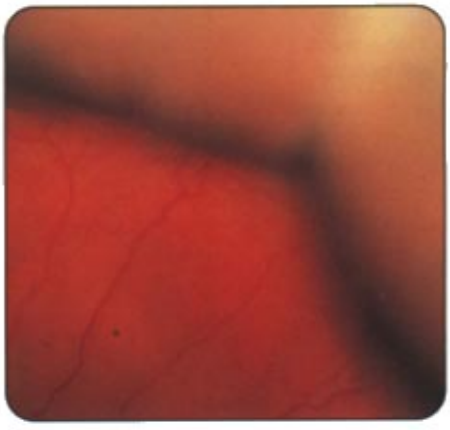

Fig. 12.316

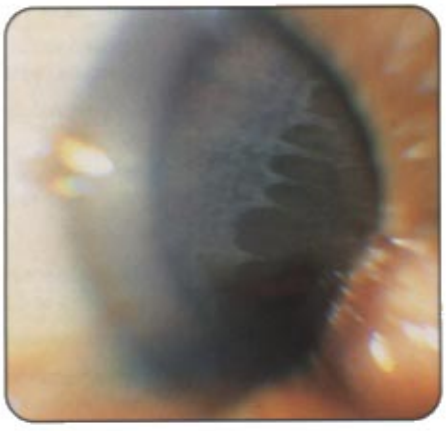

Fig. 12.317

Uveal effusion syndrome

Uveal effusion syndrome is a rare, usually idiopathic condition which may be mistaken for either a retinal detachment complicated by choroidal detachment or a ring choroidal melanoma. In some cases it is associated with nanophthalmos.

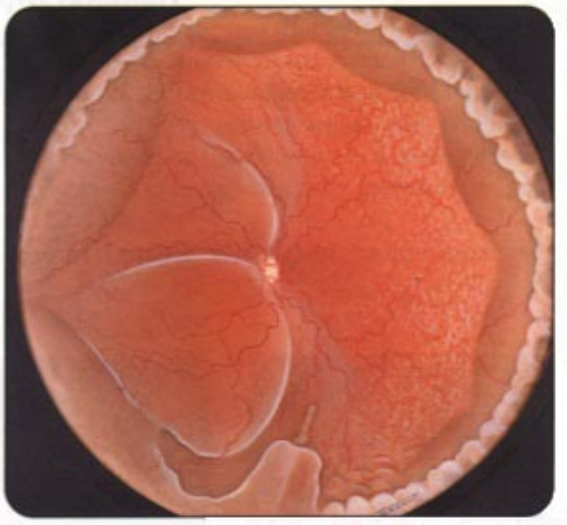

Fig. 12.318

Signs

- Ciliochoroidal detachment associated with exudative retinal detachment (**Fig. 12.318**).
- Absence of retinal breaks.

POOR VISION BUT NORMAL FUNDUS

Table 12.12 Poor vision but normal fundus

1. With normal eye movements	2. In children with abnormal eye movements
In children	• Leber congenital amaurosis – rotary eye movements, high hypermetropia, and 'eye poking' sign
• refractive error	• achromatopsia (rod monochromatism) – nystagmus and photophobia
• amblyopia – check refraction and look for squint	• X-linked recessive congenital stationary night-blindness – nystagmus, nyctalopia, and myopia
• nonorganic visual loss	
• delayed visual maturation – vision improves to normal by 6 months	
• early retinal dystrophies – cone and Stargardt	
• cortical visual impairment	
In adults	
• retrobulbar neuritis – check pupillary reactions and color vision	
• nutritional amblyopia – check color vision	
• cancer-associated retinopathy – fundus may be initially normal	

13 Strabismus

ESODEVIATIONS IN PRIMARY POSITION OF GAZE 382

Angle almost the same for near and distance fixation
- Comitant
- Incomitant

Angle greater for distance than for near fixation
- Comitant
- Congenital incomitant
- Acquired incomitant

Angle greater for near than for distance fixation
- Refractive accommodative esotropia
- Nonrefractive accommodative esotropia
- Nonaccommodative convergence excess (near esotropia)

Pseudo-esodeviations

EXODEVIATIONS IN PRIMARY POSITION OF GAZE 389

Angle almost the same for near and distance fixation
- Congenital comitant
- Acquired comitant
- Incomitant

Angle different for near and distance fixation

Pseudo-exodeviations

ALPHABET PATTERN DEVIATIONS 352

'V' pattern

'A' pattern

VERTICAL DEVIATIONS IN PRIMARY POSITION 396

Hyperdeviation (of nonfixating eye)
- Ipsilateral causes
- Contralateral causes

Hypodeviation (of nonfixating eye)
- Ipsilateral causes
- Contralateral causes

Skew deviation

TORSIONAL DEVIATIONS IN PRIMARY POSITION 401

UPSHOOTS 401

In adduction on version testing

In abduction on version testing

DOWNSHOOTS 403

LIMITATION OF HORIZONTAL EYE MOVEMENTS 403

Limitation of abduction on version testing
- Unilateral
- Bilateral

Limitation of adduction on version testing
- Unilateral
- Bilateral

LIMITATION OF HORIZONTAL VERSIONS (GAZE PALSIES) 407

LIMITATION OF VERTICAL EYE MOVEMENTS 408

LIMITATION OF VERTICAL VERSIONS (GAZE PALSIES) 410

GENERALIZED LIMITATION OF OCULAR MOTILITY 412

Without multiple ocular motor palsies

With multiple ocular motor nerve palsies

PAINFUL OPHTHALMOPLEGIA 414

COMPENSATORY HEAD POSTURES 414

General principles

Head turn
- Strabismic causes
- Nonstrabismic causes

Chin elevation and depression
- Chin elevation
- Chin depression

Head tilt

ESODEVIATIONS IN PRIMARY POSITION OF GAZE

Angle almost the same for near and distance fixation

Most esodeviations will be slightly greater for near than for distance fixation because of physiologic convergence tone. If this difference is less than l0 pd then the deviations can be considered to be almost the same.

Comitant

In comitant deviation the angle is the same, or nearly the same, for all positions of gaze.

1. Infantile esotropia

This is a common condition which develops before the age of 6 months.

Signs

- Large and stable angle (>30 pd) (**Fig. 13.1**).
- Fixation may be alternating in the primary position (**Fig. 13.2a and b**), with cross fixation in side-gaze.
- Normal refraction for age.
- Poor potential for binocular single vision.
- Latent and manifest-latent nystagmus.
- Inferior oblique overaction may be present initially or develop later. **Figure 13.3a** shows the eyes in the primary position and **Figure 13.3b** shows left inferior oblique overaction.
- Asymmetry of optokinetic nystagmus.
- Dissociated vertical deviation (DVD) may develop later.

2. Basic nonaccommodative esotropia

Onset is during childhood but after the age of 6 months.

Signs

- Insignificant refractive error.
- No excess accommodative element (**Fig. 13.4**).

3. Sensory esotropia

This is caused by a unilateral reduction in visual acuity which interferes with or abolishes fusion.
Figure 13.5 shows a left esotropia in a child with iris and retinochoroidal colobomas.

4. Consecutive esotropia

This occurs after surgical overcorrection of an exodeviation. It may be incomitant if there is an immediate large overcorrection caused by a slipped muscle.

5. Microtropia

This is commonly seen following surgery for infantile esotropia. There is a relative scotoma at the fixation point which may, or may not, be foveal.

Signs

- Very small angle which may, or may not, be detected on cover testing. **Figure 13.6** shows a microtropia in which no angle of deviation could be detected on cover test (**Fig. 13.6b and c**) because of the presence of left parafoveal fixation.
- Amblyopia with a marked crowding phenomenon.
- Anisometropia.
- Abnormal retinal correspondence.
- Normal or almost normal peripheral fusional amplitudes.
- Only gross stereoacuity at best.

6. Convergence spasm

This is an intermittent phenomenon which is usually hysterical but may occasionally have an organic cause such as trauma or a posterior fossa tumor.

Signs during an attack

- Esotropia.
- Pseudomyopia due to accommodative spasm.
- Miosis of both pupils.

7. Cyclic esotropia

This rare condition is characterized by alternating manifest esotropia lasting 24 hours followed by normal binocularity for 24 hours. These cycles may last months or years and the patient may eventually develop a constant esotropia which requires surgery.

Signs during an attack

- Large deviation (40–50 pd).
- Diplopia is not usually present.
- Fusional amplitudes are subnormal or absent.

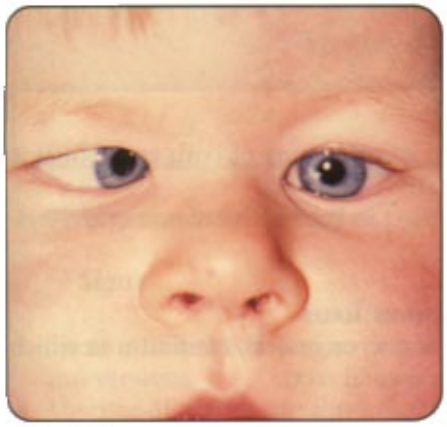

Fig. 13.1

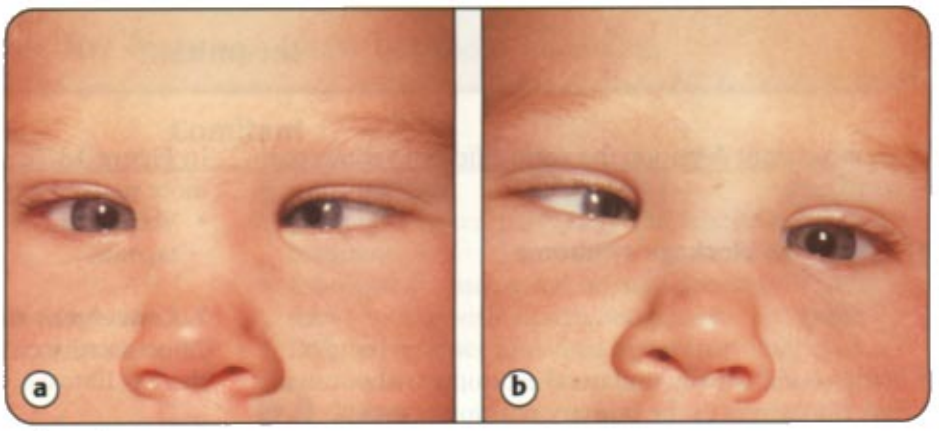

Fig. 13.2

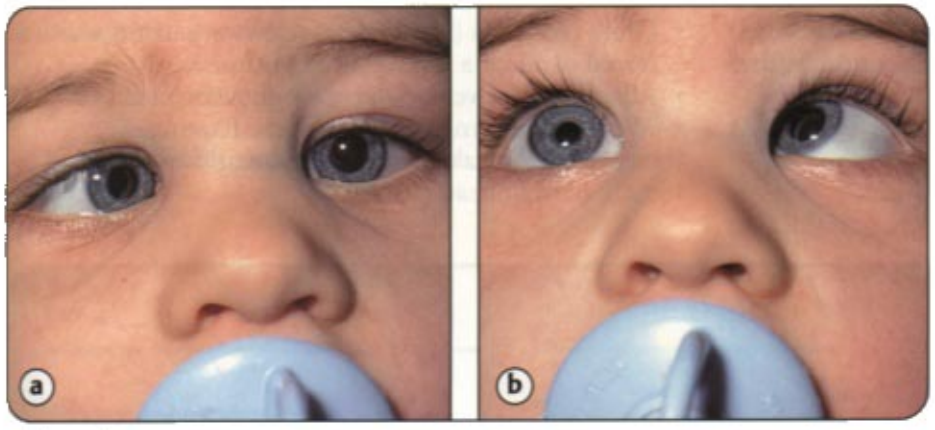

Fig. 13.3

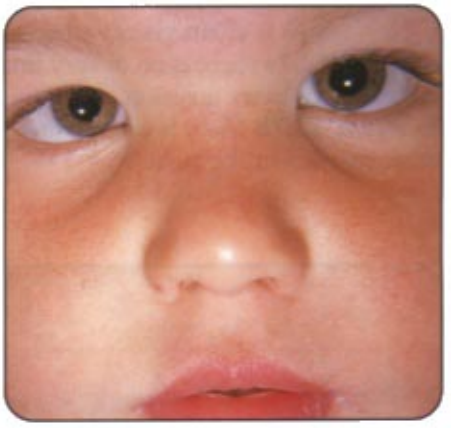

Fig. 13.4

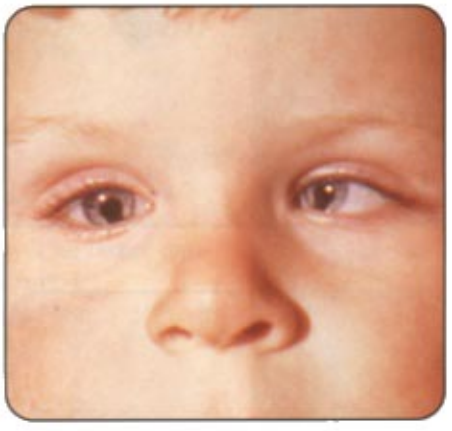

Fig. 13.5

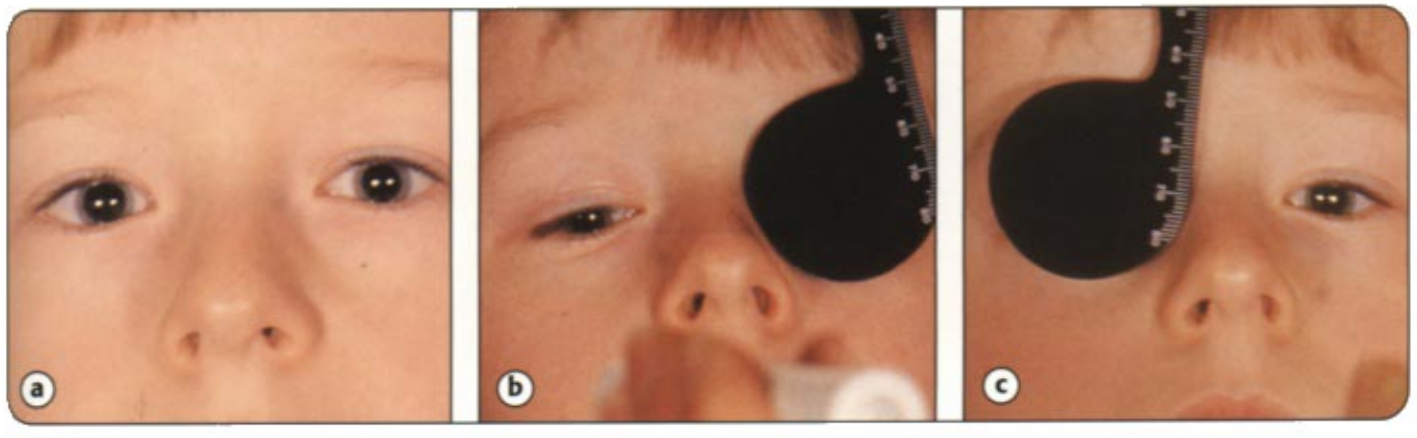

Fig. 13.6

Incomitant

In an incomitant deviation the angle differs in various positions of gaze.

1. Nystagmus blockage syndrome

Signs

- Early-onset, usually unilateral esotropia without manifest nystagmus in the primary position of gaze.
- Angle of deviation is inversely proportional to the amplitude of nystagmus.
- Nystagmus is ellicited only when either eye is abducted.

In **Figure 13.7a** there is no or very fine nystagmus of either eye in the primary position.
In **Figure 13.7b** there is nystagmus of both eyes but it is more marked in the right.
In **Figure 13.7c** there is nystagmus of both eyes but it is more marked in the left.

- Visual acuity is reduced in both eyes because of the nystagmus.

2. Convergent strabismus fixus

This is a very rare, usually congenital, condition in which there is fibrosis of the medial recti.

Signs

- Gross esotropia (**Figure 13.8** shows a patient who was forced to adopt a normal head posture.).
- Absence of active or passive eye movements.
- Marked face turn to the side of the fixating eye.

Figure 13.8 was taken when the patient was forced to adopt a normal head posture.

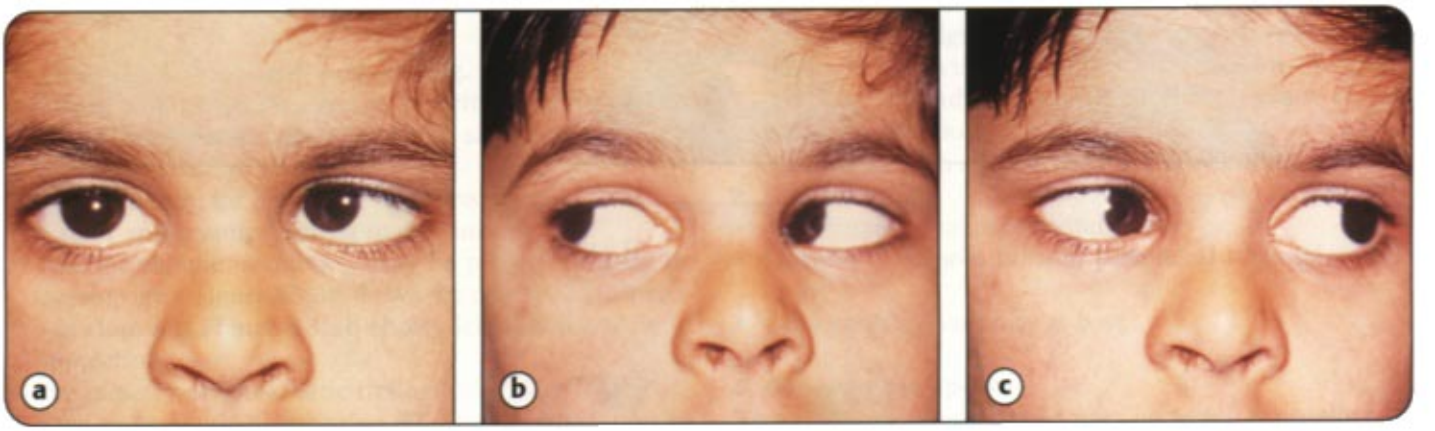

Fig. 13.7

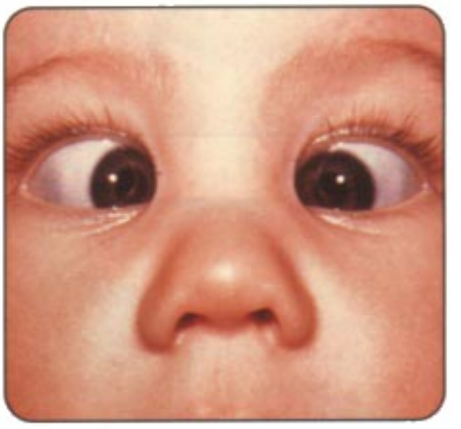

Fig. 13.8

Angle greater for distance than for near fixation

Comitant

1. Divergence insufficiency

Signs

- Intermittent or constant esotropia with normal ductions and versions.
- Absence of neurologic abnormality.
- Reduced fusional divergence amplitude.

2. Divergence paralysis

Signs

- May be difficult to differentiate from unilateral or bilateral sixth nerve palsy, but this condition is usually comitant.
- Esotropia is unchanged or may decrease on laterover-sion – unlike with a sixth nerve palsy.
- Reduced or absent fusional divergence amplitude.

Causes

- Head trauma.
- Intracranial space-occupying lesions.
- Cerebrovascular accidents.

Congenital incomitant

1. Congenital sixth nerve palsy

This is thought to be caused by increased intracranial pressure passage through the birth canal. Spontaneous resolution is the rule if it is infranuclear in origin.

2. Möbius syndrome

Signs

- Bilateral sixth and seventh nerve palsy. The latter typically spares the lower face and is asymmetric.
- Esotropia in the primary position is present in 50% of cases (**Fig. 13.9**).
- Systemic features (*see* Appendix).

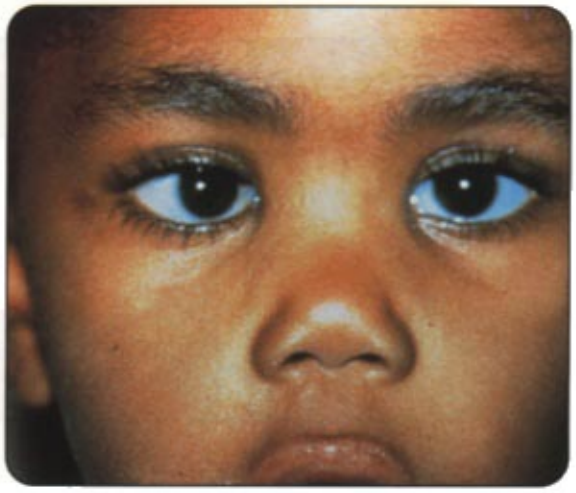

Fig. 13.9

3. Duane syndrome I

Signs (right eye)

- In the primary position the eyes are straight or slightly esotropic (**Fig. 13.10a**).
- Normal or very mildly limited adduction (**Fig. 13.10b**).
- Narrowing of the ipsilateral palpebral fissure and retraction of the globe on adduction (**Fig. 13.10b**).
- Limited abduction on dextroversion (**Fig. 13.10c**).
- Widening of the ipsilateral palpebral fissure on attempted abduction (**Fig. 13.10c**).

4. Duane syndrome III

Signs (left eye)

- In the primary position the eyes are straight (**Fig. 13.11a**) or slightly esotropic.
- Limited abduction on levoversion (**Fig. 13.11b**).
- Limited adduction on dextroversion (**Fig. 13.11c**).
- Narrowing of the palpebral fissure and retraction of the globe on attempted adduction (**Fig. 13.11c**).

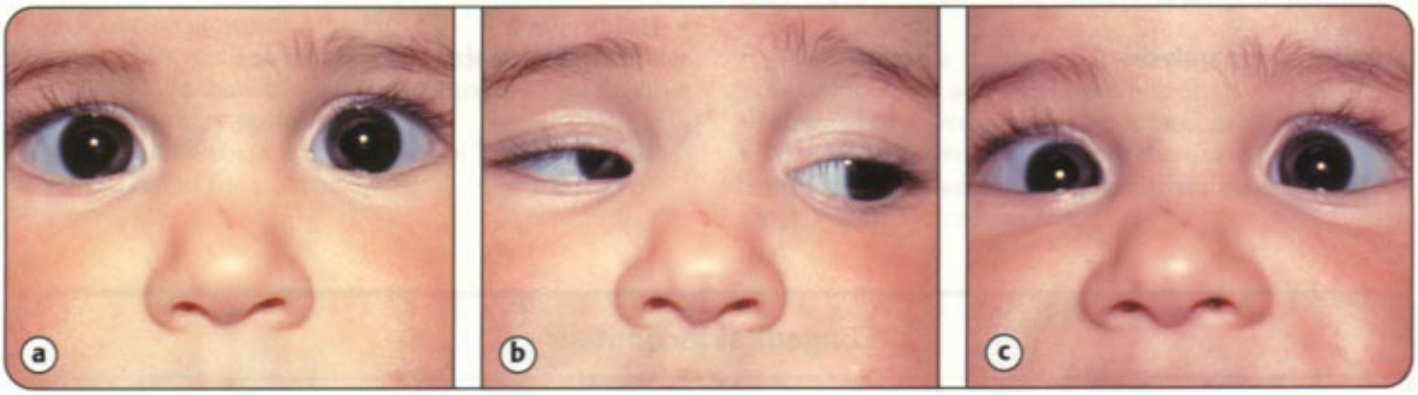

Fig. 13.10

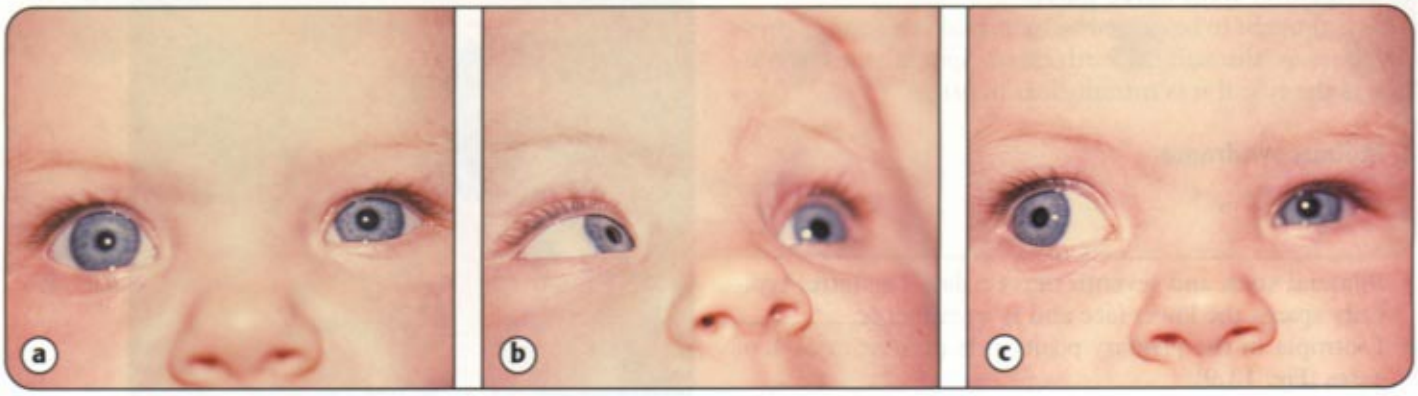

Fig. 13.11

Acquired incomitant

1. Recent-onset sixth nerve palsy

Signs (left eye)

- Esotropia which is frequently greater for distance (**Fig. 13.12a**) than for near (**Fig. 13.12b**). In this case the patient prefers to fixate with the dominant left eye.
- Normal left adduction.
- Grossly reduced left abduction and an increase in the angle on ipsilateral lateroversion (**Fig. 13.12c**).

2. High myopia

Very rarely the enormous globe compresses the lateral rectus muscles against the wall of the orbit.

3. Iatrogenic

This may occur as a result of a slipped lateral rectus muscle during squint or retinal detachment surgery.

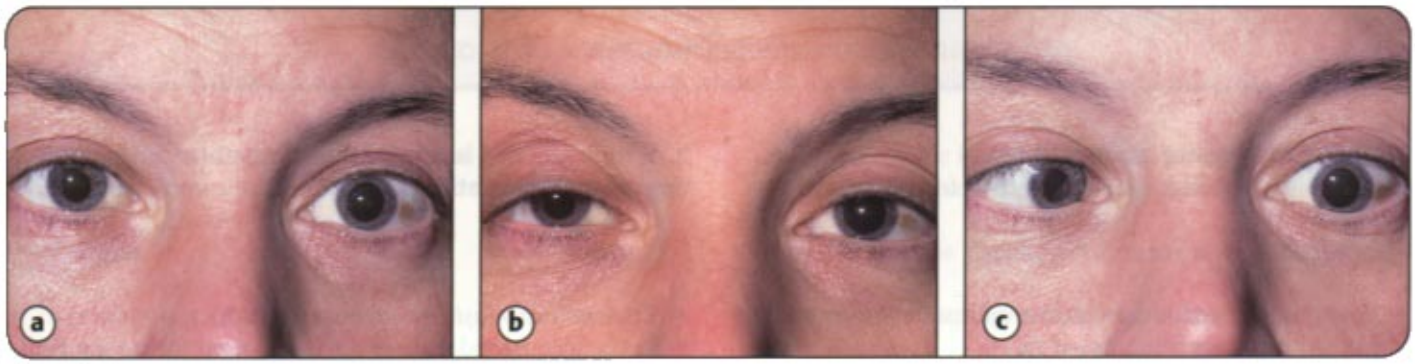

Fig. 13.12

Angle greater for near than for distance fixation

Refractive accommodative esotropia

1. Fully accommodative

Onset is usually between 2 and 3 years and rarely earlier.

Signs

- Hypermetropia.
- Spectacles fully correct the deviation (**Fig. 13.13a and b**).

2. Partially accommodative esotropia

Signs

- Spectacles partly correct the deviation (**Fig. 13.14**).

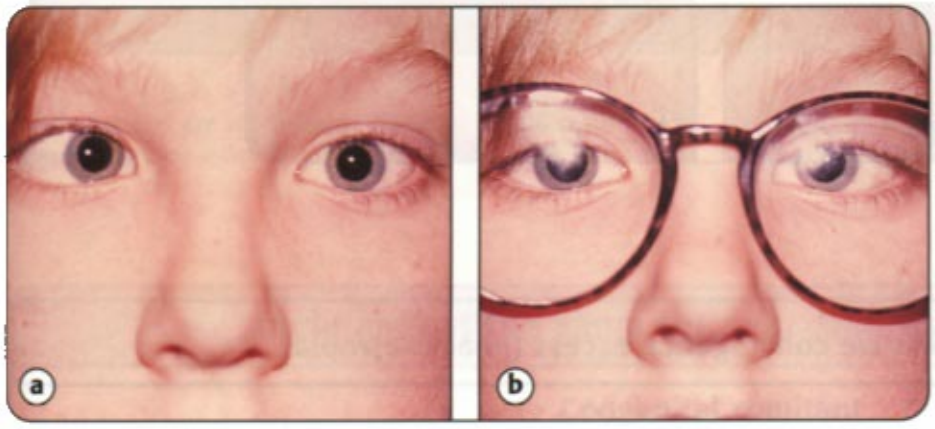

Fig. 13.13

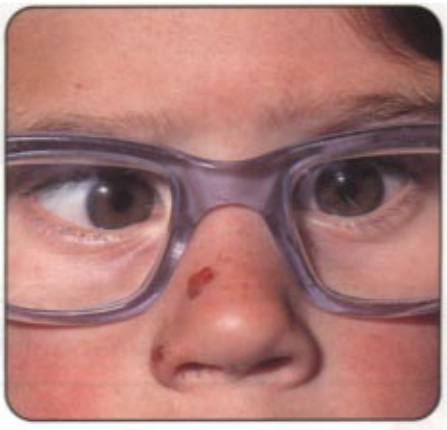

Fig. 13.14

Nonrefractive accommodative esotropia

1. With convergence excess

Onset is usually after 6 months but before 3 years.

Signs

- High accommodative convergence/accommodation (AC/A) ratio, with increased AC.
- Normal near point of accommodation.
- No significant refractive error.
- Straight eyes for distance (**Fig. 13.15a**) but esotropia for near (**Fig. 13.15b**).

Figure 13.15 was taken without an accommodative target. The deviation seen is therefore smaller than it would be with an accommodative target.

2. With defective accommodation (hypoaccommodative)

Signs

- Remote near point of accommodation.
- Extra accommodative effort is required for near, resulting in convergence excess.
- High AC/A ratio is due to decreased accommodation.

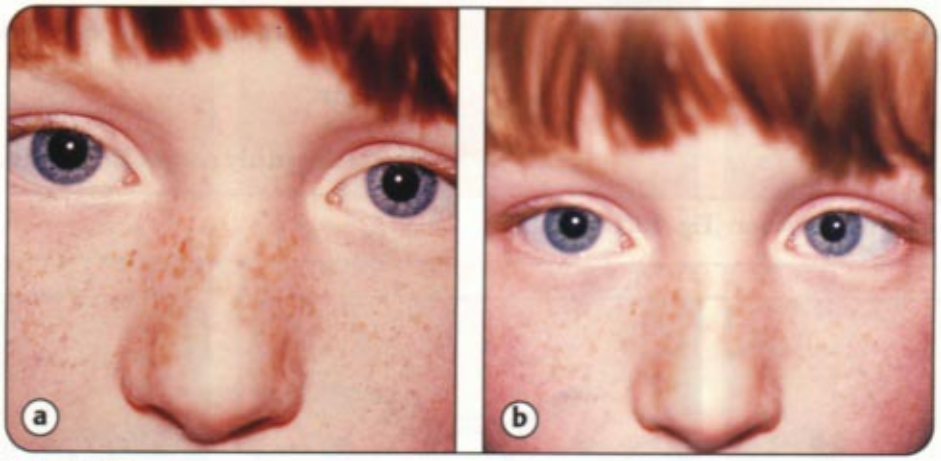

Fig. 13.15

Nonaccommodative convergence excess (near esotropia)

Signs

- Normal or low AC/A ratio.
- Normal near point of accommodation.
- No significant refractive error.
- Normal visual acuity in both eyes.

Pseudo-esodeviations

1. Epicanthic folds and a broad nasal bridge
This is very common and may may simulate an esotropia.

Signs

- Symmetric and central corneal light reflexes (**Fig. 13.16**).
- No deviation on cover–uncover test.

2. Negative angle kappa
Angle kappa is the angle formed by the intersection of the pupillary and visual axes. The pupillary axis runs perpendicular to the cornea and through the center of the pupil. The visual axis runs from the point of fixation to the foveola.

Signs

- Normally, a corneal light reflex is slightly displaced nasally (positive angle kappa).
- In high myopia the reflection may be displaced temporally (negative angle kappa).
- A negative angle kappa may therefore simulate an esodeviation, accentuate an existing esodeviation, or mask an exodeviation.
- A negative angle kappa is not necessarily pathologic.

3. Narrow interpupillary distance in hypotelorism

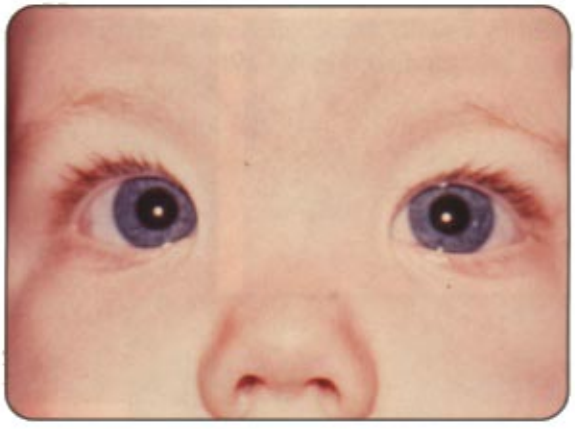

Fig. 13.16

EXODEVIATIONS IN PRIMARY POSITION OF GAZE

Exodeviations are much more likely to be intermittent or latent than are esodeviations. Constant comitant exotropia is usually encountered in the older patient with either a sensory or a decompensating intermittent exodeviation.

Angle almost the same for near and distance fixation

Congenital comitant

1. Congenital exotropia
This is much less common than infantile esotropia.

Signs

- Constant alternating exotropia.
- DVD may be seen.

2. Hypertelorism
This is a congenital anomaly in which there is unusually wide separation of the orbits (*see* Chapter 2).
Figure 13.17 shows a child with Crouzon disease and gross exotropia.

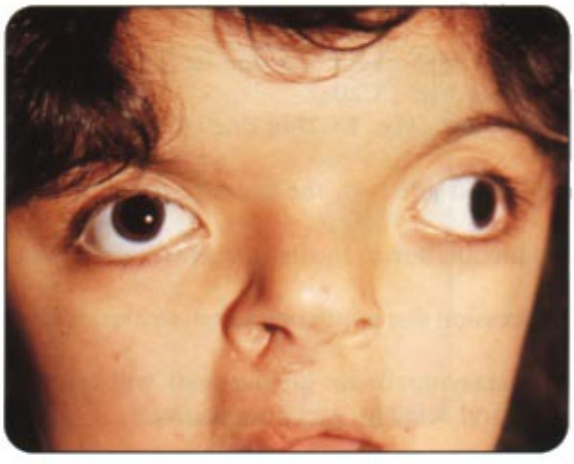

Fig. 13.17

Acquired comitant

1. Basic exotropia

Signs

- Frequently intermittent exodeviation.
- Alternating fixation (**Fig. 13.18**) usually present.

2. Sensory exotropia

This is caused by usually uniocular sensory visual deprivation such as by a cataract (**Fig. 13.19**).

Signs

- Constant exodeviation.

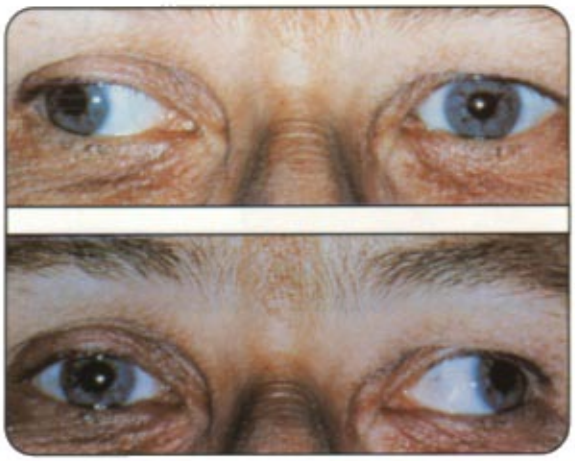

Fig. 13.18

3. Microexotropia

This may occur in isolation or in association with a primary intermittent or constant exotropia.

Signs

- Associated amblyopia and anisometropia.

4. Consecutive exotropia

Causes

- Spontaneous.
- Following strabismus surgery for esotropia – the deviation may be incomitant if a muscle has slipped.

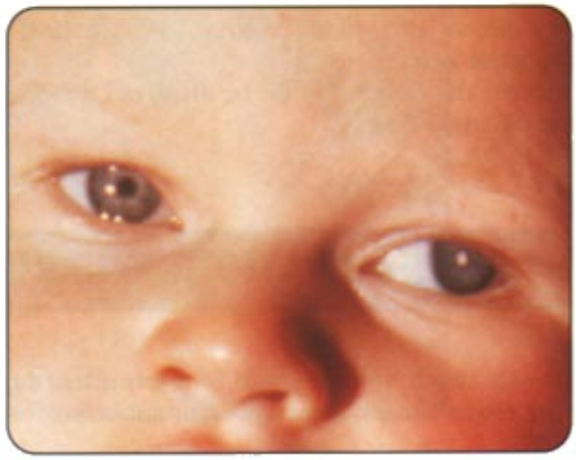

Fig. 13.19

Incomitant

1. Third nerve palsy

Signs (right eye)

- Limited elevation in abduction due to right superior rectus weakness (**Fig. 13.20a**).
- Normal abduction (**Fig. 13.20b**) due to intact lateral rectus.
- Exodeviation and ptosis in the primary position (**Fig. 13.20c**).
- Limited adduction due to medial rectus weakness (**Fig. 13.20d**).
- Limited depression due to inferior rectus weakness (**Fig. 13.20e**).
- If present, parasympathetic involvement will cause a dilated pupil and defective accommodation.

2. Duane syndrome II

Signs

- Limited adduction.
- Narrowing of palpebral fissure on attempted adduction.
- Associated A and V patterns are common.

3. Divergent strabismus fixus

This is a very rare usually congenital condition in which there is fibrosis of the lateral recti.

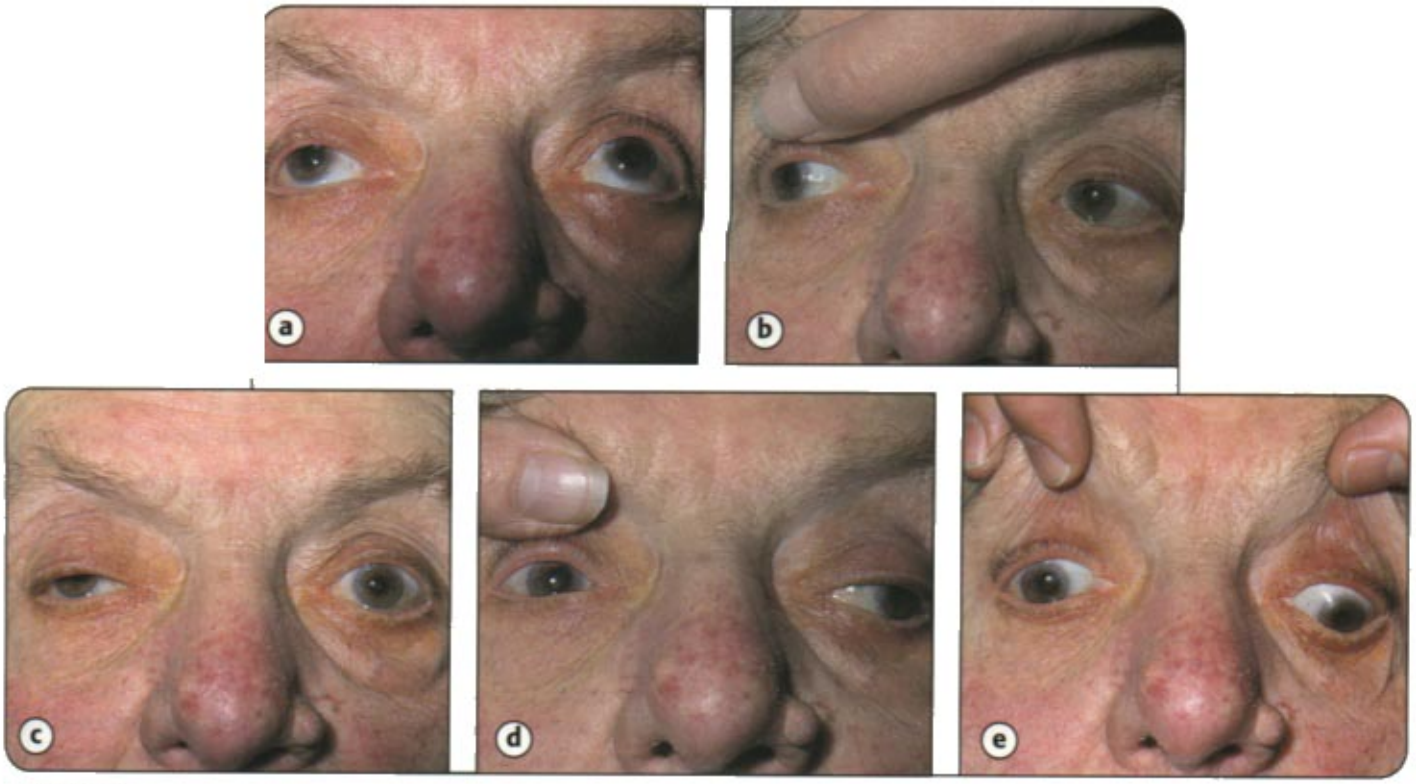

Fig. 13.20

Angle different for near and distance fixation

1. Convergence insufficiency (near exotropia)
This is an acquired comitant condition seen in older children and adults.

Signs

- Exotropia greater for near.
- Usually intermittent.
- Poor or absent binocular convergence.
- May be associated with acquired myopia.

2. Divergence excess
This is an acquired comitant condition which is usually intermittent. It may be true or simulated. The latter is associated with a high AC/A ratio.

Signs

- In true divergence excess, angle for near (**Fig. 13.21a**) is consistently less than for distance (**Fig. 13.21b**).
- In simulated divergence excess, deviations for near and distance are similar when the angle is remeasured with the patient looking through a +3.00 D lens or after a period of uniocular occlusion.

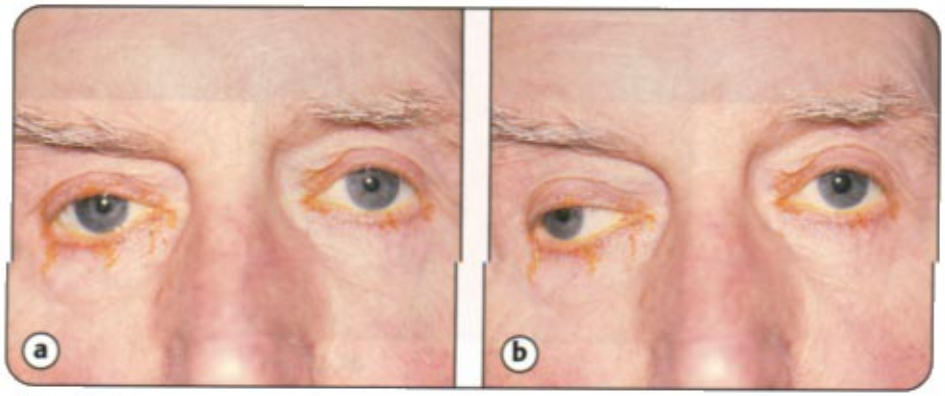

Fig. 13.21

Pseudo-exodeviations

1. Positive angle kappa without ocular anomalies

Signs

- Corneal light reflex displaced slightly nasally when fixating a light.
- This may give a false impression of an exodeviation.

2. Positive angle kappa with ocular anomalies

This is caused by macular heterotopia with temporal displacement of the fovea; consequently there is a marked nasal displacement of the corneal light reflex.

ALPHABET PATTERN DEVIATIONS

Horizontal deviations can vary when measured in the primary position, in upgaze and downgaze. Regardless of whether a horizontal deviation is comitant or incomitant it can still have vertical incomitance.

- 'V' pattern – this is significant if the difference between upgaze and downgaze is 15 pd or more (**Fig. 13.22**).
- 'A' pattern – this is significant if the difference between upgaze and downgaze is 10 pd or more (**Fig. 13.23**).
- 'X' pattern – orthophoria in the primary position but exotropia in upgaze and downgaze. This is usually seen in cases with both inferior and superior oblique overactions.
- 'Y' pattern – divergence only in upgaze.
- Inverted 'Y' (lambda) pattern – divergence only in downgaze.

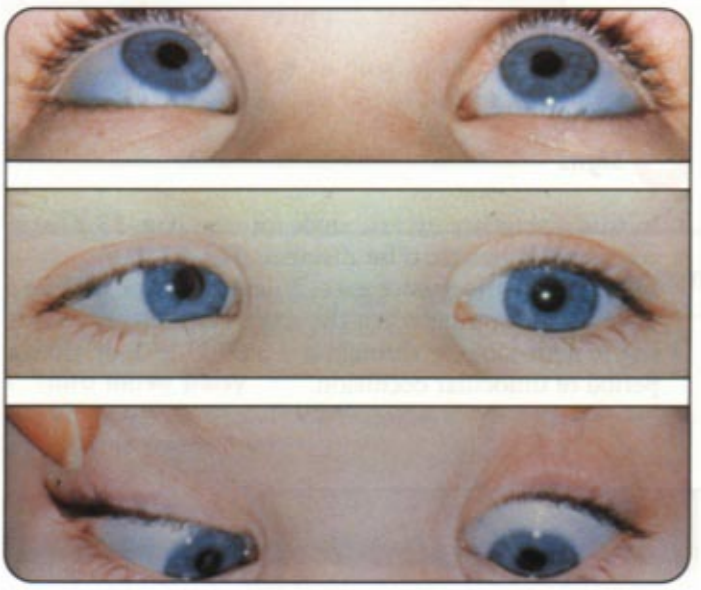

Fig. 13.22

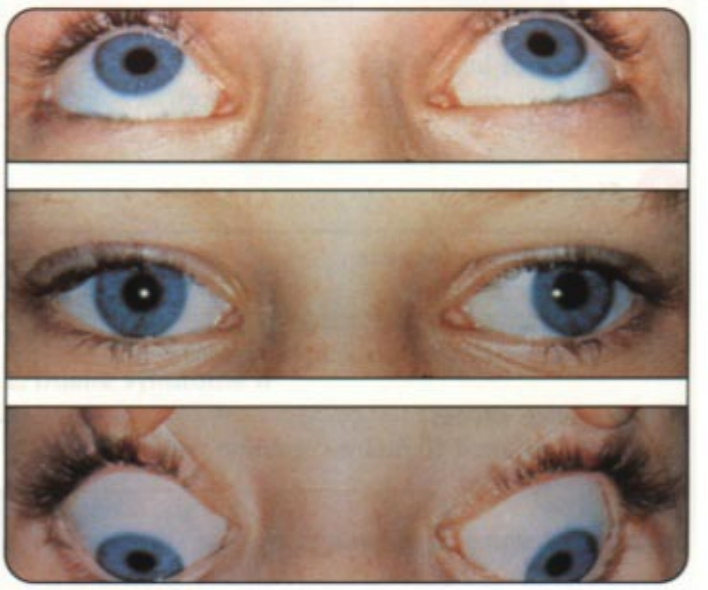

Fig. 13.23

'V' pattern

1. Brown syndrome

Signs (left eye)

- Eyes straight in the primary position (**Fig. 13.24a**).
- Limited elevation in adduction (**Fig. 13.24b**) and occasionally also in the primary position.
- Usually normal elevation in abduction (**Fig. 13.24c**) .
- Positive forced duction test on elevating globe in adduction.
- Downshoot in adduction – variable.
- Hypotropia in primary position – variable.
- Anomalous head position – ipsilateral head tilt with chin up – variable.

Causes

- Idiopathic congenital.
- Congenital click syndrome – impaired movement of tendon through trochlea.
- Iatrogenic damage to trochlea or superior oblique tendon.
- Inflammation of the tendon which may be caused by rheumatoid arthritis (**Fig. 13.25** shows involvement of the left eye), pansinusitis and scleritis.

Differential diagnosis

a. Inferior oblique underaction

- 'A' pattern.
- Negative forced duction test on elevation in adduction.
- More pronounced vertical deviation in primary position.

b. Double elevator palsy

- Restricted elevation in all positions of gaze.
- Frequently associated with ptosis.

NB Occasionally, an 'A' pattern may be seen in Brown syndrome.

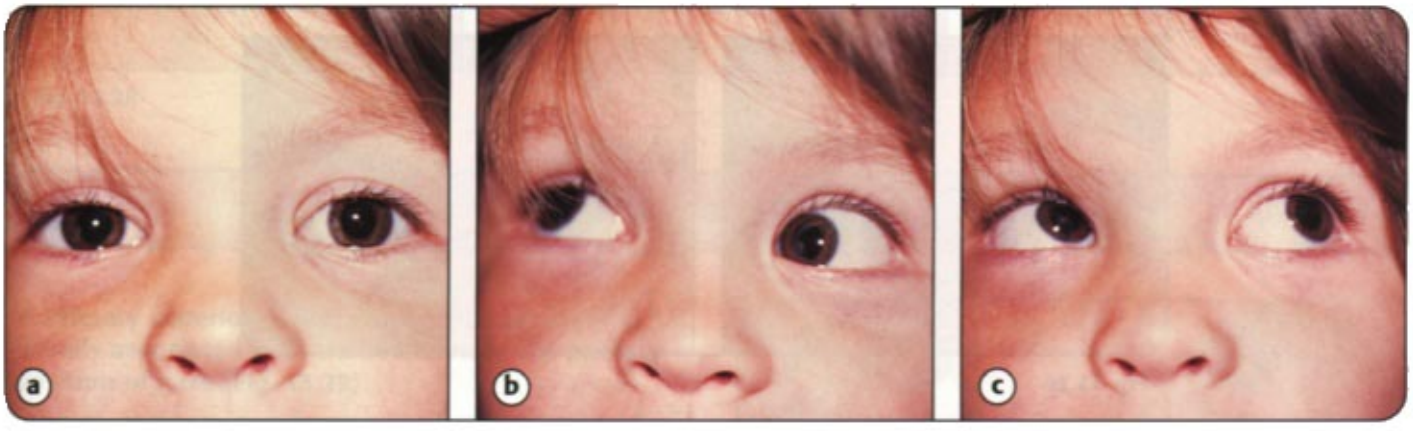

Fig. 13.24

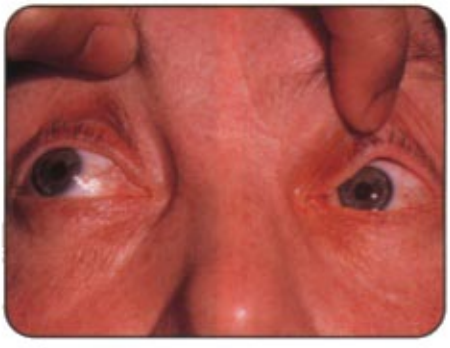

Fig. 13.25

2. Primary inferior oblique overaction

Causes

- Infantile esotropia.
- Other esotropias with onset during first few years of life.

3. Superior oblique underaction with consequent inferior oblique overaction

Signs

- Eyes straight in upgaze (**Fig. 13.26a**).
- Marked esodeviation in downgaze (**Fig. 13.26b**).
- Left inferior oblique overaction (**Fig. 13.26c**).
- Right inferior oblique overaction (**Fig. 13.26d**).

Causes

- Bilateral fourth nerve palsy after closed head trauma.
- Congenital fourth nerve palsy – usually unilateral.

4. Craniofacial anomalies

These are associated with shallow orbits and downslanting palpebral fissures, as in this patient with Crouzon disease.

Signs

- Gross exotropia in upgaze (**Fig. 13.27a**).
- Almost straight eyes in downgaze (**Fig. 13.27b**).
- Exotropia in primary position view (*see* **Fig. 13.17**).

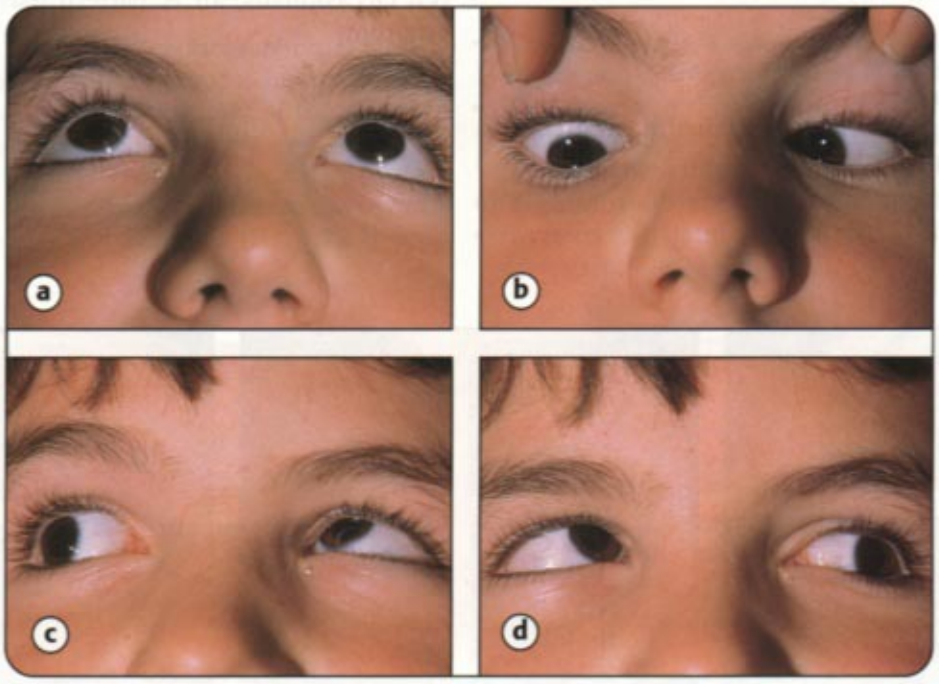

Fig. 13.26

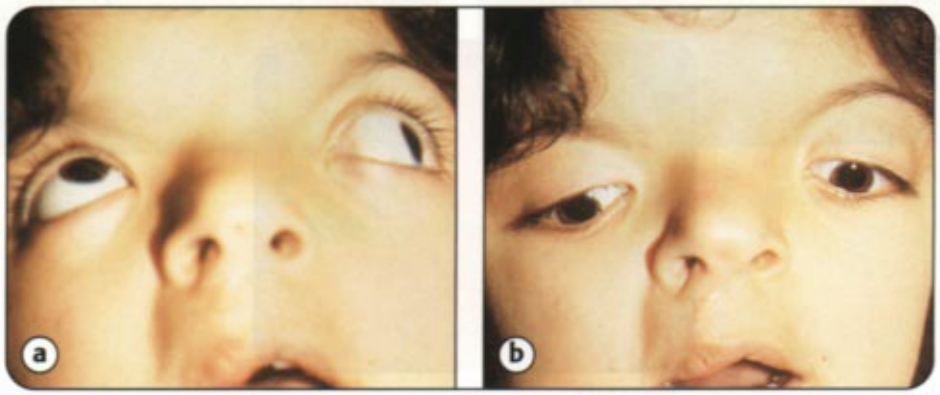

Fig. 13.27

5. Desagittalization of the superior oblique

This occurs in craniofacial anomalies, especially coronal suture synostosis (plagiocephaly) in which there is recession of the frontal bones (**Fig. 13.28a**) and therefore also of the trochlea. The mechanical disadvantage of the superior oblique tendon results in underaction. **Figure 13.28c** shows left superior oblique underaction.

Signs

- Exotropia in upgaze (**Fig. 13.28b**).
- Eyes almost straight in downgaze (**Fig. 13.28d**).
- Gross left inferior oblique overaction (**Fig. 13.28e**).

6. Superior rectus underaction

7. Lateral rectus overaction

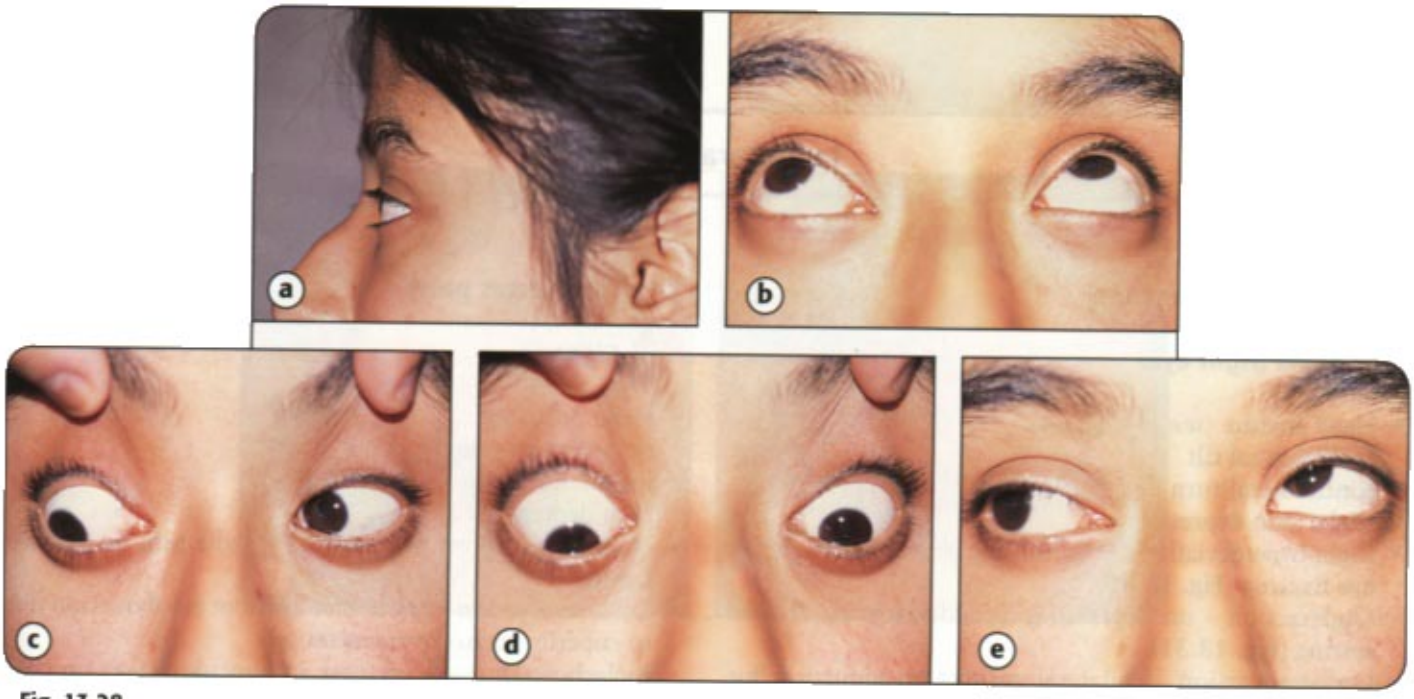

Fig. 13.28

'A' pattern

1. Primary superior oblique overaction which is usually associated with exodeviation in the primary position of gaze (Fig. 13.29)

2. Inferior oblique underaction with consequent superior oblique overaction

3. Lateral rectus underaction

4. Inferior rectus underaction

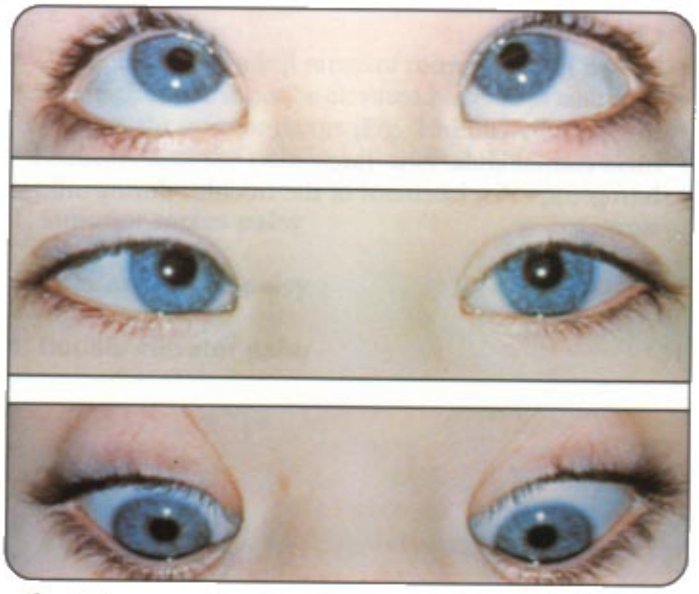

Fig. 13.29

VERTICAL DEVIATIONS IN PRIMARY POSITION

Hyperdeviation (of nonfixating eye)

Hyperdeviation may be primary, caused by compromise of ipsilateral depressors, with the nonparetic eye fixating. It may also be secondary, caused by compromise of contralateral elevators, with the paretic eye fixating. All the following conditions are incomitant.

Ipsilateral causes

1. Superior oblique palsy

Signs (right eye)

- Head posture (*see* **Fig. 13.64**):
  contralateral tilt
  contralateral turn
  slight chin depression.
- Right hyperdeviation in the primary position with the left eye fixating (**Fig. 13.30**).
- Underaction – on depression in adduction on version testing (**Fig. 13.31**).
- On lateroversion – ipsilateral upshoot on adduction due to inferior oblique overaction.
- Bielschowsky test – increase in hyperdeviation on ipsilateral head tilt (**Fig. 13.32a and b**).
- Torsion – excyclotorsion.

Causes

- Congenital.
- Closed head trauma.
- Vascular.
- Intra-cranial tumor.
- Intra-cranial aneurysm (rare).
- Iatrogenic – displacement of the trochlea during orbital surgery.

2. Inferior rectus palsy

Signs

- Head posture:
  tilt to either side
  ipsilateral turn
  chin depressed.
- Underaction – on depression in abduction on version testing.
- On lateroversion – ipsilateral upshoot on abduction due to superior rectus overaction.
- Bielschowsky test – increase in hyperdeviation on contralateral tilting.
- Torsion – incyclotorsion.

Causes

- Congenital.
- Trauma.
- Myasthenia gravis.

3. Congenital absence or structural anomaly of superior oblique

This may be seen in certain craniofacial anomalies such as Apert syndrome.

4. Double depressor palsy (very rare)

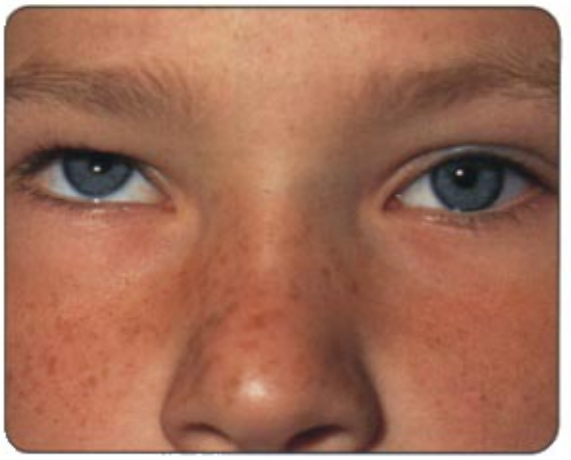

Fig. 13.30

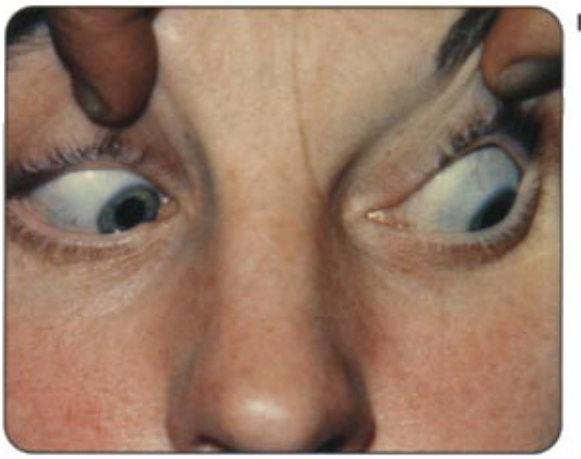

Fig. 13.31

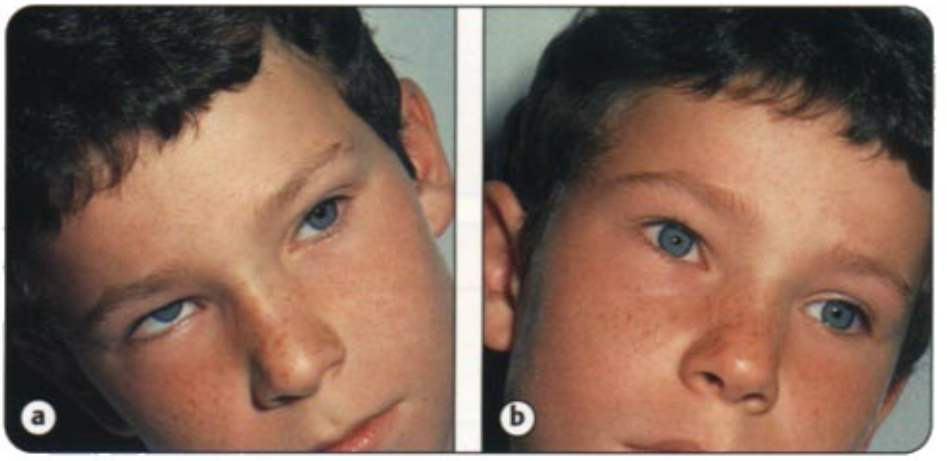

Fig. 13.32

Contralateral causes

1. Non-neurological inferior rectus lesions

Signs in thyroid eye disease (right inferior rectus)

- The patient chooses to fixate with the right (dominant) eye, which gives the impression of a left hypertropia in the primary position (**Fig. 13.33a**).
- The left eye has a full range of movements.
- The right eye cannot be elevated because of contracture of the right inferior rectus (**Fig. 13.33b and c**).
- Depression of the right eye is normal (**Fig. 13.33d**).

2. Superior rectus palsy

3. Inferior oblique palsy

4. Double elevator palsy

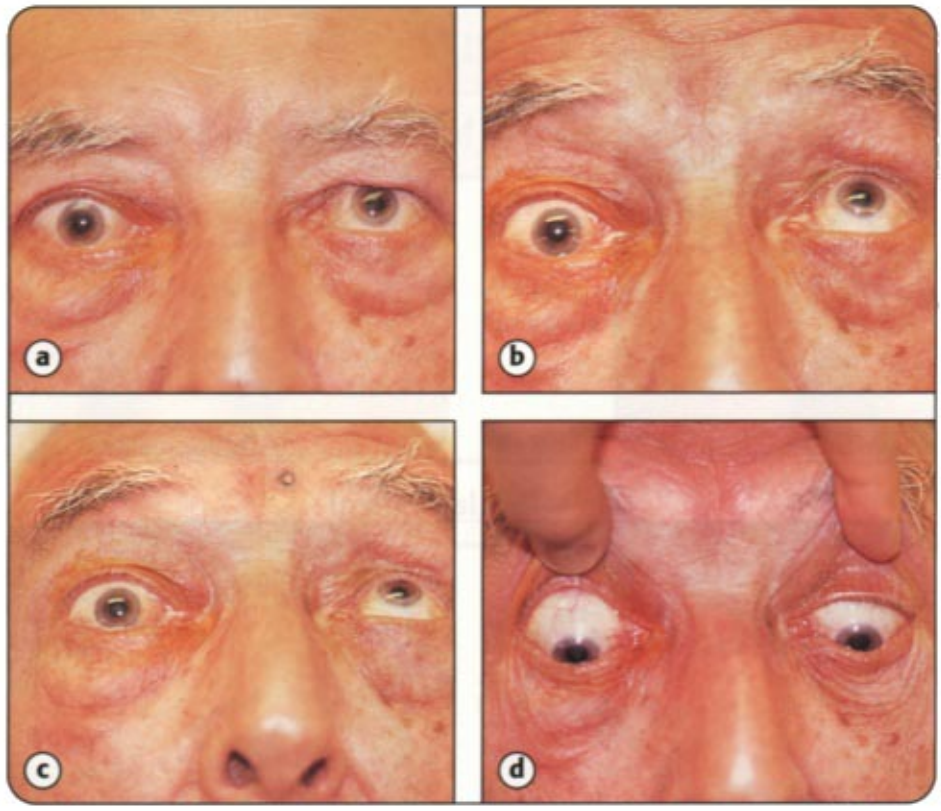

Fig. 13.33

Hypodeviation (of nonfixating eye)

Hypodeviation may be primary, with the nonparetic eye fixating, or secondary, with the paretic eye fixating.
All the following conditions are incomitant.

Ipsilateral causes

1. Superior rectus palsy

Signs (left eye)

- Head posture:
  tilt usually to contralateral side but may be to either ipsilateral turn (if recent onset)
  chin elevated (if recent onset).
- In the primary position the left eye is hypotropic as noted by the corneal light reflexes (**Fig. 13.34e**).
- Underaction – on elevation in abduction on version testing (**Fig. 13.34c**)
- Elevation in adduction:
  normal if recent onset (**Fig. 13.34a**)
  limited if longstanding.
- Absent Bell phenomenon.
- On lateroversion – contralateral upshoot on adduction due to inferior oblique overaction and ipsilateral downshoot due to inferior rectus overaction (**Fig. 13.34f**).
- Bielschowsky test – increase in hypodeviation on ipsilateral head tilt.
- Torsion – small excyclotropia.
- Other movements are normal (**Fig. 13.34a, b, d g, h and i**).

Causes

- Congenital.
- Trauma during surgery by traction suture.
- Myasthenia gravis in which there may also be ptosis (**Fig. 13.34e**).

2. Inferior oblique palsy

Signs

- Head posture:
  ipsilateral tilt
  contralateral turn
  chin elevation.

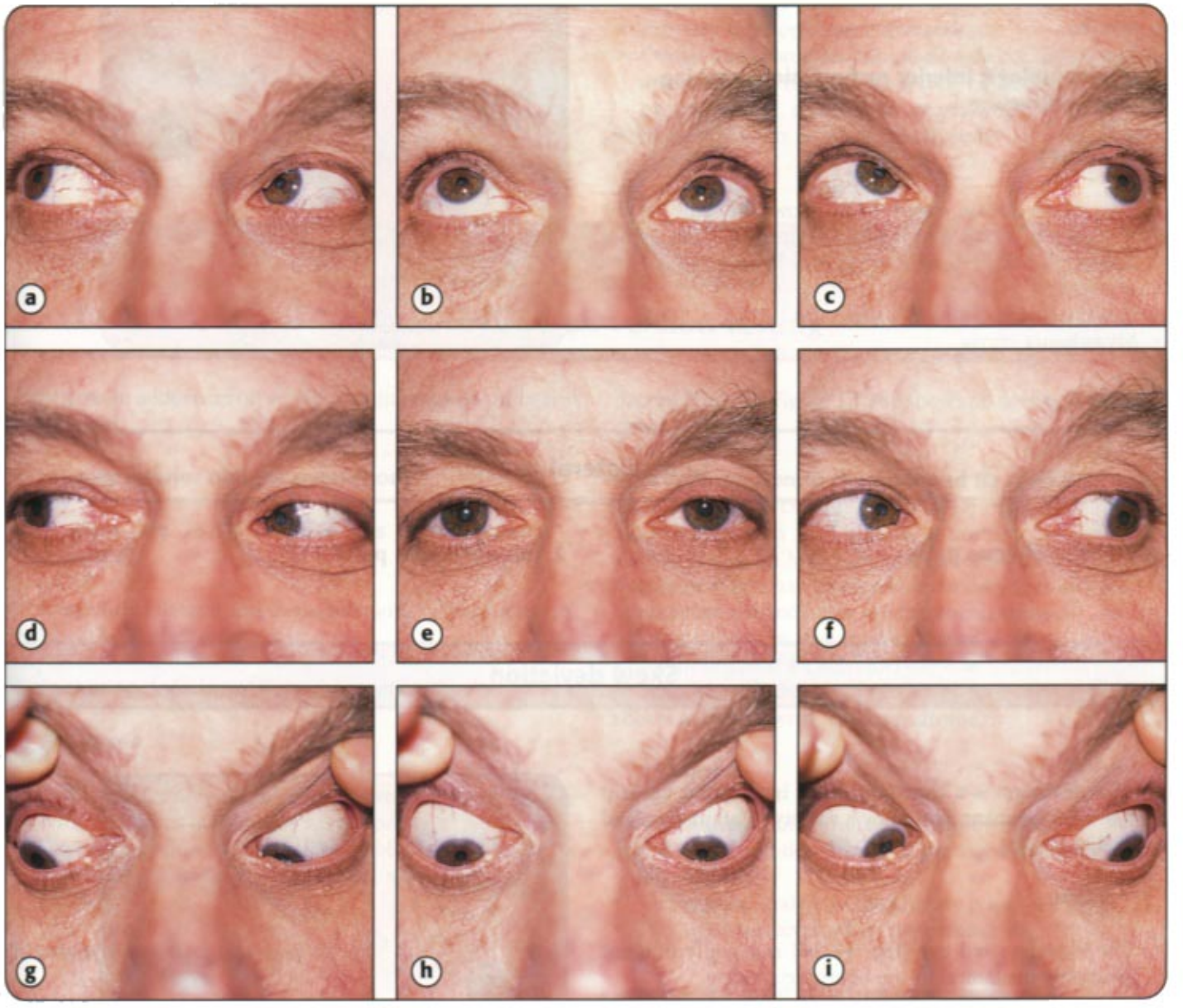

Fig. 13.34

- Underaction – on elevation in adduction on version testing.
- On lateroversion – contralateral upshoot on abduction due to superior rectus overaction.
- Bielschowsky test – increase in hypodeviation on contralateral tilting.
- Torsion – incyclotorsion.

Causes

- Isolated involvement is very rare and almost always congenital.

3. Non-neurologic inferior rectus lesions causing muscle contracture

Causes

- Thyroid eye disease. **Figure 13.35** shows the same patient as in **Figure 13.33** who is now fixating with the normal left eye; the affected right eye is now hypotrophic.
- Myositis.
- Entrapment in orbital floor fracture.
- Myasthenia gravis.

4. Double elevator palsy

5. Brown syndrome – if severe

6. Congenital absence of superior rectus
This may be seen in certain craniofacial anomalies.

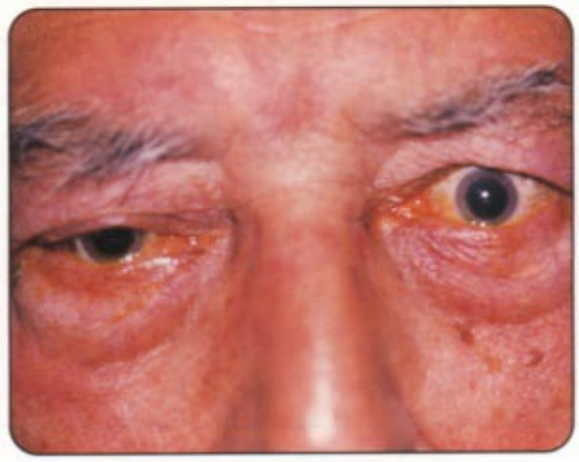

Fig. 13.35

Contralateral causes

1. Superior oblique palsy

2. Inferior rectus palsy

Skew deviation

A skew deviation is a concomitant or incomitant vertical ocular misalignment caused by a variety of brainstem and cerebellar lesions.

Signs

- The hypodeviated eye (**Fig. 13.36**) is usually but not invariably ipsilateral to the neurologic lesion.
- Incomitant cases can be differentiated from cyclovertical muscle palsy by the coexistence of central neurologic signs.
- The hypodeviated eye is excyclotorted.

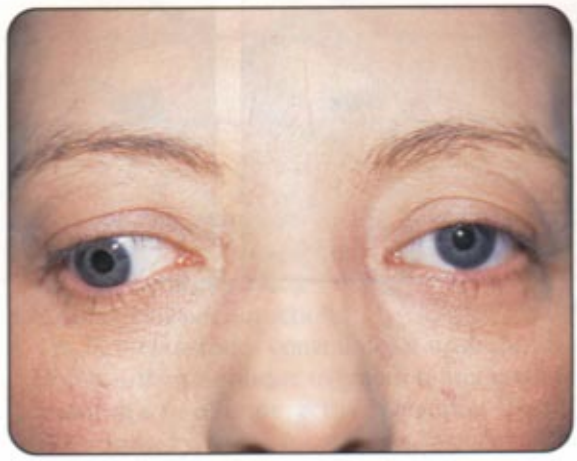

Fig. 13.36

TORSIONAL DEVIATIONS IN PRIMARY POSITION

1. Incyclotorsion

Causes

- Inferior rectus palsy.
- Inferior oblique palsy.

2. Excyclotorsion

Causes

- Superior oblique palsy.
- Superior rectus palsy.
- Primary inferior oblique overaction.
- Ocular and orbital excyclorotation. In certain craniofacial anomalies the entire orbit and its contents may become excyclorotated, resulting in excyclotropia of the globe.
- Skew deviation in which the hypodeviated eye is excyclotorted.

UPSHOOTS

In adduction on version testing

Upshoots in adduction on version testing may be unilateral or bilateral.

1. Primary inferior oblique overaction

Signs (Fig. 13.37; see also Figs 13.3b, 13.26c and d)

- Frequently bilateral and associated with 'V' esodeviation.
- Negative Bielschowsky test.

2. Secondary inferior oblique overaction

Causes

- With superior oblique palsy – positive Bielschowsky test.
- With contralateral superior rectus palsy.
- Desagittalization of the superior oblique muscle, most commonly associated with coronal synostosis (*see* **Fig. 13.28**).

3. Duane syndrome I, II, and III

4. Dissociated vertical deviation (DVD)

This may on cursory examination resemble an inferior oblique overaction.

Distinguishing features include:

- Upshoot in all positions of gaze.
- Covered eye is higher in DVD, whereas in inferior oblique overaction the covered eye is lower if the affected eye is fixating.

Figure 13.38 shows consecutive frames of a video clip of a child with left DVD. **Figure 13.38a** shows left hyperdeviation just as the eye is being uncovered; **Figure 13.38b** shows the patient blinking; **Figure 13.38c** shows no deviation with both eyes open.

5. Excyclorotation of the orbit and its contents

This is seen in certain craniofacial syndromes. **Figure 13.39a** shows an MRI scan of a normal patient. **Figure 13.39b** shows an MRI scan of a patient with craniofrontal dysplasia which has given rise to an excyclorotation of the orbital contents including the extraocular muscles. Because of excyclorotation, the medial rectus is no longer a pure adductor but also an elevator; likewise the lateral rectus is no longer a pure abductor but also a depressor. **Figure 13.40** shows a patient with Apert syndrome.

Signs

- On dextroversion there is upshoot of the left eye in adduction and downshoot of the right eye in abduction (**Fig. 13.40a**).
- The eyes are straight in the primary position (**Fig. 13.40b**).
- The signs are reversed in levoversion (**Fig. 13.40c**).

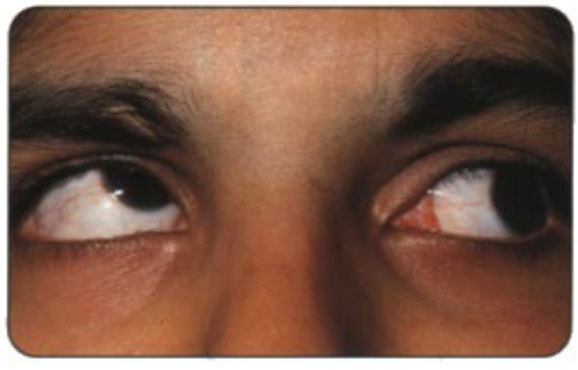

Fig. 13.37

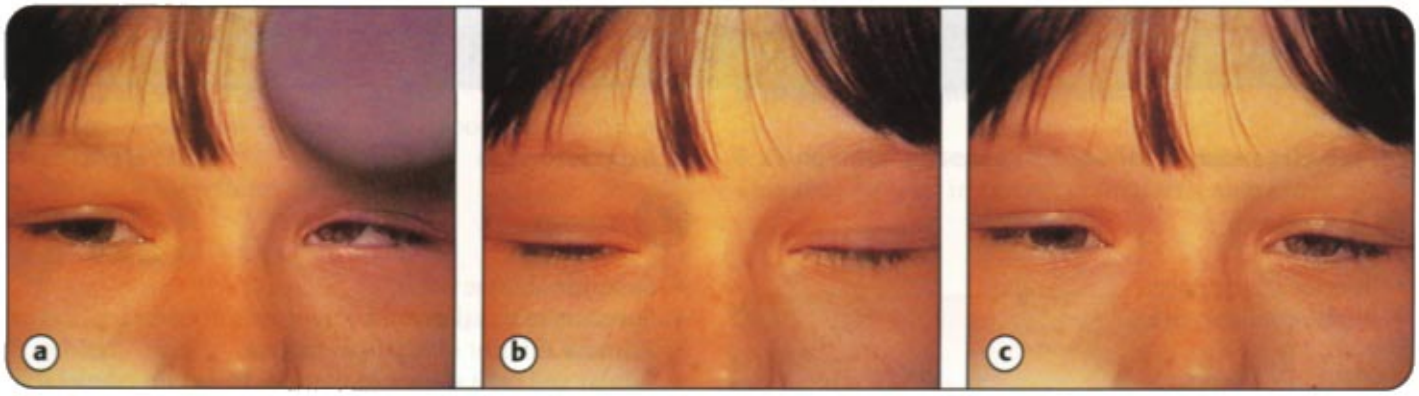

Fig. 13.38

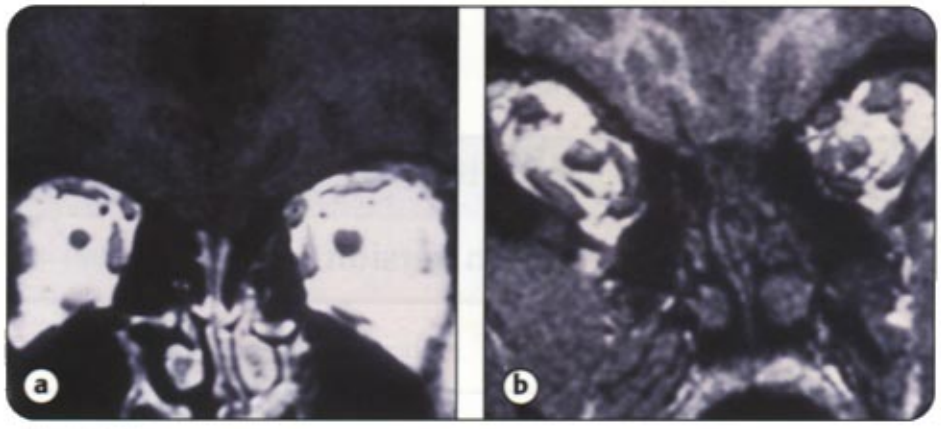

Fig. 13.39

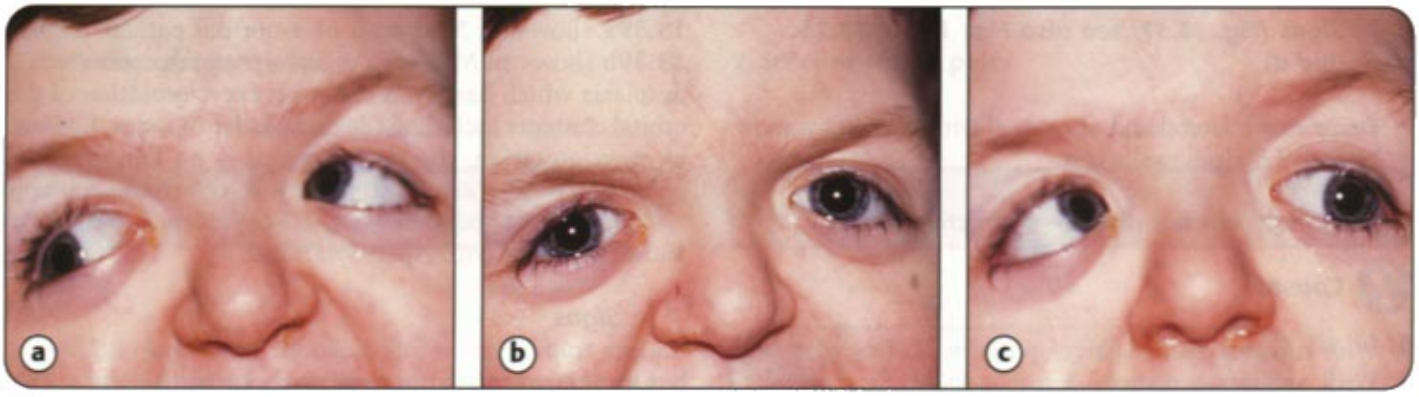

Fig. 13.40

In abduction on version testing

1. Dissociated vertical deviation
This may cause upshoot in abduction in contrast to an inferior oblique overaction.

2. Unilateral congenital ptosis

Signs

- Occasionally on upgaze the eye shoots up and laterally due to excessive innervation of the superior rectus–levator complex.
- The upshoot is abolished if the lid is physically elevated.

3. Ipsilateral inferior rectus palsy
With superior rectus overaction.

4. Contralateral inferior oblique palsy
With ipsilateral superior rectus overaction.

DOWNSHOOTS

1. In adduction on version testing

Causes

- Duane syndrome I, II, and III.
- Brown syndrome.
- Secondary superior oblique overaction which may occur as a result of ipsilateral inferior oblique palsy or contralateral inferior rectus palsy.
- Apparent superior oblique overaction. In some cases of longstanding superior oblique palsy there is contracture of the ipsilateral superior rectus. The extra innervation required by the contralateral synergist (superior oblique) in depression appears as an apparent overaction.

2. In abduction on version testing

Causes

- Excyclorotation of the orbit and contents is seen in certain craniofacial syndromes. If the globe is excyclorotated the lateral rectus acts not only as a pure abductor but also as a depressor (**Fig. 13.41a–c**).

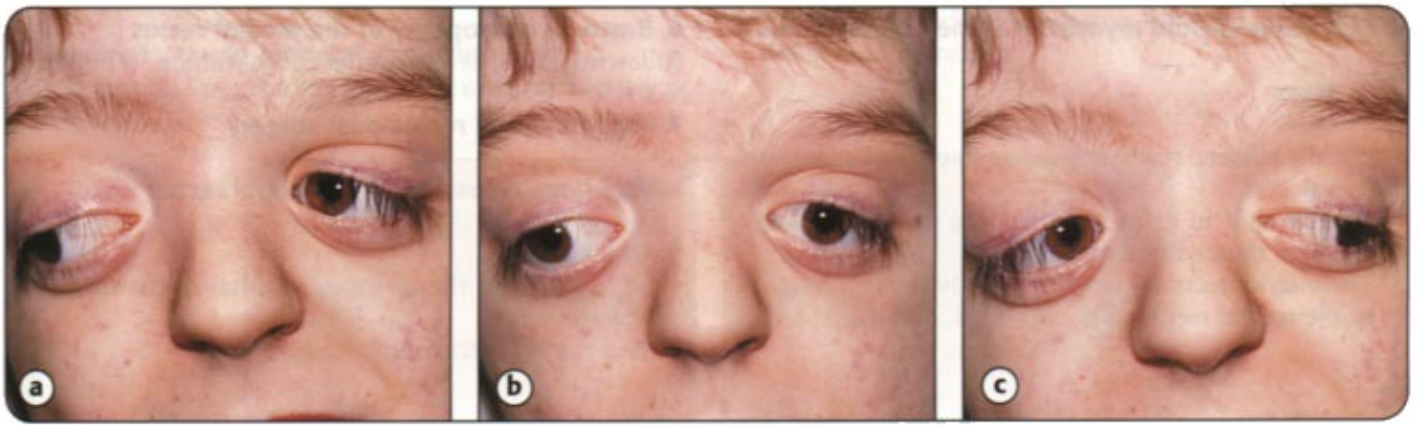

Fig. 13.41

LIMITATION OF HORIZONTAL EYE MOVEMENTS

Limitation of abduction on version testing

Unilateral

1. Acute onset sixth nerve palsy

Signs (right eye)

- Right esotropia in the primary position (**Fig. 13.42a**).
- Marked limitation of right abduction (**Fig. 13.42b**).
- Negative forced duction test.
- Reduced forced generation test.

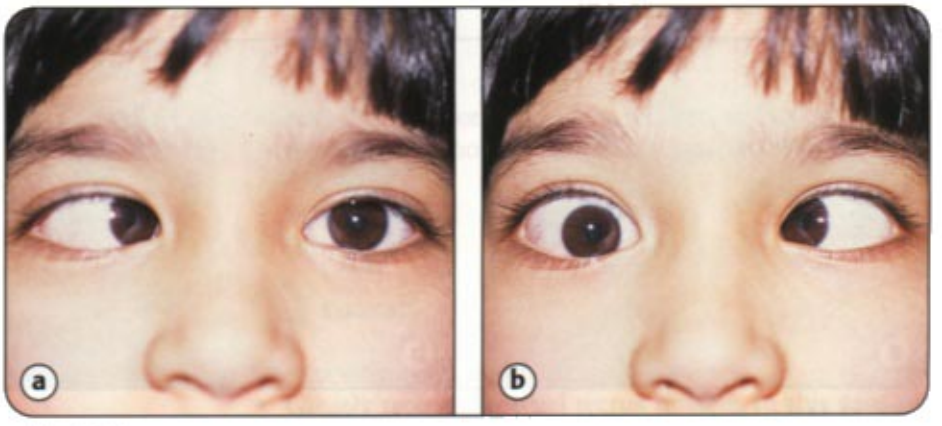

Fig. 13.42

2. Longstanding sixth nerve palsy

Signs (left eye)

- Normal left adduction (**Fig. 13.43a**).
- Straight eyes in primary position (**Fig. 13.43b**).
- Limitaton of left abduction (**Fig. 13.43c**).
- Positive forced duction test due to contracture of the ipsilateral medial rectus.
- Reduced forced generation test.

3. Following resection of the ipsilateral medial rectus

Signs

- Positive forced duction test.
- Normal forced generation test.

4. Restrictive thyroid myopathy of the medial rectus

Signs

- Usually bilateral but may be asymmetric and appear unilateral.
- Restriction is in a direction opposite that of the field of action of the involved muscle.
- The inferior rectus is most frequently affected, followed by the medial rectus, superior rectus, and lateral rectus.

Look for associated

- Lid retraction and lag.
- Chemosis.
- Superior limbic keratoconjunctivitis.
- Proptosis.

5. Duane syndromes I and II

Signs

- In a left Duane I syndrome there is slightly reduced left adduction and narrowing of the palpebral fissure (**Fig. 13.44a**).
- The eyes are straight in the primary position (**Fig. 13.44b**).
- Restriction of abduction with widening of palpebral fissure (**Fig. 13.44c**).

6. Restrictive myopathy of the medial rectus

Following myositis and orbital pseudotumor.

7. Slipped lateral rectus post surgery

8. Very large recession of the lateral rectus

9. Trauma

- Entrapment of the ipsilateral medial rectus in medial wall fracture.
- Very rarely, direct muscle laceration.

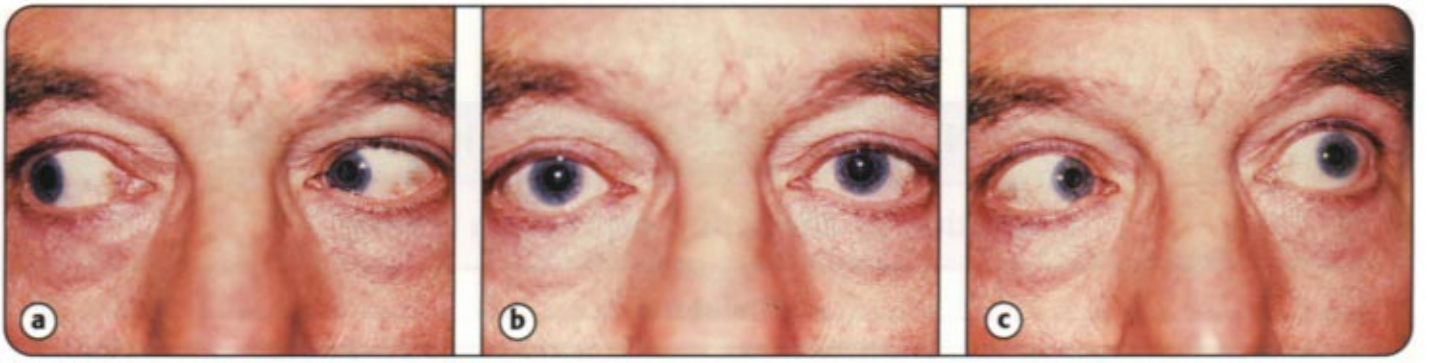

Fig. 13.43

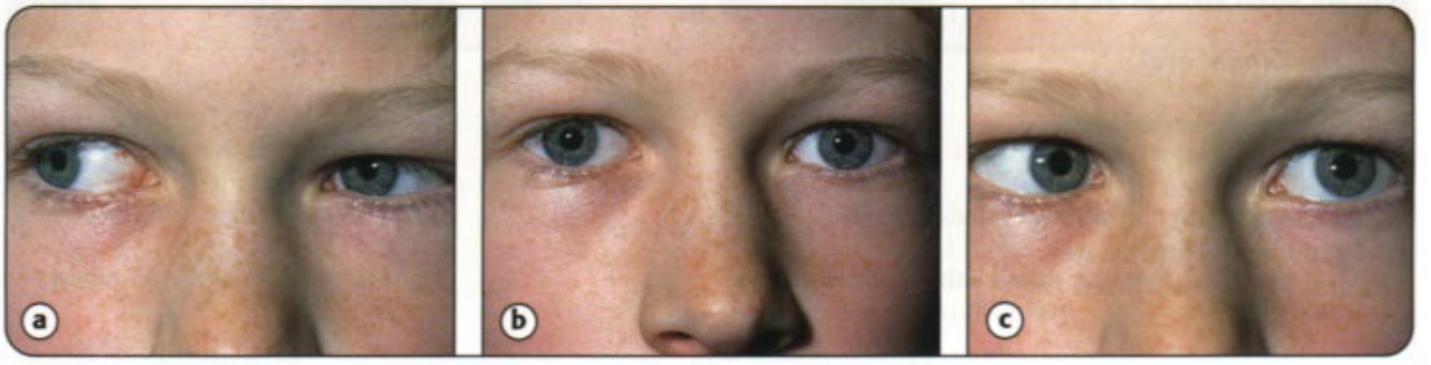

Fig. 13.44

Bilateral

1. Bilateral sixth nerve palsy

2. Restrictive thyroid myopathy
Affecting both medial recti.

3. Bilateral Duane syndromes I and III
Bilateral Duane syndrome I is shown in **Figure 13.45a and b.**

Signs

- On doll's-head maneuver, abduction is restricted.

4. Möbius syndrome

5. Acquired pseudo-bilateral abduction defect
This occurs on longstanding unilateral sixth nerve palsy in which the ipsilateral antagonist (medial rectus) requires less innervation to contract. According to Hering law, the contralateral synergist of the medial rectus (contralateral lateral rectus) will get less innervation and may appear to be underacting. Forced generation test will differentiate a true from an inhibitional palsy.

6. Congenital or infantile pseudo-bilateral abduction defect

Causes

- Infantile esotropia with cross-fixation – normal abduction on doll's-head maneuver.
- Nystagmus blockage syndrome – normal abduction on doll's-head maneuver.

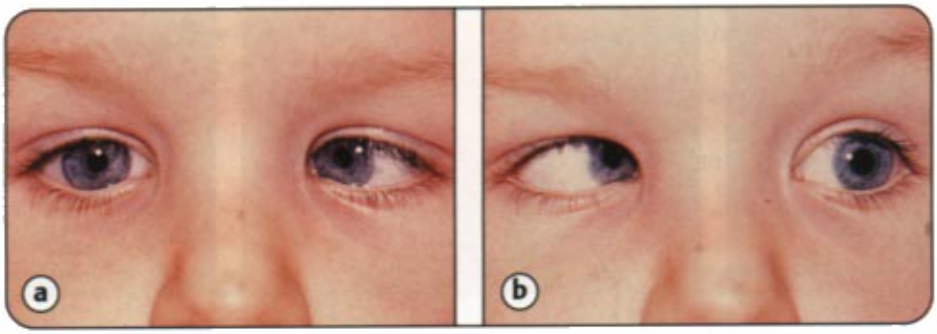

Fig. 13.45

Limitation of adduction on version testing

Unilateral

1. Acute third nerve palsy
This is shown in **Figure 13.20**.

Signs

- Complete – will affect pupillary parasympathetic supply (dilated pupil).
- Incomplete – will not affect pupil.
- Inferior division – involves the inferior rectus, medial rectus, inferior oblique, and pupillary parasympathetic supply (dilated pupil).
- Forced generation test will be reduced, but forced duction test will be normal.

2. Longstanding third nerve palsy

Signs

- Forced generation test will be reduced and forced duction test may be abnormal because of contraction of the lateral rectus.

3. Iatrogenic

Causes

- Slipped medial rectus muscle at surgery.
- Greatly recessed medial rectus muscle, in this case the left medial (**Fig. 13.46**).

Signs

- Left exotropia in primary position (**Fig. 13.46a**).
- Decrease in the deviation on levoversion (**Fig. 13.46b**).
- Defective left adduction on dextroversion (**Fig. 13.46c**).

4. Internuclear ophthalmoplegia (INO)

This is caused by a lesion of the medial longitudinal fasciculus.

Signs of bilateral INO

- Ipsilateral failure of adduction which is frequently asymmetric in bilateral cases (**Fig. 13.47a and b**).
- Nystagmus of abducting eye on conjugate gaze.
- Convergence is intact if the lesion is discrete (**Fig. 13.47c**).
- Vertical nystagmus on attempted upgaze.

These signs may be mimicked by myasthenia gravis and Fisher syndrome.

Table 13.1 Causes of internuclear ophthalmoplegia

- demyelination (usually bilateral)
- infarction
- hemorrhage
- tumors of the brainstem and 4th ventricle
- infection
- hydrocephalus
- progressive supranuclear palsy
- drug-induced
- remote carcinoma

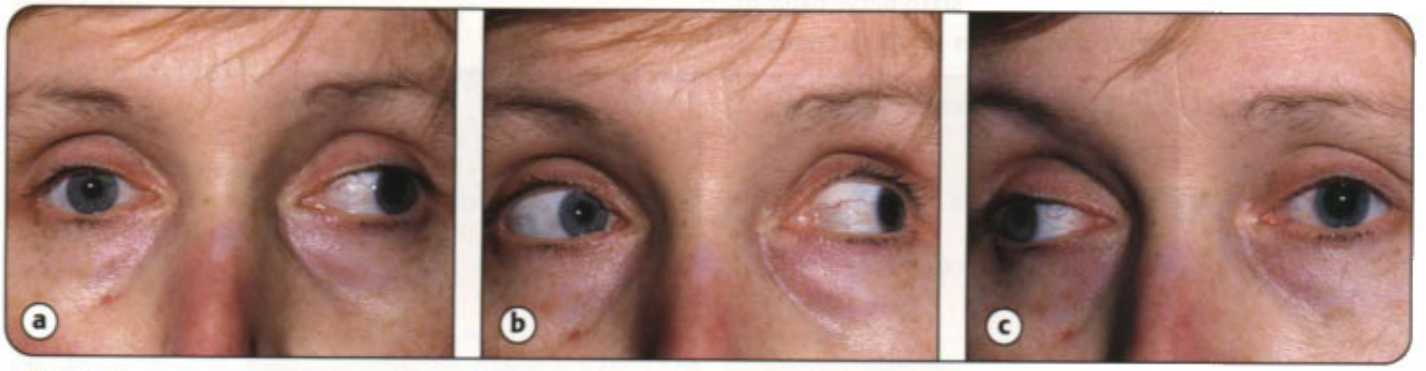

Fig. 13.46

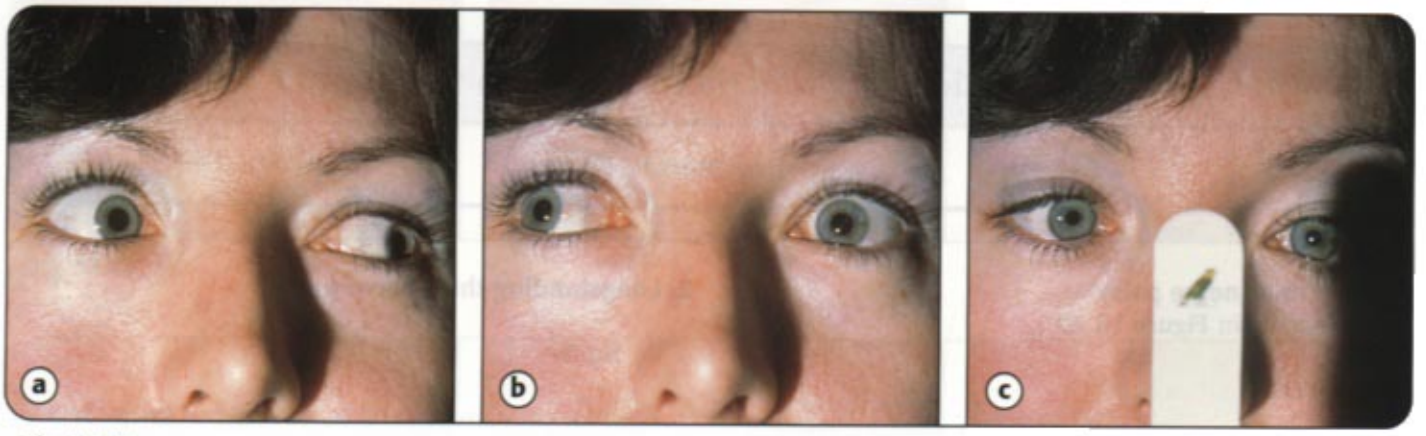

Fig. 13.47

5. Myasthenia gravis

Look for

- Variability of signs with fatiguability.
- Ptosis.
- Cogan lid twitch in upgaze.
- Involvement of other muscles – especially vertical.

6. Acute myositis
This may affect any extraocular muscle. Restriction of movement is in the direction of the field of action of the affected muscle.

Look for

- Pain.
- Chemosis.
- Proptosis.

7. Restrictive thyroid myopathy
Lateral rectus involvement is very rare and when it occurs other muscles are also involved.

8. Duane syndromes II and III

9. Congenital third nerve palsy

Bilateral

1. Bilateral third nerve palsy

2. Pseudo-bilateral third nerve palsy
This is the result of inhibitional palsy of the contralateral medial recti.

3. Bilateral internuclear ophthalmoplegia
Limitation of adduction is usually asymmetric (*see* **Fig. 13.47a and b**) with intact convergence (*see* **Fig. 13.47c**).

4. Myasthenia gravis

5. Bilateral Duane syndromes II and III

LIMITATION OF HORIZONTAL VERSIONS (GAZE PALSIES)

1. Acute unilateral lesion of premotor cortex (frontal eyefield)

Signs

- In destructive lesions (e.g. cerebrovascular accidents) there is tonic deviation of eyes to the ipsilateral side which usually resolves within a few weeks, with subsequent saccadic paralysis to the contralateral side.
- Rarely, in acute irritative lesions there is tonic deviation of the eyes to the contralateral side.

2. Unilateral destructive lesions of paramedian pontine reticular formation (PPRF)
These result in an ipsilateral gaze palsy.

3. One-and-a-half syndrome
This is caused by a lesion involving the PPRF (or abducens nucleus) and the adjacent medial longitudinal fasciculus.

Signs

- Ipsilateral gaze palsy.
- Ipsilateral internuclear ophthalmoplegia.
- The only residual movement is abduction of the contralateral eye, which exhibits abducting nystagmus.
- Exodeviation on distance fixation may be present.

Figure 13.48 show a rare case of a left one-and-a-half syndrome in a child.
Figure 13.48a shows gaze palsy on looking to the left.
Figure 13.48b – on attempted dextroversion the left eye does not move at all and the right eye abducts and exhibits nystagmus.

4. Bilateral pontine lesions
These result in total loss of horizontal gaze movements.

5. Fisher syndrome
This is a variant of Guillain–Barré syndrome.

Signs

- External ophthalmoplegia.
- Areflexia.
- Ataxia of gait and limbs.

This may initially resemble a horizontal gaze palsy or an internuclear ophthalmoplegia.

6. Ocular motor apraxia

This is a very rare condition.

Signs

- Saccadic paralysis of horizontal gaze.
- Head thrusts and blinking which allow refixation.
- Normal vertical gaze.

7. Wernicke encephalopathy

This is a triad of ophthalmoplegia, mental confusion, and gait ataxia. It is caused by thiamine deficiency and is mostly seen in alcoholics.

Initial signs

- Weakness of abduction.
- Internuclear ophthalmoplegia.
- Gaze-evoked nystagmus.
- Horizontal or vertical gaze palsies which may progress to ophthalmoplegia.

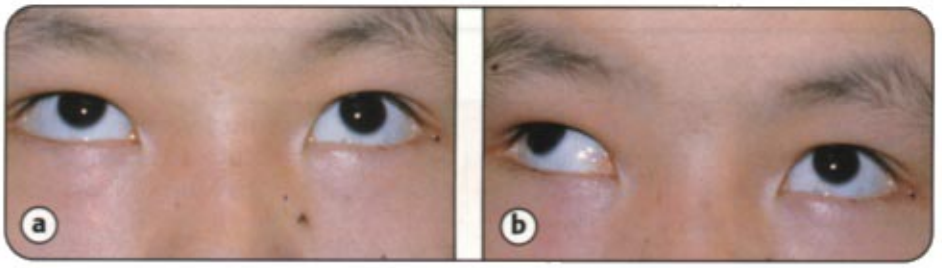

Fig. 13.48

LIMITATION OF VERTICAL EYE MOVEMENTS

1. Palsy of a vertically acting muscle (superior rectus, inferior rectus, superior oblique, or inferior oblique).

2. Restrictive thyroid myopathy of the inferior or superior rectus

3. Inferior rectus entrapment in an orbital blow-out fracture

Signs

- Limitation of left elevation (**Fig. 13.49a**).
- Normal depression (**Fig. 13.49b**).
- In some cases both elevation and depression may be restricted.

4. Inferior rectus contracture and fibrosis

This may occur following myositis.

5. Iatrogenic

- Retinal detachment surgery with placement of the explant under the superior or inferior rectus.
- Molteno tube in which the reservoir footplate is placed under the superior rectus.

6. Orbital space-occupying lesions

Signs

Figure 13.50 shows a patient with Paget's disease of the skull and a sarcoma in the superotemporal quadrant of the orbit which has caused restriction of left elevation.
Figure 13.51a shows a boy with a left plexiform neurofibroma of the anterior orbit.
Figure 13.51b shows slight restriction of left elevation.

7. Severe symblepharon

Figure 13.52 shows a child with congenital symblepharon which is restricting upgaze.

8. Brown syndrome

Signs

- Limitation of elevation in primary gaze or in adduction (**Fig. 13.53** shows right Brown syndrome).

9. Double elevator palsy

This may be congenital or acquired.

Signs (right eye)

- Limited elevation in all positions of upgaze (**Fig. 13.54a–c**).
- Normal abduction (**Fig. 13.54d**) and adduction (**Fig. 13.54f**).
- Straight in primary position (**Fig. 13.54e**). A slight ptosis may be present in some cases.
- Normal depression in all positions of downgaze (**Fig. 13.54g–i**).

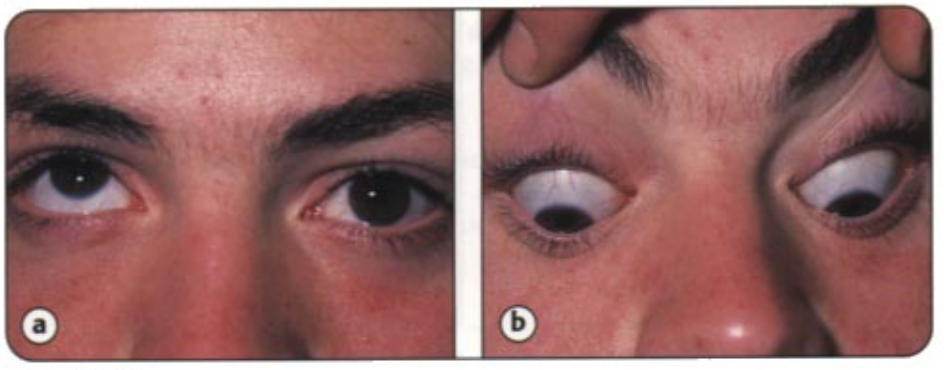

Fig. 13.49

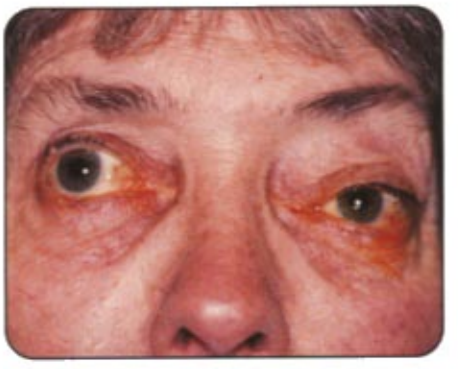

Fig. 13.50

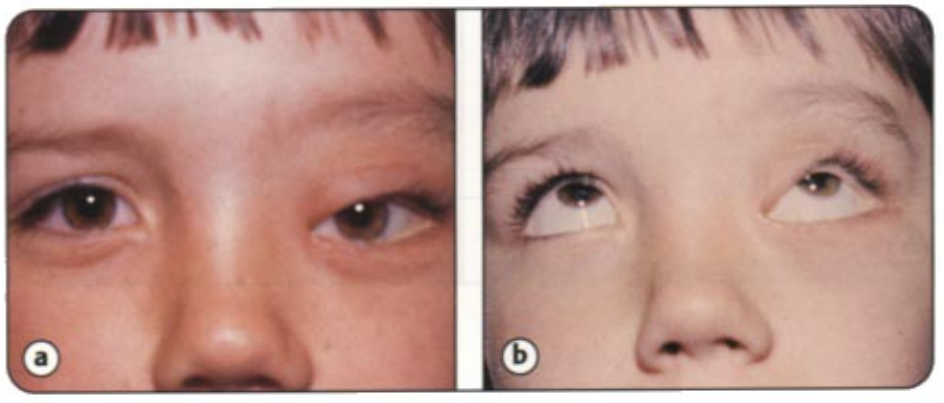

Fig. 13.51

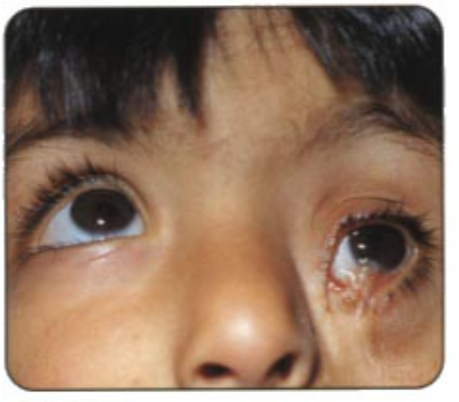

Fig. 13.52

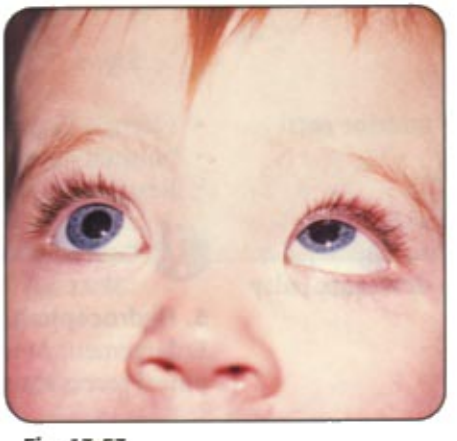

Fig. 13.53

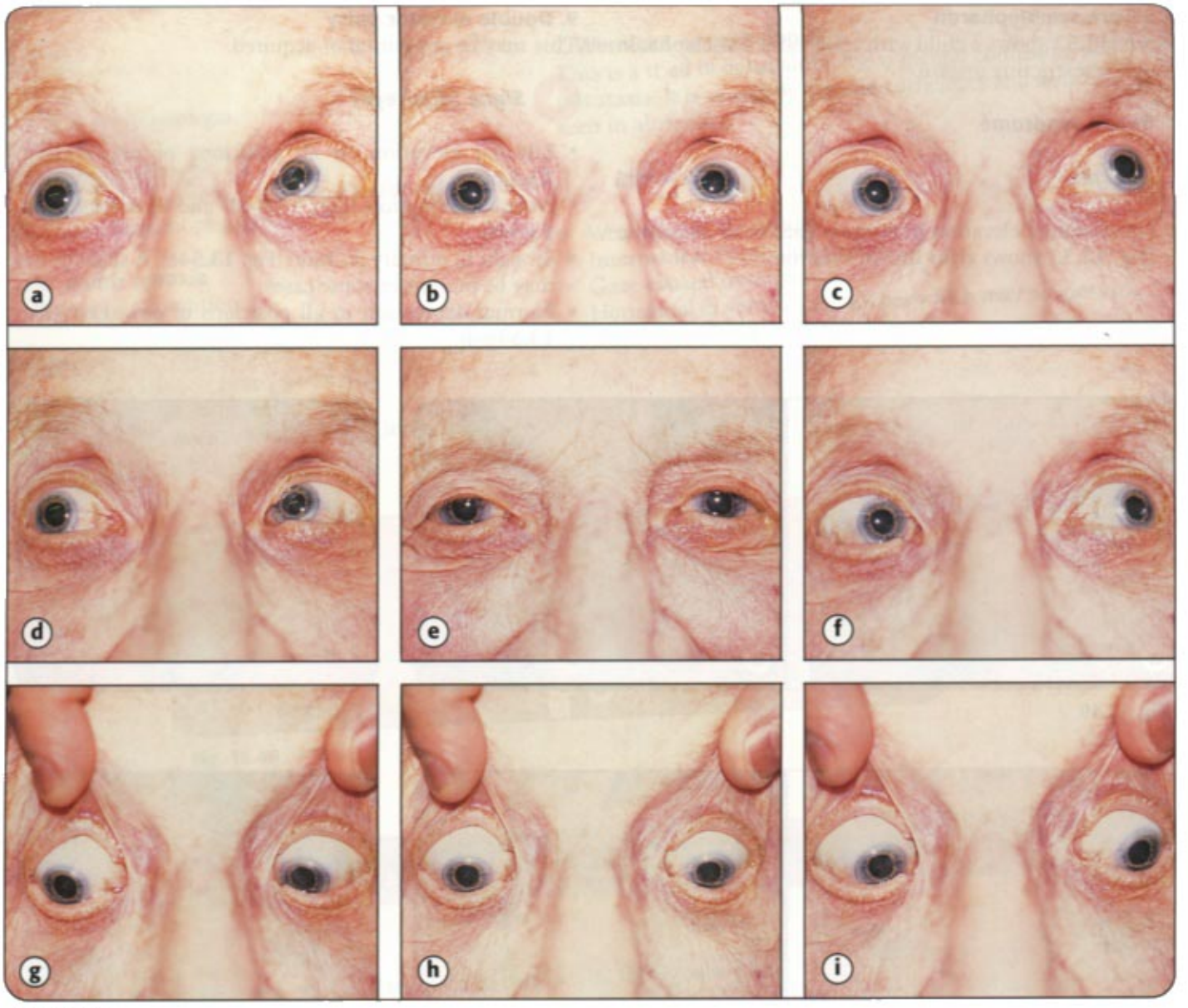

Fig. 13.54

LIMITATION OF VERTICAL VERSIONS (GAZE PALSIES)

1. Age-related limitation of upgaze

2. Restrictive thyroid myopathy of both inferior recti

3. Cerebrovascular accidents
- Midbrain infarction – complete vertical gaze palsy.
- Multi-infarct syndrome – incomplete vertical gaze palsy.
- Bilateral lesions of periaqueductal gray – downgaze palsy with upward tonic deviation of eyes.
- Thalamic hemorrhages or infarcts.

4. Parinaud syndrome

Signs

- Normal in primary position of gaze (**Fig. 13.55a**).
- Normal downgaze (**Fig. 13.55b**).
- Restriction of upgaze (**Fig. 13.55c**).

Causes – see Chapter 7.

5. Hydrocephalus

Enlargement of the aqueduct and third ventricle or suprapineal recess causes stretching of the posterior commisure.

Signs

- Loss of upgaze.
- Tonic downward deviation of eyes ('sun-set' sign) (**Fig. 13.56**).

6. Progressive supranuclear palsy

This is a degenerative disorder characterized by ophthalmoplegia, abnormal posture and tone, difficulty in swallowing, and mental slowing.

Signs

- Impairment of upgaze in all positions (**Fig. 13.57a–c**).
- Impairment of downgaze in all positions (**Fig. 13.57g–i**).
- Limitation of lateral gaze (**Fig. 13.57d and f**).
- Straight eyes in primary position (**Fig. 13.57e**).

7. Parkinson disease

This may resemble progressive supranuclear palsy as both are associated with limited upgaze, although limited downgaze is more characteristic of progressive supranuclear palsy.

8. Congenital fibrosis syndrome

This is a rare, familial condition.

Signs

- Downward fixed position of both eyes.
- Severe ptosis.
- Chin elevation.

9. Metabolic causes

- Tay–Sachs disease – impairment of vertical and, later, horizontal gaze.
- Adult-onset hexosaminidase deficiency – limited vertical gaze.
- Wernicke encephalopathy.
- Niemann–Pick variants.

10. Miscellaneous causes

- Oculogyric crisis – impaired downgaze with eyes deviated up and laterally.
- Whipple disease.
- Bilateral double elevator palsy.

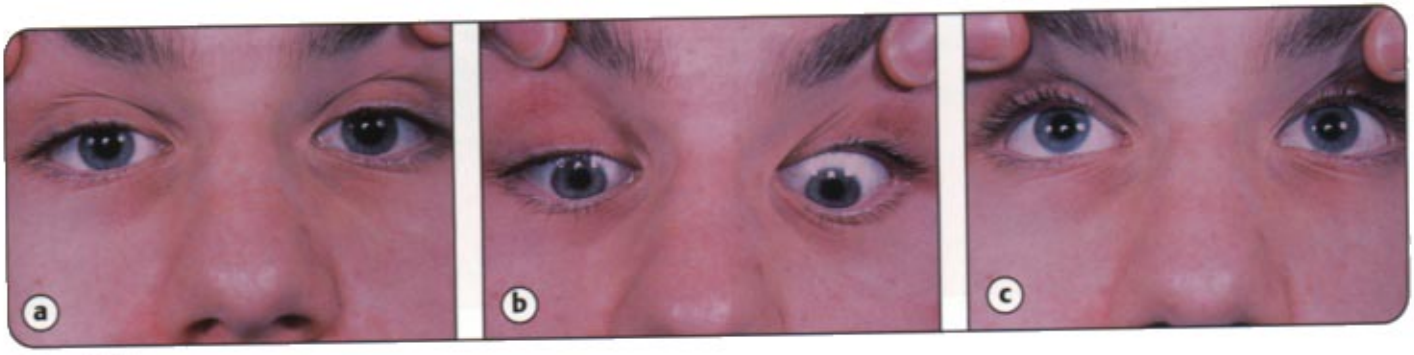

Fig. 13.55

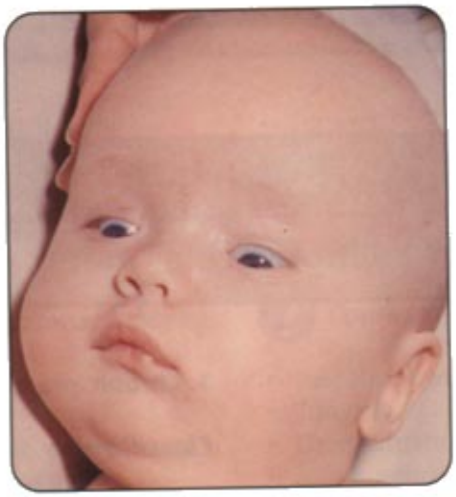

Fig. 13.56

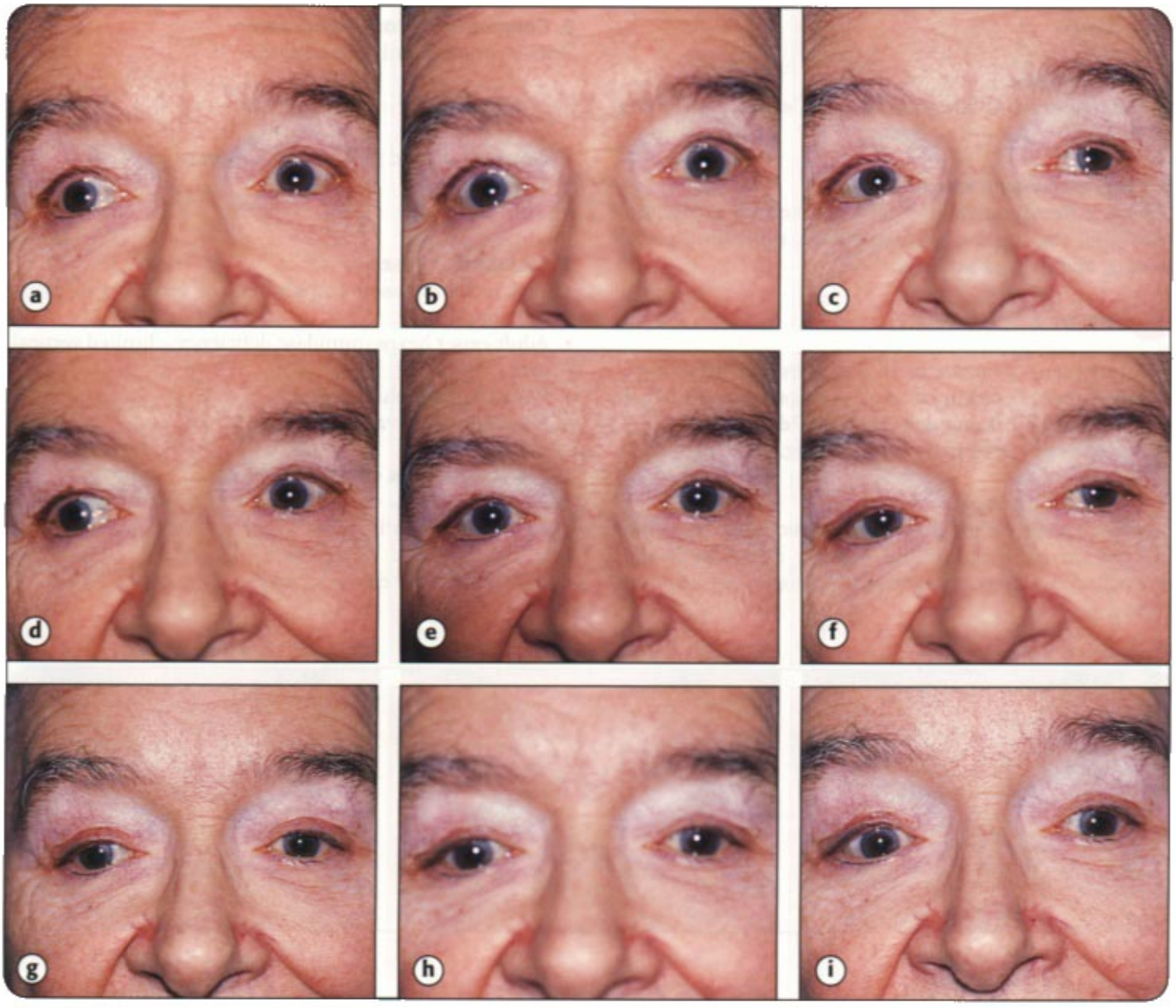

Fig. 13.57

GENERALIZED LIMITATION OF OCULAR MOTILITY

Without multiple ocular motor palsies

1. Frozen orbit

Causes

- Thyroid orbitopathy (**Fig. 13.58**).
- Pseudotumor.

2. Myasthenia gravis

3. Chronic progressive external ophthalmoplegia

Classification

- Primary isolated.
- Oculopharyngeal dystrophy – weakness of the pharyngeal muscles and wasting of temporalis.
- Kearns–Sayre syndrome – pigmentary retinopathy and heart block.

Signs

- Myopathic facies.
- Bilateral ptosis (**Fig. 13.59**).
- Initially, defective upgaze.
- Later, generalized symmetric impairment of movement.

4. Myotonic dystrophy

Look for

- Frontal balding.
- Ptosis.
- Myopathic mournful facies.
- Difficulty in relaxing grip.
- Presenile posterior stellate cataracts.
- Small pupils with light–near dissociation.
- Mesh-like peripheral retinal pigment clumping.

5. Drug-induced

- Phenytoin.
- Tricyclic antidepressants – may cause complete or internuclear ophthalmoplegia in patients with stupor.

6. Acquired ocular motor apraxia

This is associated with lesions of the frontoparietal cortex. Unlike in the congenital type, both horizontal and vertical gaze are affected.

7. Metabolic causes

- Wernicke encephalopathy.
- Tay–Sachs disease.
- Other lipid-storage diseases.

8. Late progressive supranuclear palsy

9. Congenital fibrosis syndrome

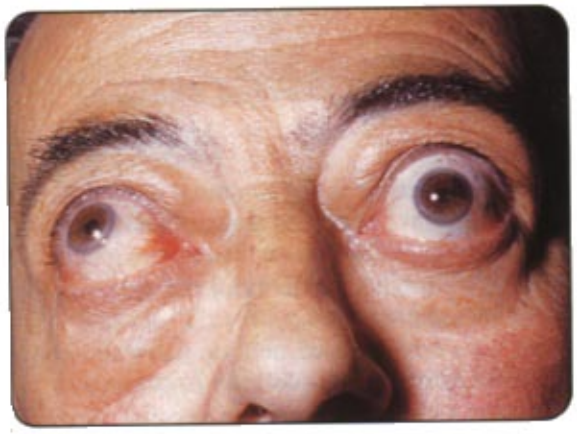

Fig. 13.58

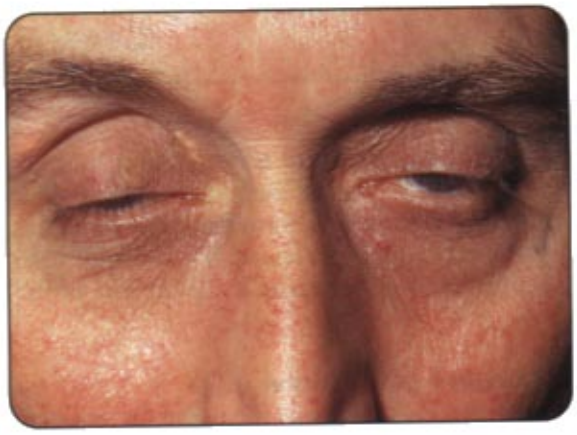

Fig. 13.59

With multiple ocular motor nerve palsies

1. Cavernous sinus and superior orbital fissure lesions

Causes

- Aneurysm.
- Tumors – meningioma, pituitary adenoma, nasopharyngeal carcinoma.
- Cavernous sinus thrombosis.
- Carotid–cavernous fistula.
- Tolosa–Hunt syndrome – an idiopathic granulomatous inflammation that affects the cavernous sinus.

2. Orbital lesions

Causes

- Infections – cellulitis and mucormycosis.
- Trauma.
- Metastases at orbital apex – such as from prostatic carcinoma.

3. Brainstem lesions

Causes

- Vascular lesions.
- Tumors.
- Demyelination.
- Motor neuron disease.
- Encephalitis, especially in AIDS.

PAINFUL OPHTHALMOPLEGIA

1. Orbital causes
- Acute cellulitis.
- Acute myositis.
- Pseudotumor.
- Direct carotid–cavernous fistula (*see* Chapter 2).

2. Posterior communicating aneurysm
This typically gives rise to an acute painful third nerve palsy with involvement of the pupil.

3. Herpes zoster ophthalmicus

4. Diabetes
This may also give rise to an acute painful ophthalmoplegia but the pupil is usually spared.

5. Giant-cell arteritis
This may occasionally give rise to an ophthalmoplegia associated with headache.

6. Superior orbital fissure syndrome (e.g. Tolosa–Hunt)

7. Cavernous sinus thrombosis (*see* Chapter 2)

COMPENSATORY HEAD POSTURES

General principles

Compensatory head postures are adopted to minimize diplopia by increasing the field of binocular single vision, most frequently in incomitant deviations. However, occasionally a patient may adopt an abnormal head posture to maximize diplopia so that it is easier to ignore one of the two images.

1. The head-turn and chin position
This is such that the face points in the same direction as the field of action of the affected muscle.

2. The head tilt
This is to the same direction as the torsional movement of the globe that would have occurred if the affected muscle were normal.

Another rule of thumb is that the head tilt occurs to the side with the hypotropic eye.

For example, in a left superior oblique palsy the compensatory head posture will be as follows:
- Head turn to the right and chin down because the field of action is down and to the right.
- Head tilt to the right because the muscle is an intorter (Also the right is the hypotropic eye).

Head turn

Strabismic causes

1. Lesions ipsilateral to direction of head turn
- Lateral rectus palsy.
- Duane syndrome I. **Figure 13.60a** shows left Duane syndrome with a head turn to the left. When the child's head is turned to the right the limitation of left abduction is seen (**Fig. 13.60b**).
- Duane syndrome III.
- Medial rectus contracture (e.g. following medial wall fracture).

2. Lesions contralateral to direction of head turn
- Third nerve palsy.
- Duane syndromes II and III.
- Lateral rectus fibrosis (e.g. following myositis).
- Medial rectus trauma.

3. Lesions nonspecific to direction of head turn
- Manifest congenital nystagmus to achieve null position.
- Infantile esotropia, usually associated with manifest latent nystagmus.
- DVD – very rarely.
- Spasmus nutans – head turn or tilt is seen in two-thirds of cases.

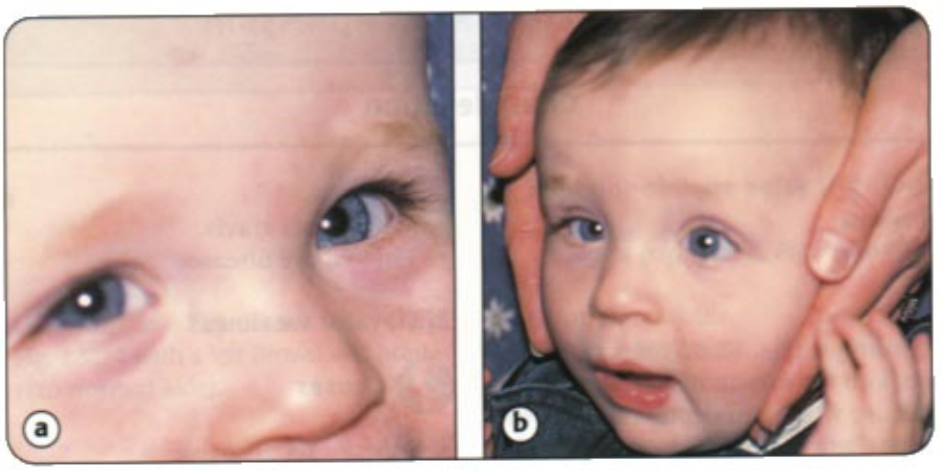

Fig. 13.60

Nonstrabismic causes

1. Congenital torticollis

Causes

- Anormal fusion of cervical vertebrae.
- Fibrosis of the sternocleidomastoid muscle. **Figure 13.61** shows a head turn to the left caused by ipsilateral involvement of the sternocleidomastoid muscle. This may also cause a head tilt.

2. Unilateral ametropia – usually astigmatic

3. Unilateral deafness

4. Nystagmus (*see* Chapter 14)

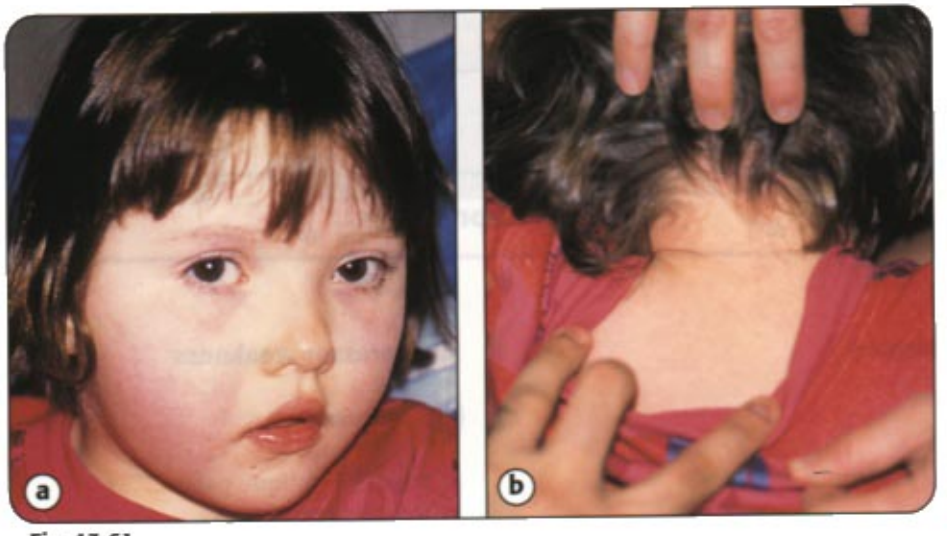

Fig. 13.61

Chin elevation and depression

Chin elevation

1. In horizontal strabismus

Look for

- 'A' esotropia and 'V' exotropia (**Fig. 13.62**). **Figure 13.62a** shows chin elevation with the eyes in relative downgaze. This takes place because with chin depression and the eyes in relative upgaze there is a marked exotropia (**Fig. 13.62b**) which is absent in downgaze (**Fig. 13.62c**).

2. Inability to look up on versions

Causes

- Congenital fibrosis syndrome.
- Chronic progressive external ophthalmoplegia.
- Myasthenia gravis.
- Thyroid eye disease.

3. Elevator weakness

Causes

- Superior rectus palsy.
- Entrapment of inferior rectus in orbital floor fracture.
- Inferior oblique palsy.
- Double elevator palsy.
- Brown syndrome.

4. Bilateral ptosis

5. Congenital nystagmus

With null position in downgaze.

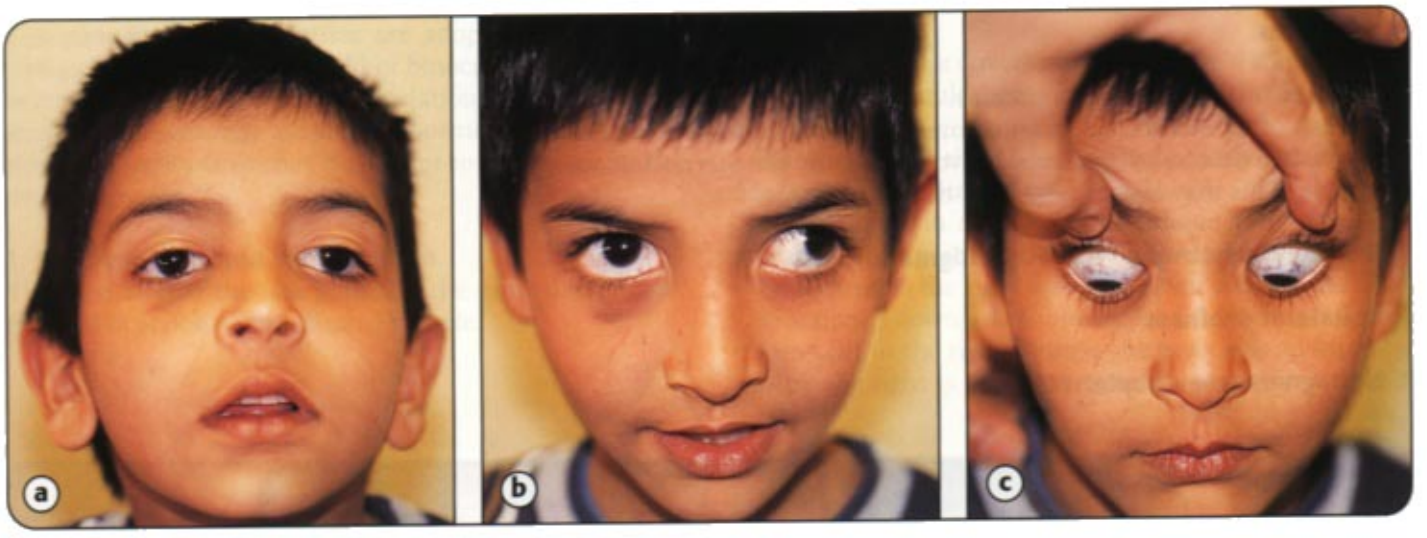

Fig. 13.62

Chin depression

1. In horizontal strabismus

Look for

- 'A' exotropia and 'V' esotropia.

2. Depressor weakness

Causes

- Superior oblique palsy.
- Inferior rectus palsy.

3. Congenital nystagmus

With null position in upgaze.

Head tilt

1. Brown syndrome

Signs

- Ipsilateral tilt (to affected side – **Fig. 13.63**. This is the same patient as in **Fig. 13.24** with a left Brown syndrome).
- Contralateral turn (to normal side).
- Chin elevated.

2. Inferior rectus palsy

Signs

- Tilt to either side.
- Ipsilateral turn.
- Chin depressed.

3. Inferior oblique palsy

Signs

- Ipsilateral tilt.
- Contralateral turn.
- Chin elevated.

4. Superior oblique palsy (Fig. 13.64a)

Signs (right eye, Fig. 13.64b)

- Contralateral tilt.
- Contralateral turn.
- Slight chin depression.

5. Superior rectus palsy

- Contralateral tilt usually, but may be to either side.
- Ipsilateral turn – if recent onset.
- Chin elevated – if recent onset.

6. Ocular tilt reaction

This is seen in skew deviation associated with ocular torsion and subjective deviation in the vertical plane. It may be tonic and either sustained or paroxysmal.

7. Central vestibular nystagmus

This may be suppressed by head tilt (e.g. upbeat nystagmus).

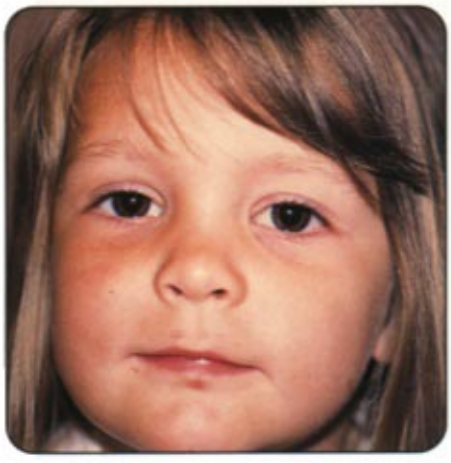

Fig. 13.63

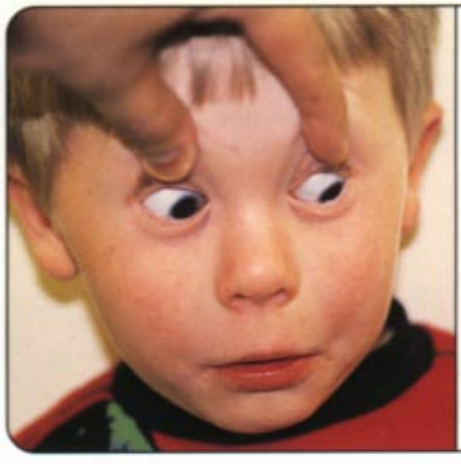

Fig. 13.64a

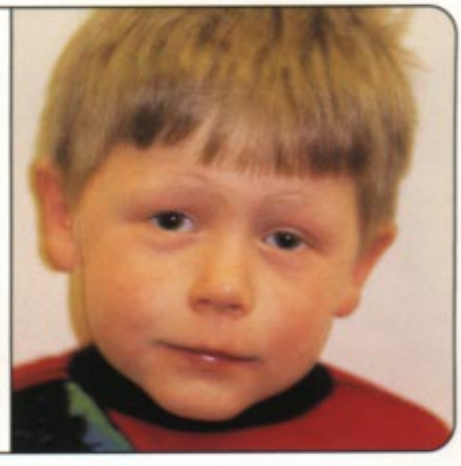

Fig. 13.64b

14 Nystagmus

CHARACTER OF OSCILLATIONS 420

PLANE OF OSCILLATIONS 420

Early-onset horizontal nystagmus
- Manifest nystagmus
- Latent nystagmus
- Manifest–latent nystagmus

Later-onset horizontal nystagmus
- Fine horizontal jerk nystagmus
- Coarse horizontal jerk nystagmus

Torsional nystagmus

Vertical nystagmus
- Downbeat nystagmus
- Upbeat nystagmus

Nystagmus with no specific plane of oscillation
- Nystagmus only present in eccentric gaze
- Reversal of nystagmus direction
- Seesaw nystagmus

MISCELLANEOUS TYPES OF NYSTAGMUS 424

- Dissociated nystagmus
- Disjunctive nystagmus
- Roving nystagmus
- Nystagmoid movements

CHARACTER OF OSCILLATIONS

Nystagmus is a repetitive, to and fro oscillation of the eyes. The three main types are:

1. Jerk nystagmus

Signs

- There is a slow drift and then a fast 'corrective' phase.
- The direction of nystagmus is described in terms of the direction of the fast component.

2. Pendular nystagmus

Signs

- The velocity of nystagmus is equal in both directions.
- Congenital pendular nystagmus is horizontal, conjugate, and tends to convert to jerk on lateral gaze.
- Acquired pendular nystagmus has horizontal, vertical, and torsional components.
- If the horizontal and vertical components of pendular nystagmus are in phase the perceived direction becomes oblique.
- If the horizontal and vertical components are out of phase the direction becomes ellipitic or rotary.

3. Mixed (jerk and pendular) nystagmus

PLANE OF OSCILLATIONS

The plane of nystagmus may be horizontal, vertical, torsional, or nonspecific. Congenital nystagmus displays uniplanar oscillations even though the character may vary, and therefore congenital horizontal pendular nystagmus may become jerk on lateral gaze but with its plane remaining horizontal. Acquired nystagmus may be uniplanar or multiplanar.

Early-onset horizontal nystagmus

Manifest nystagmus

Manifest nystagmus is very rarely present at birth and usually develops a few weeks later.

Signs

- Usually conjugate (equal amplitude in both eyes) and horizontal, although it may be difficult to discern whether it is jerk or pendular.
- Rarely, it may be purely vertical or torsional.

Table 14.1 Causes of early-onset nystagmus

Idiopathic motor	Associated with less obvious bilateral ocular disease
Hereditary	• ocular albinism with foveal hypoplasia
Associated with obvious bilateral ocular disease	• optic nerve hypoplasia
• corneal opacities	• optic atrophy
• microcoria	• achromatopsia
• cataract	• Leber congenital amaurosis
• buphthalmos	• X-linked congenital stationary night-blindness
• microphthalmos	• X-linked congenital retinoschisis
• oculocutaneous albinism	

Latent nystagmus

Latent nystagmus is associated with infantile esotropia and dissociated vertical deviation (DVD).

Signs

- Horizontal jerk nystagmus which becomes apparent on covering one eye or reducing the amount of light reaching the eye.
- The fast phase is in the direction of the uncovered fixating eye.
- Nystagmus disappears when both eyes are open.

Manifest–latent nystagmus

Manifest–latent nystagmus is also associated with infantile esotropia and DVD.

Signs

- Increase in amplitude when one eye is covered.
- With both eyes open, the amplitude is reduced but manifest nystagmus remains.

Later-onset horizontal nystagmus

Fine horizontal jerk nystagmus

Fine horizontal jerk nystagmus is usually caused by peripheral vestibular disease. However, unilateral labyrinthine damage may also display a torsional component.

Signs

- Transient nystagmus with the fast phase contralateral to the side of the lesion.
- Amplitude increases to the side away from the lesion.
- Nystagmus is accentuated or may only become apparent if fixation is prevented, as occurs when using a high-power convex lens.

Coarse horizontal jerk nystagmus

Coarse horizontal jerk nystagmus is usually caused by cerebellar disease, which may also cause vertical nystagmus.

Signs

- Persistent nystagmus with the fast phase toward the side of the lesion.
- Amplitude increases when the eyes move toward the side of the lesion.

Torsional nystagmus

Purely torsional nystagmus is only seen in central nervous system disease including the central vestibular system.

Causes

- Syringobulbia.
- Syringomyelia with Arnold–Chiari malformation.
- Demyelination.
- Lateral medullary syndrome.

Vertical nystagmus

Downbeat nystagmus

Signs

- Vertical nystagmus with fast phase downward.
- More easily elicited with the patient looking down and laterally.

Table 14.2 Causes of downbeat nystagmus

Lesions at craniocervical junction	Miscellaneous
• Arnold–Chiari malformation	• trauma
• cerebellar degeneration	• vitamin B_{12} deficiency
• syringobulbia	• Wernicke encephalopathy
• Paget disease	• hydrocephalus
Drugs	• demyelination
• lithium toxicity	
• phenytoin	
• carbamazepine	
• barbiturates	

Upbeat nystagmus

Signs

- Vertical nystagmus with fast phase upward.

Table 14.3 Causes of upbeat nystagmus

• cerebellar degeneration
• vascular disease of the cerebellum or medulla
• encephalitis
Drugs
• organophosphates
• tobacco
Miscellaneous
• Wernicke encephalopathy
• Leber congenital amaurosis

Nystagmus with no specific plane of oscillation

The following types of nystagmus are distinctive because of specific features. The plane of oscillation may be horizontal, vertical, torsional, or multidirectional.

Nystagmus only present in eccentric gaze

1. Physiologic end-point nystagmus

2. Gaze-evoked nystagmus

Causes

- Lesions of the vestibulocerebellum (flocculonodular lobe), brainstem, and cerebral hemispheres.
- Gaze palsy – fast phase in direction of defective gaze.
- Drugs – lithium, phenytoin, carbamazepine, and barbiturates.
- Nystagmus associated with peripheral vestibular disease usually apparent only in eccentric positions of gaze but becoming apparent in all positions of gaze if fixation is interrupted with high-power convex lenses.

Reversal of nystagmus direction

1. Acquired periodic alternating nystagmus

Signs

The nystagmus reverses every 2 minutes. As the first half of the cycle finishes, there may be a transitional period during which there may be upbeating, downbeating, or square wave jerks before the second, reversed phase of the cycle begins.

Causes

- Arnold–Chiari malformation.
- Cerebellar disease.
- Demyelination.
- Ataxia telangiectasia.
- Trauma.
- Drugs such as phenytoin.
- Associated with sudden visual loss.

2. Congenital periodic alternating nystagmus

This is not as regular as the acquired type.

3. Rebound nystagmus

This is usually seen in patients with cerebellar disease but may occasionally occur in normal individuals.

Signs

- Continued effort to maintain eccentric gaze in gaze-evoked nystagmus may result in dampening and sometimes reversal of direction of nystagmus.
- On returning to the primary position of gaze, a transient nystagmus develops, with the fast phase opposite to the original gaze-evoked nystagmus.

Seesaw nystagmus

Signs

- Pendular nystagmus in which one eye elevates and intorts while the other eye depresses and extorts. The eyes then reverse direction.

Causes

The lesion is located in the diencephalon but specific causes include:

- Large parasellar tumors (most common).
- Head trauma.
- Syringobulbia.
- Brainstem stroke.
- Transient finding in albinism – later changes to horizontal.
- Congenital – very rare.

In dissociated nystagmus the oscillations in the two eyes have different amplitudes.

MISCELLANEOUS TYPES OF NYSTAGMUS

Dissociated nystagmus

1. End-point physiologic nystagmus
Nystagmus may be slightly greater in the abducting eye than in the adducting eye.

2. Latent nystagmus
This is occasionally disconjugate, with greater amplitude of nystagmus in the viewing eye.

3. Internuclear ophthalmoplegia
There is marked nystagmus of the abducting eye with little or no nystagmus of the adducting eye.

4. Brun nystagmus
This is caused by a cerebellopontine angle tumor, usually an acoustic neuroma.

Signs

- Coarse gaze-evoked nystagmus on ipsilateral gaze (cerebellar damage).
- Fine nystagmus on contralateral gaze (vestibular nerve damage).

5. Spasmus nutans
Onset is between 3 and 18 months.

Signs

- Unilateral or bilateral, small-amplitude, high-frequency horizontal nystagmus associated with head nodding.
- Frequently asymmetric with increased amplitude in adbuction, but sometimes strictly monocular.
- May have vertical and torsional components.
- Over the course of a few minutes the nystagmus may be conjugate, disconjugate, disjunctive, and even monocular.

Causes

- Idiopathic, with spontaneous resolution by age 3 years.
- Gliomas of the optic nerve or chiasm.
- Empty sella syndrome.
- Porencephalic cyst.

6. Monocular nystagmus (Table 14.4)

Table 14.4 Causes of monocular nystagmus

Spasmus nutans
Deep amblyopia, usually ametropic (Heimann–Bielschowsky phenomenon)
Superior oblique myokymia – rapid, monocular, torsional–vertical oscillations
• idiopathic • longstanding superior oblique palsy • associated with dyemyelination
Pendular nystagmus
• monocular blindness • demyelination • vascular brainstem disease
Vertical pendular – optic nerve tumor must be excluded
Voluntary

Disjunctive nystagmus

1. Convergence retraction nystagmus
This is typically seen in Parinaud syndrome and rarely in Arnold–Chiari malformation.

Signs of Parinaud syndrome

- Retraction–convergence nystagmus.
- Bilateral large pupils.
- Light–near dissociation of pupillary reactions.
- Paralysis of upgaze and convergence.

2. Divergence retraction nystagmus
This is occasionally seen in Parinaud syndrome.

Roving nystagmus

Any severe disruption of vision during infancy may lead to nystagmus. The two most common causes are.

- Leber congenital amaurosis.
- Bilateral optic nerve hypoplasia – exclude intracranial anomalies such as absent septum pellucidum in De Morsier syndrome.

Nystagmoid movements

1. Ocular flutter and opsoclonus

Signs

- Saccadic oscillations with no intersaccadic interval. In ocular flutter they are purely horizontal and in opsoclonus they are multiplanar.

Causes

- Viral encephalitis.
- Metastatic neuroblastoma in children.
- Paraneoplastic syndrome in adults (bronchial carcinoma).
- Demyelination.
- Drugs – lithium, amitriptyline, phenytoin, diazepam, phenelzine, and imipramine.
- Idiopathic and transient in healthy neonates.
- Monoclonic encephalopathy in infants ('dancing eyes and dancing feet').
- Toxins – organophosphates.

2. Ocular bobbing

Signs

- Rapid, conjugate, downward eye movements with a slow return to the primary position.

Causes

- Intrinsic pontine lesions – usually hemorrhage.
- Cerebellar lesions compressing the pons.
- Metabolic encephalopathy.

Appendix – Uncommon systemic disorders

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ADRENOLEUKODYSTROPHY

This is a peroxisomal disorder characterized by progressive central nervous system (CNS) demyelination and adrenocortical deficiency due to an inability to metabolize saturated, unbranched, very-long-chain fatty acids. There are two types: the neonatal type and the more common childhood type.

1. Neonatal

Mode of inheritance

- Autosomal recessive.

Ocular features

- Poorly reactive pupils.
- Optic atrophy with early loss of vision.
- Pigmentary retinopathy.
- Nystagmus.
- Anterior polar cataracts.

Systemic features

- Onset before 1 year of age.
- Delayed development.
- Neurologic defects often present at birth, with deterioration leading to death in early childhood.
- Seizures.
- Adrenal insufficiency.
- Dysmorphic features.

2. Childhood

Mode of inheritance

- X-linked recessive.

Ocular features

- Optic atrophy with visual loss.
- Nystagmus.

Systemic features

- Onset between 4 and 10 years of age.
- Behavioral disturbance and dementia.
- Impaired hearing.
- Mild to severe adrenal insufficiency.
- Individuals die within 1 to 4 years of onset.

ADULT-ONSET HEXOSAMINIDASE DEFICIENCY

– *see* Tay–Sachs disease

AICARDI SYNDROME

Mode of inheritance

- X-linked dominant (lethal in males).

Ocular features

- Bilateral chorioretinal lesions consisting of sharply demarcated, yellow–white lacunae.
- Optic disc coloboma.
- Uveal coloboma.
- Microphthalmos.

Systemic features

- Infantile spasms.
- Agenesis of corpus callosum.
- Delayed development.
- Mental deficiency.
- Hypotonia.

Comment

- Infantile spasms are usually the presenting feature in the first 2 years of life. Death occurs in the majority of patients before early adulthood.

ALAGILLE SYNDROME
(arteriohepatic dysplasia)

Mode of inheritance

- Autosomal dominant mapped to 20p.

Ocular features

- Posterior embryotoxon.
- Optic disc drusen.
- Pigmentary retinopathy.

Systemic features

- Chronic cholestasis due to paucity of intrahepatic bile ducts.
- Peripheral pulmonary stenosis or hypoplasia.
- Peculiar facies.
- Complex cardiovascular abnormalities.
- Butterfly vertebral arch defects.

Comment

- Most patients present with neonatal jaundice and about 25% eventually die of the disease.

ALKAPTONURIA

Mode of inheritance

- Autosomal recessive defect of homogentisic acid oxidase, resulting in excessive deposition of homogentisic acid in tissues.

Ocular features

- Bluish-gray or black pigmentation (ochronosis) of the conjunctiva, episclera, sclera, and tendons of the horizontal recti.

Systemic features

- Ochronosis of cartilage.
- Arthritis.
- Dark sweat stains on clothes.

ALPORT SYNDROME

Mode of inheritance

- X-linked affecting basement membrane collagen.

Ocular features

- Anterior lenticonus.
- Cataract.
- Microspherophakia.
- Flecked retinopathy.
- Posterior polymorphous dystrophy.

Systemic features

- Progressive hemorrhagic nephritis.
- Bilateral sensorineural deafness.

Comment

- Hematuria usually begins in childhood, whereas renal failure and secondary hypertension occur later.

ALSTRÖM SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Pigmentary retinopathy (cone–rod dystrophy).
- Optic atrophy.
- Nystagmus.
- Photophobia.

Systemic features

- Diabetes mellitus.
- Obesity.
- Deafness.
- Acanthosis nigricans.
- Hypogenitalism.
- Dilated cardiomyopathy.

AMYLOIDOSIS

This is either a localized or systemic condition in which there is extracellular deposition of a fibrillar protein. There are six types.

1. Hereditary systemic

- Portuguese type I (neuropathic amyloidosis).
- Meretoja syndrome (lattice corneal dystrophy type IV).
- Familial Mediterranean fever.

2. Primary systemic

Ocular features

- Prominent corneal nerves.
- Vitreous opacities.
- Light–near dissociation of pupillary reactions.

Systemic features

- Polyneuropathy.

3. Immunocyte-related systemic

This type is associated with lymphoproliferative disease of B-cell origin.

Systemic features

- Macroglossia (only seen in this form).
- Other organ involvement, e.g. heart – cardiac failure; kidney – nephrotic syndrome; nerves – peripheral neuropathy.

4. Reactive systemic

This type is associated with longstanding chronic infections (e.g. TB), inflammation (e.g. rheumatoid arthritis), and malignancy.

Systemic features

- Hepatosplenomegaly.

5. Local

This type may affect any organ.

- Primary, e.g. lattice corneal dystrophy type I.
- Secondary.

6. Age-related

ANGELMAN SYNDROME

Mode of inheritance

- All cases are sporadic and 60–80% have interstitial deletion of 15q.

Ocular features

- Strabismus.
- Pale-blue eyes.
- Deep-set eyes.

Systemic features

- Severe mental retardation.
- Speech deficiency.
- Ataxia and jerky arm movements resembling a puppet gait.
- Bursts of inappropriate laughter.

ANTIPHOSPHOLIPID ANTIBODY SYNDROME

There are two antiphospholipid antibodies: lupus anticoagulant and anticardiolipin antibody.

Ocular features

- Cotton-wool spots.
- Retinal vascular thromboses.
- Retinal venous dilatation.

Systemic features

- Thrombocytopenia.
- Vascular thromboses resulting in strokes in young patients and recurrent spontaneous abortions.
- Valvular heart disease.
- Atheroma.
- Skin changes, e.g. ulceration, livedo reticularis.

Comment

- Look for anticardiolipin antibody, which is diagnostic. Antinuclear antibody is usually negative.

APERT SYNDROME

Mode of inheritance

- Autosomal dominant, but the vast majority of cases are sporadic and associated with older paternal age. There is a mutation in the FGFR2 (fibroblast growth factor receptor) gene on 10q.

Ocular features

- Shallow orbits and proptosis.
- Hypertelorism.
- Strabismus.
- Megalocornea.
- Keratoconus.
- Ectopia lentis.
- Congenital glaucoma.
- Downslanting palpebral fissures.

Systemic features

- Irregular craniosynostosis, especially of the coronal suture.
- Osseous or cutaneous syndactyly.
- Midfacial hypoplasia.
- Broad distal phalanx of thumb and big toe.
- Narrow palate with median groove.
- Mental handicap.
- Moderate to severe acne.

Comment

- Mental handicap may occur despite early surgery for craniosynostosis.

ARTERIOHEPATIC DYSPLASIA

– see Alagille syndrome

ATAXIA TELANGIECTASIA

(Louis-Bar syndrome)

Mode of inheritance

- Autosomal recessive, with gene locus identified on 11q.

Ocular features

- Bulbar conjunctival telangiectasia (usually appears between 2 and 8 years of life).
- Eye movement disorders.

Systemic features

- Progressive ataxia.
- Skin telangiectasia.
- Lymphopenia and immune deficit.
- Growth deficiency (more obvious in late childhood).
- Mental deficiency.
- Hematopoietic malignancy.

Comment

- Most patients die by the third decade.

BARDET–BIEDL SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Pigmentary retinopathy in all cases, but typical retinitis pigmentosa in less than 10%.
- Myopia.
- Cataract.
- Bull's-eye maculopathy.
- Astigmatism.

Systemic features

- Polydactyly.
- Obesity.
- Hypogenitalism.
- Renal disease.
- Mental handicap.

Comment

- Although only 15% of patients show pigmentary retinopathy by 10 years of age, almost 75% of patients are blind by the age of 20 years. Mental deficiency is mild to moderate. No affected males have fathered a child, but females have reproduced.

BASSEN–KORNZWEIG SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Pigmentary retinopathy secondary to vitamin A deficiency (atypical retinal pigmentosa, with disc pallor, attenuated vessels, and white spots).
- Ptosis.
- Progressive external ophthalmoplegia.
- Xanthelasma.
- Arcus.

Systemic features

- Abetalipoproteinemia.
- Fat malabsorption leading to secondary vitamin A deficiency.
- Ataxia.
- Acanthocytosis.

Comment

- Plasma levels of cholesterol, phospholipids, and triglycerides are very low. High doses of vitamin A may reverse visual impairment.

BATTEN–VOGT SYNDROME
(neuronal ceroid lipofuscinosis)

Mode of inheritance

- Autosomal recessive with accumulation of lipopigments in tissues. There are four types:

1. Infantile (Haltia–Santavuori)

Ocular features/Systemic features

- Onset before 2 years of age, with rapid psychomotor deterioration and blindness.

2. Late infantile (Jansky–Bielschowsky)

Ocular features

- Variable pigmentary retinopathy.
- Slowly progressive optic disc pallor.
- Attenuation of retinal vessels.
- Bull's-eye maculopathy in some cases.

Systemic features

- Onset at 2–4 years of age, with rapid CNS deterioration.

3. Juvenile (Spielmeyer–Sjögren)

Ocular features

- Severe retinal changes with bull's-eye maculopathy.

Systemic features

- Onset at 6–8 years of age.
- CNS deterioration.

4. Adult (Kufs disease)

The adult type is similar to the juvenile type but has a late onset.

BEHR SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Childhood-onset optic atrophy.

Systemic features

- Ataxia.
- Spasticity.
- Mental retardation.

BLEPHAROPHIMOSIS SYNDROME

Mode of inheritance

- Autosomal dominant condition associated with interstitial deletion on 3q. There are two types: type I is associated with infertility in females and complete penetrance, and type II is associated with incomplete penetrance.

Ocular features

- Epicanthus inversus.
- Ptosis.
- Telecanthus.
- Short horizontal palpebral fissure.
- Lateral ectropion.
- Hypoplasia of the superior orbital rim.
- Amblyopia.

Systemic features

- Menstrual irregularity and infertility (type I).
- Variable hypotonia.
- Incomplete development of the ears.
- Poorly developed nasal bridge.

Comment

- If the patient (male or female) is from a family in which the disorder is passed only through males, it is most likely to be type I.

BLOCH–SULZBERGER SYNDROME
(incontinentia pigmenti)

Mode of inheritance

- X-linked dominant, with linkage of the familial form to Xq28 (lethal in males).

Ocular features

- Avascularity of peripheral temporal retina.
- Retrolental cicatricial retinal detachment, giving rise to leukocoria.
- Strabismus.
- Congenital cataract.
- Microphthalmos.
- Retinal dysplasia.

Systemic features

- Recurrent vesicobullous dermatitis, giving rise to patches or whorls of hyperpigmentation on the trunk and extremities.
- Hypodontia or delayed eruption of teeth.
- Patchy alopecia.
- CNS abnormalities (e.g. seizures, spasticity).
- Skeletal anomalies.

Comment

- Bullous skin lesions are usually present in early childhood. Seizures in childhood usually indicate poor prognosis for neurologic development.

CAT-EYE SYNDROME

Mode of inheritance

- Partial trisomy 22.

Ocular features

- Uveal coloboma.
- Downslanting palpebral fissures.
- Microphthalmos.

Systemic features

- Anal atresia.
- Cardiac defects.
- Renal agenesis.
- Preauricular pits or tags.
- Mild mental deficiency.

CAT-SCRATCH FEVER

At 1–2 weeks after a bite or a scratch from an infected cat, a papule appears with regional lymphadenopathy which persists for weeks. The organism is a Gram-positive bacillus.

Ocular features

- Parinaud oculoglandular fever.
- Neuroretinitis.

Systemic features

- The illness may progress to encephalitis, hepatitis, arthritis, bone lesions, or pleurisy.

CEREBELLAR ATAXIA TYPE I

Mode of inheritance

- Autosomal dominant, with gene thought to be located on chromosome 6p.

Ocular features

- One-third of patients have optic atrophy with mild visual loss.
- Nystagmus.
- Supranuclear ophthalmoplegia.
- Lid retraction.

Systemic features

- Progressive gait ataxia, usually in the second decade.
- Later, limbs and speech are affected, with reduced life-expectancy.

CHANCROID

This is an acute sexually transmitted disease that is endemic in areas of Africa and Asia and is caused by *Hemophilus ducreyi*.

Ocular features

- Parinaud oculoglandular fever.

Systemic features

- Ulcers on the penis in men or at the vaginal entrance in women.
- Unilateral inguinal lymphadenopathy, which may develop into suppurating buboes.

CHARCOT–MARIE–TOOTH DISEASE
(peroneal muscular atrophy)

This is a hereditary motor and sensory neuropathy (HMSN). There are three types: HMSN I (demyelinating neuropathy), HMSN II (axonal neuropathy), and distal spinal muscular atrophy.

Mode of inheritance

- Autosomal dominant or X-linked recessive.

Ocular features

- Optic atrophy and pigmentary retinopathy occasionally seen.

Systemic features

- Progressive distal limb wasting and weakness usually of the legs, which may lead to 'inverted champagne bottle' appearance of the legs.
- Loss of sensation and reflexes.
- Spastic paresis.
- Deafness.

C.H.A.R.G.E. (see Pagon syndrome).

CHÉDIAK–HIGASHI SYNDROME

Mode of inheritance

- Autosomal recessive, characterized by giant granules in myeloid cells and granular lymphocytes.

Ocular features

- Clinically appears to be ocular albinism, but histologically is oculocutaneous.
- Photophobia.
- Prone to retinal detachment.

Systemic features

- Repeated infections.
- Lympadenopathy due to infiltration of immature leukocytes.

Comment

- Diagnosis depends largely on history. Death usually occurs during the second decade.

CHRYSIASIS

This manifests as blue–black skin pigmentation, most evident in sun-exposed areas, and is seen in patients taking gold parenterally or orally.

Ocular features

- Gold deposition in cornea and lens.

CHURG–STRAUSS SYNDROME
(allergic granulomatosis)

Mode of inheritance

- Not inherited but thought to be an unusual progression of allergic disease seen in patients with rhinitis or asthma.

Ocular features

- Scleritis.
- Conjunctival granulomas.

Systemic features

- Severe eosinophilia.
- Small-vessel vasculitis, usually involving the lungs, peripheral nerves, and skin (subcutaneous nodules and purpuric patches).
- Extravascular granulomas.

Comment

- Look for p-ANCA.

COCCIDIOIDOMYCOSIS

This is caused by the fungus *Coccidioides immitis*, usually found in the central part of the Americas.

Ocular features

- Phlyctenular conjunctivitis.
- Parinaud oculoglandular fever.
- Posterior uveitis.

Systemic features

- Pulmonary infection, rarely with effusions.
- Polyarthritis with erythema nodosum.
- In severe cases only, pulmonary cavitation, meningitis, and hepatosplenomegaly.

COCKAYNE SYNDROME

Mode of inheritance

- Autosomal recessive condition with a defect in fibroblast DNA metabolism.

Ocular features

- Pigmentary retinopathy with optic disc pallor.
- Cataract.
- Miotic pupils which dilate poorly.
- Dry eyes.
- Strabismus.
- Nystagmus.

Systemic features

- Premature aging beginning in infancy.
- Photosensitive dermatitis.
- Postnatal growth deficiency, usually apparent after 2 years of age.
- Microcephaly in all cases by 2 years of age.
- Mental handicap.
- Sensorineural hearing loss.
- Skeletal malformations.
- Dental caries.

Comment

- No affected person has successfully reproduced offspring. Average age at death is 12 years. There is an early-onset severe form (type II) in which there is intrauterine onset of growth deficiency, early congenital cataracts, and structural eye defects, with death occurring by 7 years.

COGAN SYNDROME

Ocular features

- Interstitial keratitis.

Systemic features

- Acute tinnitus.
- Vertigo.
- Deafness.
- In some cases, associated polyarteritis nodosa.

COHEN SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Bull's-eye maculopathy.
- Pigmentary retinopathy.
- Optic atrophy.
- Strabismus.
- Constricted visual fields.

Systemic features

- Hypotonia beyond infancy.
- Open mouth with prominent central incisors.
- Obesity.
- Microcephaly.

CORNELIA DE LANGE SYNDROME

Mode of inheritance

- Sporadic, although autosomal dominant cases have been reported.

Ocular features

- Bushy eyebrows and synophrys.
- Long curly eyebrows.
- Myopia.
- Nystagmus.
- Ptosis.
- Occasionally, microcornea, optic nerve coloboma.

Systemic features

- Retarded bone maturation.
- Early hypertonicity.
- Thin upper lip with long philtrum.
- Micromelia.
- Microcephaly.
- Hearing loss with speech delay.
- Mental retardation.

Comment

- Growth retardation is marked even at birth, with failure to thrive.

CRANIOFRONTONASAL DYSPLASIA

Mode of inheritance

- Probably X-linked dominant, but females are more severely affected than males.

Ocular features

- Hypertelorism.
- Strabismus ('V' exotropia).

Systemic features

- Craniosynostosis in females only.
- Bifid nasal tip.
- Longitudinal split in nails.
- Syndactyly of fingers in females only.

Comment

- To make a diagnosis in a male, the patient must have a diagnosed female relative.

CRI-DU-CHAT SYNDROME

Mode of inheritance

- Partial deletion of 5p.

Ocular features

- Congenital cataract.
- Hypertelorism.
- Strabismus.
- Downslanting palpebral fissures.

Systemic features

- Growth retardation, cat-like cry, mental deficiency, and microcephaly in all cases.
- Low-set ears.

CROUZON SYNDROME

Mode of inheritance

- Autosomal dominant, but 25% of cases represent fresh mutation. Caused by mutation in the FGFR2 (fibroblast growth factor receptor) gene mapped to 10q.

Ocular features

- Shallow orbits with proptosis which is the most consistent feature.
- Hypertelorism.
- Divergent squint ('V' exotropia).
- Exposure keratopathy.
- Optic atrophy.

Systemic features

- Craniosynostosis, leading to short anteroposterior diameter and wide cranium.
- Maxillary hypoplasia.
- Curved parrot-like nose.
- Inverted V-shaped palate.
- Acanthosis nigricans.

CUTIS MARMORATA TELANGIECTASIA CONGENITA

Ocular features

- Infantile glaucoma.

Systemic features

- Localized or generalized networks of dilated veins in the skin, associated with spider nevi and ulcers.
- Hypertrophy of involved extremities.

CYSTINOSIS

There are three types:

1. Infantile nephropathic

Mode of inheritance

- Autosomal recessive.

Ocular features

- Corneal needle-like crystals evident by 4–6 months of age.
- Photophobia.
- Reduction in vision.
- Crystals in conjunctiva and uvea.
- Peripheral pigmentary retinopathy.

Systemic features

- Growth failure in the first year of life.
- Progressive renal tubular then glomerular dysfunction, resulting in uremia by 10 years of age.
- Blond hair and fair complexion.

2. Non-nephropathic (formerly adult type)

Mode of inheritance

- Autosomal recessive, originally described in adults but also found in children.

Ocular features

- Corneal crystals, but no ocular symptoms and no pigmentary retinopathy.

Systemic features

- No renal dysfunction, compatible with survival into adulthood.

3. Intermediate (adolescent cystinosis)

Mode of inheritance

- Autosomal recessive, with recognition often in teenage years.

Ocular features

- Conjunctival and corneal crystal deposits.
- Variable retinopathy and photophobia.

Systemic features

- Variable nephropathy.
- Normal hair and skin color.

DELLEMAN SYNDROME

Ocular features

- Microphthalmos with orbital cyst.

Systemic features

- Skin tags.
- Punched-out skin lesions over ears and other areas.
- Mental retardation.
- Hydrocephalus.
- Brain malformations.

DE MORSIER SYNDROME
(septo-optic dysplasia)

Ocular features

- Bilateral optic disc hypoplasia.

Systemic features

- Agenesis of the anterior commissure and septum pellucidum.
- Secondary hypopituitarism.
- Secondary growth retardation.

Comment

- If growth hormone is given before epiphyseal closure, growth retardation can be reversed.

DERMATOMYOSITIS – *see* Polymyositis

DEVIC DISEASE

Ocular features

- Bilateral optic neuritis with variable recovery.

Systemic features

- Paraplegia from transverse myelitis.
- Very high CSF protein concentrations.

D.I.D.M.O.A.D. – *see* Wolfram syndrome

DOWN SYNDROME

Mode of inheritance

- Trisomy for part or all of chromosome 21, which may be associated with increased maternal age.

Ocular features

- Upslanting palpebral fissures.
- Chronic blepharitis.
- Ectropion.
- Epicanthic folds.
- Squint.
- Refractive error, usually myopia.
- Nystagmus.
- Keratoconus.
- Iris hypoplasia.
- Brushfield spots.
- Blue dot lens opacities.
- Acquired cataract.
- Epiphora.
- Anomalous optic disc vasculature.

Systemic features

- Hypotonia.
- Flat facies.
- Small ears.
- Mental deficiency.
- Single transverse palmar crease (simian crease).
- Congenital cardiac anomaly.
- Recurrent respiratory infection.
- Thyroid dysfunction.
- Excess skin on back of neck.
- Stunted growth.

Comment

- A major cause of early death is cardiac disease.

DYSTROPHIA MYOTONICA
(myotonic dystrophy/Steinert syndrome)

Mode of inheritance

- Autosomal dominant (variable expressivity), with gene mapped to chromosome 19q.

Ocular features

- Cataract – evident by age 20, on slit-lamp examination.
- Bilateral symmetric ptosis.
- Light–near dissociation of pupillary reactions.
- Symmetric ophthalmoplegia.
- Mesh-like peripheral retinal pigment clumping.

Systemic features

- Myotonia with muscle atrophy.
- Hypogonadism.
- Expressionless myopathic facies.
- Premature frontal balding.
- Cardiac conduction defects.

Comment

- This condition displays genetic anticipation which is an increase in the phenotypic severity with successive generations.

ECHINOCOCCUS INFECTION (hydatid disease)
Infection with dog tapeworm *E. granulosus* or *E. multilocularis* occurs usually in early childhood by direct contact with infected dogs or ingestion of inadequately prepared vegetables contaminated with infected dog feces.

Ocular features

- Orbital myositis.
- Vitreous cysts.

Systemic features

- Cyst(s) in the liver, lung, kidney, or brain.

EDWARD SYNDROME

Mode of inheritance

- Trisomy 18.

Ocular features

- Ptosis.
- Corneal opacity.
- Uveal coloboma.
- Microphthalmos.
- Congenital cataract.
- Retinal dysplasia.

Systemic features

- Clenched hand.
- Short sternum.
- Low-arch dermal ridge pattern on six or more fingers.
- Cardiac anomalies.
- Mental deficiency.
- Low-set ears.

Comment

- Only 5–10% of patients survive the first year and those that do have severe mental deficiency.

EHLERS–DANLOS SYNDROME
There are ten distinct types, but only type I has occasional ocular features, which are described here.

Mode of inheritance

- Autosomal dominant.

Ocular features

- Blue sclera.
- Keratoconus.
- Lens subluxation.
- High myopia.
- Retinal detachment.
- Angioid streaks.

Systemic features

- Hyperextensible joints and skin.
- Defective wound healing.
- Easy bruising.
- Cardiac anomalies, e.g. mitral valve prolapse.

EPIDERMOLYSIS BULLOSA (EB)

There are four types. The main ocular feature is cicatricial conjunctivitis, but only when there is mucosal involvement, and blisters which heal with scarring.

1. EB simplex
- Blisters develop within the first year of life over contact areas of the skin, and heal without scarring.

2. EB dystrophica
- Blisters develop in early infancy on limbs (autosomal dominant) or large areas of blisters of skin and mucosa are present at birth which heal with scarring (autosomal recessive).

3. EB letalis
- Fatal in infancy, with mucosal and skin involvement.

4. EB acquisita
- Develops in adulthood.

FABRY DISEASE

Mode of inheritance

- X-linked recessive sphingolipidosis due to deficiency of alpha-galactosidase A.

Ocular features

- Corneal verticillata (vortex keratopathy).
- Conjunctival microaneurysms.
- Fundal arterial vascular tortuosities.
- Congenital spoke-like posterior cataracts.
- Periorbital puffiness.

Systemic features

- Purple telangiectatic skin lesions (angiokeratomas).
- Cardiovascular and progressive renal disease.
- Periodic crises of severe pain in the extremities.

Comment

- Female carriers display corneal verticillata. Death in affected males is usually due to renal failure, typically in the third to fifth decade.

FAMILIAL DYSAUTONOMIA

– *see* Riley–Day syndrome

FETAL VALPROATE SYNDROME

This is due to prenatal valproic acid exposure.

Ocular features

- Telecanthus.

Systemic features

- Narrow bifrontal diameter with high forehead.
- Midface hypoplasia.
- Relatively small mouth.
- Aortic coarctation.
- Cardiac defects.
- Long thin fingers with hyperconvex fingernails.
- Meningomyelocele.

FRASER SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Bilateral cryptophthalmos with defective eyes.
- Poor visual prognosis.

Systemic features

- Ear anomalies.
- Anomalous genitalia.
- Mental deficiency in 50% of cases.
- Cutaneous syndactyly.
- Renal hypoplasia or agenesis.

FREEMAN–SHELDON SYNDROME
(whistling face syndrome)

Mode of inheritance

- Autosomal dominant.

Ocular features

- Telecanthus.
- Blepharophimosis.

Systemic features

- Mask-like facies with small mouth, giving a 'whistling appearance'.
- Nasal speech.
- Hypoplastic alae nasi.
- Talipes equinovarus.
- Kyphoscoliosis.

FRIEDREICH ATAXIA

Mode of inheritance

- Usually autosomal recessive condition, with gene mapped to chromosome 9.

Ocular features

- Nystagmus.
- Pigmentary retinopathy.
- Optic atrophy.

Systemic features

- Progressive ataxia starting in the legs.
- Diabetes mellitus.
- Dysarthria.
- Absent knee and ankle jerks.
- Scoliosis.
- Loss of joint-position sense.
- Cardiac involvement.

Comment

- Presents in the first decade, with patients usually surviving until about 30 years of age.

FRONTONASAL DYSPLASIA SEQUENCE
(median clefting syndrome)

Mode of inheritance

- Unclear.

Ocular features

- Hypertelorism.
- Occasional microphthalmos.

Systemic features

- Notched broad nasal tip to complete division of nostrils.
- Widow's peak.

Comment

- Also known as median clefting syndrome. May be seen as part of craniofrontonasal dysplasia.

FUCOSIDOSIS

Mode of inheritance

- Autosomal recessive. There is a fatal infantile form (I) and a less severe form (II) with survival into adulthood.

Ocular features

- Conjunctival and retinal vessel tortuosity.

Systemic features

- Psychomotor and growth retardation.
- Coarse facies.
- Hepatosplenomegaly.
- Angiokeratomas (type II only) which are identical to those seen in Fabry disease.

GALACTOSEMIA

Mode of inheritance

- Autosomal recessive disorder characterized by an error in galactose metabolism due to deficiency of any one of three enzymes: galactokinase, transferase, and epimerase. Classic galactosemia has been mapped to a gene on chromosome 9 which codes for galactose-1-phosphate uridyl transferase.

Ocular features

- Cataract.

Systemic features

- Hepatomegaly.
- Mental retardation.

Comment

- Galactokinase deficiency alone may manifest only in adults with early-onset cataracts.

GANGLIOSIDOSIS

Gangliosides are glycosphingolipids that have oligosaccharide chains containing sialic acid. Disorders of ganglioside degradation can be divided into two types:

1. G_{M1} gangliosidosis (generalized)

Mode of inheritance

- Autosomal recessive condition due to deficiency of ganglioside G_{M1} beta-galactosidase. The gene has been assigned to chromosome 3p.The commoner infantile type displays:

Ocular features

- Macular cherry-red spot in 50% of cases.
- Corneas may be cloudy, but this is very subtle, even on slit-lamp examination.

Systemic features

- Coarse facies.
- Joint limitation.
- Hypotonia.
- Macroglossia.
- Postnatal growth deficiency.
- Variable hepatosplenomegaly.
- Kyphosis.
- Infants usually die by 2 years of age.

Comment

- The juvenile form presents at approximately 1 year of age and is characterized by ataxia, esotropia, and sensorimotor deterioration. Vision is not impaired and the fundi are normal.

2. G_{M2} gangliosidosis

This group includes three types:
- Type I – Tay–Sachs disease, classic.
- Type II – Sandhoff disease.
- Type III –Tay–Sachs disease, juvenile.

GARDNER SYNDROME

Mode of inheritance

- Autosomal dominant.

Ocular features

- Multiple, widely spaced, usually bilateral areas of congenital hypertrophy of the retinal pigment epithelium.

Systemic features

- Familial polyposis coli with increased incidence of carcinoma of the colon.
- Bony tumors.
- Ampullary carcinoma.
- Fibroadenomas.

GAUCHER DISEASE (GD)

Mode of inheritance

- Autosomal recessive lysosomal storage disorder that is due to a deficiency of glucocerebrosidase (β-glucosidase). The gene for this enzyme is located on chromosome 1q. There are three types:

1. GD I (adult type)

Ocular features

- Pinguecula-like conjuctival lesions.

Systemic features

- Splenomegaly presenting early with or without hypersplenism.
- Skeletal complications, e.g. aseptic necrosis of the femoral head, fractures.
- Skin pigmentation.

Comment

- Non-neuropathic, commoner type.

2. GD II (infantile type)

Ocular features

- Strabismus.

Systemic features

- Presents in the first 6 months of life.
- Marked hepatosplenomegaly.
- Progressive spasticity.
- Head retroflexion.

Comment

- Acute neuropathic form with death usually by 3 years of age.

3. GD III (juvenile type)

Ocular features

- Horizontal supranuclear gaze palsy.
- White retinal spots.

Systemic features

- Early onset.
- Short and underweight for age compared with GD I.
- Hepatosplenomegaly.
- Neurologic involvement.

GOLDENHAR SYNDROME
(oculo-auriculo-vertebral spectrum)

Mode of inheritance

- Usually sporadic.

Ocular features

- Epibulbar dermoid.
- Upper-lid notch.
- Microphthalmos.
- Disc coloboma.

Systemic features

- Hypoplasia of the malar, maxillary, and mandibular regions (may be unilateral).
- Macrostomia.
- Microtia.
- Accessory preauricular tags and/or pits.
- Hemivertebrae, usually cervical.

Comment

- Incidence of mental deficiency increases with presence of microphthalmos. It is thought that hemifacial microsomia and Goldenhar syndrome are part of the same spectrum of anomaly.

GOLTZ SYNDROME (focal dermal hypoplasia)

Mode of inheritance

- Unclear, but thought to be X-linked dominant, with the condition being fatal for males. Most cases represent fresh mutations.

Ocular features

- Uveal and disc coloboma.
- Microphthalmos.
- Strabismus.
- Anophthalmos.

Systemic features

- Fatty herniation through areas of focal dermal hypoplasia.
- Areas of altered skin pigmentation.
- Syndactyly.
- Hypodontia and/or adontia.
- Facial hemihypertrophy.
- Dystrophic nails.

GORLIN–GOLTZ SYNDROME (nevoid basal cell carcinoma syndrome)

Mode of inheritance

- Autosomal dominant, mapped to chromosome 9q.

Ocular features

- Hypertelorism.
- Synophrys.
- Coloboma of iris.
- Cataract.
- Basal cell carcinoma of eyelids.

Systemic features

- Macrocephaly.
- Basal cell carcinomas over upper body and face.
- Jaw cysts.
- Broad facies.
- Rib anomalies.
- Dyskeratotic palmar pitting.
- Other neoplasms.

Comment

- Basal cell carcinomas usually develop between the second and fourth decades of life. Patients must be kept under close review in order to detect other neoplasms.

GREIG SYNDROME

Mode of inheritance

- Autosomal dominant.

Ocular features

- Hypertelorism.

Systemic features

- Frontal bossing.
- Macrocephaly.
- Polydactyly.
- Syndactyly.

GRONBLAD–STRANDBERG SYNDROME

This is the association of pseudoxanthoma elasticum (PXE) with angioid streaks. PXE is an inherited disorder of elastin.

Mode of inheritance

- Autosomal dominant and recessive types have been described.

Ocular features

- Angioid streaks with or without choroidal neovascularization.

Systemic features

- Lax loose skin ('plucked-chicken appearance'), especially of the neck.
- Gastrointestinal bleeds.
- Intermittent claudication and angina.

HALLERMANN–STREIFF–FRANÇOIS SYNDROME

Ocular features

- Bilateral microphthalmos.
- Cataract which may be membranous.
- Nystagmus.
- Strabismus.
- Disc coloboma.

Systemic features

- Small thin nose (parrot-like).
- Brachycephaly with frontal bossing.
- Hypotrichosis.
- Malar hypoplasia with micrognathia.
- Postnatal growth retardation in two-thirds of cases.

Comment

- Craniofacial anomaly results in a narrow upper respiratory airway which may necessitate tracheostomy in early infancy.

HALLERVORDEN–SPATZ SYNDROME

Mode of inheritance

Autosomal recessive.

Ocular features

Flecked retina.
Bull's-eye maculopathy.

Systemic features

Progressive ataxia and dementia.
Dystonia.
Muscle tone abnormalities.
Acanthosis.

HAPPY PUPPET SYNDROME

see Angelman syndrome

HEMOCHROMATOSIS

Mode of inheritance

Autosomal recessive.

Ocular features

Dry eyes.
Rusty-brown perilimbal conjunctival and scleral discoloration.

Systemic features

Classic triad – bronze complexion, hepatomegaly, and diabetes mellitus – is unusual.
More commonly, hypogonadism, heart failure, and arthropathy.
Cirrhosis with increased incidence of hepatic carcinoma.

Comment

- Elevated serum iron and ferritin.

HERMANSKY–PUDLAK SYNDROME

Mode of inheritance

- Autosomal recessive form of tyrosinase-positive albinism, common in Puerto Rico and the Netherlands.

Ocular features

- Features of albinism, e.g. iris transillumination, lack of pigment in fundus.

Systemic features

- *Albinism.*
- Easy bruising due to platelet dysfunction.
- Lysosomal ceroid storage disease in the reticuloendothelial system and lung, leading to pulmonary fibrosis.

HIRSCHSPRUNG DISEASE

Congenital absence of ganglion cells in the submucosal or myenteric plexuses.

Ocular features

- Bilateral sectoral iris hypochromia.

Systemic features

- Massive abdominal distension.
- Absent bowel movements.

Comment

- Usually diagnosed in early infancy, but in milder cases may not be apparent until later in life.

HISTIOCYTOSIS X
(Langerhans-cell histiocytosis)

There are three varieties of this condition, all characterized by the presence of histiocytic granulomas.

1. Eosinophilic granuloma

Ocular features

- Rarely, may cause proptosis.

Systemic features

- Commonest form found in adults, with the lung and bones being affected.

2. Lettere–Siwe disease

Ocular features

- Orbital involvement is very unusual.

Systemic features

- Usually fatal disease of infants, with involvement of the skin, lymph nodes, liver, spleen, and bone.

3. Hand–Schüller–Christian disease

Ocular features

- Proptosis.

Systemic features

- Bone lesions.
- Diabetes insipidus.
- Pulmonary involvement mimics sarcoidosis with hilar lymphadenopathy.

Comment

- Characteristic triad is of bone lesions, proptosis, and diabetes insipidus.

HOMOCYSTINURIA

Mode of inheritance

- Autosomal recessive condition with decreased hepatic activity of cystathionine synthetase, leading to accumulation of homocystine and methionine.

Ocular features

- Lens subluxation (down), usually by 10 years of age.
- Myopia.
- Retinal detachment.

Systemic features

- Tall slim build.
- Arachnodactyly.
- Malar flush.
- Blond hair.
- Osteoporosis.
- Mental retardation.
- Tendency to thromboembolism.

Comment

- Some patients are responsive to vitamin B_6 replacement. In those that are not, a diet that is low in methionine and high in cystine may help to prevent mental retardation. The urinary nitroprusside test is used for rapid screening but not always positive; consequently, specific amino acid studies must be done.

HUNTER SYNDROME (MPS II)
– *see also* Mucopolysaccharidoses

Mode of inheritance

- There are no affected females as this condition is XL recessive. There is a mild type and a severe type. Death usually occurs in the latter by 15 years of age. Both types have deficiency of iduronate sulfatase, with urinary excretion of dermatan and heparan sulfate.

Ocular features

- Clear cornea.
- Pigmentary retinopathy.
- Glaucoma.

Systemic features

- Growth deficiency.
- Coarse facies with macroglossia and macrocephaly.
- Stiff joints by 2 to 4 years of age.
- Hepatosplenomegaly.

HURLER SYNDROME (MPS I-H)
– *see also* Mucopolysaccharidoses

Mode of inheritance

- Autosomal recessive with deficiency of lysosomal hydrolase alpha-L-iduronidase and urinary excretion of dermatan and heparan sulfate. The gene for the affected enzyme has been mapped to chromosome 4p. Survival beyond 10 years of age is unusual.

Ocular features

- Cloudy corneas by 1 to 2 years of age.
- Pigmentary retinopathy.
- Optic atrophy.
- Glaucoma.

Systemic features

- Coarse facies with full lips and flared nostrils.
- Stiff joints.
- Mental retardation.
- Hearing loss (almost always present).
- Growth deficiency.
- Hepatosplenomegaly.
- Macroglossia.
- Macrocephaly.

HURLER–SCHEIE SYNDROME (MPS I-H/S)
– *see also* Mucopolysaccharidoses

Mode of inheritance

- Autosomal recessive condition with deficiency of alpha-L-iduronidase and excretion of urinary dermatan and heparan sulfate. Gene located on chromosome 4p. Onset is after 3 years of age, with survival into the twenties.

Ocular features

- Corneal clouding.
- Pigmentary retinopathy.
- Glaucoma.

Systemic features

- Mild mental deficiency.
- Growth deficiency.
- Moderate joint limitation.
- Hearing loss.

HYPERLIPIDEMIA
The most well-known classification is that of Fredrickson (WHO):
I – chylomicronemia with raised triglycerides.
IIa – raised low-density lipoprotein (LDL) and cholesterol.
IIb – raised very-low-density lipoprotein (VLDL), LDL, cholesterol, and triglycerides.
III – raised cholesterol and triglycerides, with lowered LDL.
IV – raised VLDL and triglycerides.
V – chylomicronemia with raised VLDL and triglycerides.

However, the genetic classification (Goldstein) is much more useful:

Hypertriglyceridemia
(without hypercholesterolemia)

1. Familial hypertriglyceridemia with excess VLDL (WHO type IV)

Ocular features

- Retinal vein thrombosis.

Systemic features

- Attacks of pancreatitis.
- Associated with gout and alcoholism.
- Seen more in women.
- Eruptive cutaneous xanthomata.
- Atherosclerosis and hypertension.

2. Lipoprotein lipase deficiency and apoprotein C-II deficiency (WHO type I)

Ocular features

- Presents in childhood with eruptive xanthoma.
- Lipemia retinalis.
- Retinal vein thrombosis.

Systemic features

- Associated elevated chylomicrons (hyperchylomicronemia).
- Pancreatitis.
- Hepatosplenomegaly.
- No excess atherosclerosis.

(NB The combination of '1' and '2' is the equivalent of WHO type V).

Hypercholesterolemia
(without hypertriglyceridemia)

1. Familial hypercholesterolemia due to abnormal LDL cholesterol receptors in the liver (WHO type IIa)

Ocular features

- Xanthelasma (not diagnostic).
- Arcus lipoides.

Systemic features

- Xanthoma of Achilles tendon and extensor tendons of fingers.
- Premature coronary artery disease.

2. Homozygous hypercholesterolemia

Systemic features

- Death in late childhood or early adulthood from ischemic heart disease.

Combined hyperlipidemia

1. Familial combined hyperlipidemia (WHO type IIb)

Ocular features

- Arcus lipoides.
- Xanthelasma.

2. Remnant hyperlipidemia (WHO type III)

Systemic features

- Xanthomas in palmar creases.
- Tuberous xanthomas over extensor surfaces of elbows and joints.

HYPERLYSINEMIA

Mode of inheritance

- Autosomal recessive deficiency of lysine α-ketoglutarate reductase.

Ocular features

- Occasionally, lens dislocation.
- Microspherophakia.

Systemic features

- Mental retardation.
- Lax ligaments.
- Hypotonic muscles.
- Seizures.

HYPOPHOSPHATEMIA

Mode of inheritance

- X-linked due to renal tubular insufficiency.

Ocular features

- Shallow orbits.

Systemic features

- Normal muscle power.
- Associated metabolic changes.

ICHTHYOSIS

There are five types:

1. Congenital ichthyosis

Mode of inheritance

- Autosomal recessive condition that is usually lethal in the neonatal period.

Ocular features

- Ectropion.
- Congenital cataract.

Systemic features

- Hyperkeratosis involving the articular folds, with scaly skin and hyperhidrosis.

2. Lamellar ichthyosis

Mode of inheritance

- Autosomal recessive.

Ocular features

- Ectropion.

Systemic features

- Infant is born enveloped in dry, shining membrane that cracks, peels, and then re-forms.

3. Ichthyosis vulgaris

Mode of inheritance

- Autosomal dominant and mildest variant.

Ocular features

- Blepharitis.
- Corneal erosions.

Systemic features

- Skin changes developing after 3 months of age.

4. Bullous ichthyosiform erythroderma

Mode of inheritance

- Autosomal dominant condition that is similar to the congenital form but the skin exhibits bullae.

Ocular features

- Blepharitis.

5. X-linked ichthyosis

OF

- Blepharitis.
- Corneal erosions.

Systemic features

- Onset at birth, with similar skin changes as in the vulgaris form but the palms and soles are spared.

IgA NEPHROPATHY

This condition affects boys and young men.

Ocular features

- Scleritis.
- Anterior uveitis.

Systemic features

- Asymptomatic microscopic or macroscopic hematuria.
- Proteinuria.
- Approximately 20% develop renal failure.

INTRAUTERINE INFECTIONS (embryopathies)

1. Rubella

Ocular features

- Pigmentary retinopathy which may be associated with choroidal neovascularization in adulthood.
- Cataracts (less than a third are bilateral).
- Nystagmus.
- Strabismus.
- Microphthalmos.
- Glaucoma.
- Corneal opacity.

Systemic features

- Hearing loss.
- Cardiac defects.
- Microcephaly.
- Osteopathy.
- Hepatosplenomegaly.
- Mental deficiency.

Comment

- Cardiac defects and ocular defects are frequently associated. Almost all microphthalmic eyes have cataracts and almost all cataractous microphthalmic eyes have glaucoma.

2. Toxoplasmosis

Ocular features

- Microphthalmos.
- Cataract.
- Chorioretinitis.
- Optic atrophy.

Systemic features

- Seizures.
- Hydrocephalus.
- Microcephaly.
- Hepatosplenomegaly.
- Deafness.
- Intracranial calcification.

3. Cytomegalovirus

Ocular features

- Microphthalmos.
- Cataract.
- Keratitis.
- Chorioretinitis.
- Optic atrophy.

Systemic features

- Jaundice.
- Hepatosplenomegaly.
- Microcephaly.
- Intracranial calcification.

4. Varicella

Ocular features

- Microphthalmos.
- Cataract.
- Horner syndrome.
- Chorioretinitis.
- Optic atrophy.
- Optic disc hypoplasia.
- Nystagmus.

Systemic features

- Mental deficiency with cortical atrophy but with or without seizures.
- Cicatricial skin lesions.
- Limb hypoplasia.

Comment

- Half the affected individuals usually die in early infancy.

5. Syphilis

Ocular features

- Interstitial keratitis.
- Anterior uveitis.
- Optic atrophy.
- Pigmentary retinopathy.

Systemic features

- Deafness.
- Malformed incisors.

- Osteopathy.
- Saddle-shaped nose.

Comment

- Hutchinson triad comprises interstitial keratitis, malformed incisors, and deafness.

INCONTINENTIA PIGMENTI
– see Bloch–Sulzberger syndrome

JEUNE SYNDROME
(asphyxiating thoracic dystrophy)

Mode of inheritance

- Autosomal recessive.

Ocular features

- Pigmentary retinopathy.

Systemic features

- Small thoracic cage.
- Dwarfism with short limbs.
- Hypoplastic lungs.
- Hypoplastic iliac wings.
- Chronic nephritis.
- Biliary dysgenesis.
- Polydactyly.

Comment

- Early death resulting from asphyxia with or without pneumonia is the rule.

JUVENILE XANTHOGRANULOMA (JXG)
This is a non-neoplastic, self-limiting proliferation of histiocytes that affects children under the age of 2 years and resolves usually by the age of 5 years.

Ocular features

- Iris lesions giving rise to heterochromia.
- Spontaneous hyphema.

Systemic features

- Elevated papules on skin.
- Lesions in muscle, stomach, salivary glands, and other organs.

Comment

- Histology shows Touton giant cells.

KAWASAKI DISEASE
This is an idiopathic vasculitis-like illness that affects children, most commonly in Japan and the USA.

Ocular features

- Bilateral conjunctivitis.
- Uveitis (rare).

Systemic features

- Pyrexia.
- Diffuse erythema of the oral and pharyngeal mucosa.
- Cervical lymphadenopathy.
- Rash which later desquamates.
- Myocarditis.
- Coronary or other medium vessel thrombosis.

KEARNS–SAYRE SYNDROME

Mode of inheritance

- Mitochondrial cytopathy associated with mitochondrial DNA deletions.

Ocular features

- Progressive external ophthalmoplegia with ptosis.
- Pigmentary retinopathy.

Systemic features

- Onset before 20 years of age.
- Heart block.
- Cerebellar syndrome.
- Hearing loss.

Comment

- Sudden death can occur due to cardiac problems.

KLIPPEL–TRENAUNAY–WEBER SYNDROME

Ocular features

- Glaucoma.
- Cataract.
- Orbital varix, or other ocular hemangiomas.

Systemic features

- Hemangiomas of the legs, buttocks, abdomen, and lower trunk.
- Hypertrophy of usually one limb, but there may be multiple limb or facial involvement.

Comment

- Neurologic involvement (including mental deficiency) usually occurs in patients with hemangiomatosis.

KNIEST DYSPLASIA

Mode of inheritance

- Autosomal dominant, but most cases represent a fresh mutation.

Ocular features

- High myopia.
- Retinal detachment.
- Vitreoretinal degeneration.
- Lens subluxation.

Systemic features

- Flat facies.
- Enlarged joints.
- Platyspondyly.
- Disproportionate short stature.

Comment

- Neonates display short, stiff limbs and have apparently large heads. This condition is one of a number due to a defect in the type II collagen gene, COL2A1; the others include Stickler syndrome.

LAURENCE–MOON SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Retinitis-pigmentosa-like pigmentary retinopathy.
- Choroidal atrophy.

Systemic features

- Hypogonadism.
- Spastic diplegia.
- Mental handicap.

Comment

- Polydactyly and obesity are **not** part of this syndrome but are seen in the Bardet–Biedl syndrome.

LCAT(LECITHIN–CHOLESTEROL–ACETYLTRANSFERASE) DEFICIENCY (Norum disease)

Mode of inheritance

- Autosomal recessive.

Ocular features

- Progressive corneal opacification due to lipid crystal deposits.
- Arcus-like changes extending into the sclera.

Systemic features

- Abnormal lipid profile.
- Early atherosclerosis.
- Renal disease.
- Anemia.

Comment

- Most patients with this rare condition survive into adult years. The corneal deposits are unesterified cholesterol crystals which are present from early childhood.

LENZ MICROPHTHALMOS SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Uveal coloboma.
- Microphthalmos (colobomatous or noncolobomatous).

Systemic features

- Delayed development.
- 'Bat ears'.
- Ptosis.
- Skeletal anomalies.
- Urogenital anomalies.

LINEAR IgA DISEASE
(linear IgA bullous dermatosis)

This is a rare disorder in which there is linear deposition of IgA at the basement membrane zone.

Ocular features

- Cicatricial conjunctivitis.

Systemic features

- Skin lesions resemble those of bullous pemphigoid.

LOUIS-BAR SYNDROME
– see Ataxia telangiectasia

LOWE SYNDROME
(oculocerebrorenal syndrome)

Mode of inheritance
- X-linked recessive.

Ocular features
- Congenital cataract.
- Glaucoma.
- Microphakia.
- Posterior lenticonus.
- Miosis and poorly dilating pupils.

Systemic features
- Growth retardation.
- Mental deficiency.
- Hypotonia.
- Renal tubular acidosis.
- Aminoaciduria.
- Renal rickets.

Comment
- Female carriers may show lens opacities of varying severity.

LYME DISEASE
This condition is caused by *Borrelia burgdorferi*, which is transmitted by the deer tick *Ixodes dammini*.

Ocular features
- Uveitis.
- Conjunctivitis.
- Episcleritis.
- Interstitial keratitis.
- Ophthalmoplegia.
- Optic neuritis.

Systemic features
STAGE I
- Characteristic skin lesion – erythema chronicum migrans.
- Myalgia, arthralgia, and lymphadenopathy.

STAGE II
- Neurologic symptoms, e.g. cranial or peripheral neuropathies.
- Myocarditis.

STAGE III
- Arthritis.

LYMPHOGRANULOMA VENEREUM
This is a sexually transmitted disease caused by infection with a strain of *Chlamydia trachomatis*.

Ocular features
- Parinaud oculoglandular fever.

Systemic features
- Painless ulceration of genitalia.
- Painful enlarged regional lymph nodes which progress to buboes and which may rupture.
- An acute form may present as proctitis with or without perirectal abscesses.

MAFFUCCI SYNDROME

Mode of inheritance
- Sporadic.

Ocular features
- Hemangiomas of the eyelid and orbit.

Systemic features
- Skin hemangioma.
- Enchondromata of the hands, feet, and long bones.
- Variable early bowing of long bones.

MANDIBULOFACIAL DYSOSTOSIS
– see Treacher Collins syndrome

MANNOSIDOSIS

Mode of inheritance
- Very rare autosomal recessive disorder with deficiency of alpha-mannosidase and consequent excretion of mannose-containing oligosaccharides in the urine. There are two types:

1. Infantile type (I)

Ocular features
- Punctate lens opacities – arranged in a spoke-like pattern in the posterior lens cortex.

Systemic features
- Rapidly progressive mental deterioration.
- Hepatosplenomegaly.
- Bony deformities.

2. Juvenile–adult type (II)

Ocular features

- Similar to the infantile type.

Systemic features

- Normal early development, with mental deterioration in later childhood.
- Prominent hearing loss.

MARFAN SYNDROME

Mode of inheritance

- Autosomal dominant due to mutations in fibrillin (FBN1) gene on chromosome 15q.

Ocular features

- Lens subluxation (up).
- Microspherophakia.
- High myopia.
- Retinal detachment.
- Blue sclera.
- Dilator iris muscle hypoplasia.
- Angle anomaly and glaucoma.

Systemic features

- Skeletal – arachnodactyly with hyperextensibility, tall stature with long limbs, pectus excavatum, high-arched palate, scoliosis and kyphosis.
- Cardiovascular – aortic dilatation with or without dissecting aneurysm of the ascending aorta, aortic regurgitation, and mitral valve prolapse.

Comment

- This syndrome may occur without either ectopia lentis or obvious arachnodactyly. Cardiovascular anomalies are usually the cause of death, with the mean age of survival being in the early forties.

MAROTEAUX–LAMY SYNDROME (MPS VI-A, MPS VI-B) – *see also* Mucopolysaccharidoses

Ocular features

- Cloudy corneas in infancy.
- Optic atrophy.
- Glaucoma.
- Retinal vessel dilatation and tortuosity.

Systemic features

- Coarse facies.
- Stiff joints.
- Hepatosplenomegaly.
- Variable deafness.

Comment

- The feature that differentiates this from Hurler syndrome is the lack of mental retardation during early childhood. There is deficiency of the enzyme N-acetylgalactosamine-4-sulfatase, the gene for which has been mapped to chromosome 5q. It is associated with increased urinary excretion of dermatan sulfate. There are three subtypes, which vary in degree of severity.

MARSHALL–SMITH SYNDROME

Ocular features

- Shallow orbits.
- Blue sclera.

Systemic features

- Accelerated prenatal skeletal maturation and growth.
- Broad middle phalanges.
- Mental deficiency.
- Umbilical hernia.

McCUNE–ALBRIGHT SYNDROME (fibrous dysplasia)

Ocular features

- Proptosis.
- Compressive optic neuropathy from thickening of bone.

Systemic features

- Multiple areas of fibrous dysplasia.
- Irregular skin pigmentation.
- Endocrine dysfunction.
- Pituitary adenoma.
- Sexual precocity.

MECKEL–GRUBER SYNDROME

Mode of inheritance

- Autosomal recessive with gene locus mapped to 17q.

Ocular features

- Uveal coloboma.
- Microphthalmos.
- Cataract.

Systemic features

- Encephalocele.
- Polydactyly.
- Polycystic disease of the kidneys, liver, and pancreas.
- Microcephaly.
- Micrognathia.
- Cleft palate.

Comment

- Survival is seldom more than a few days to weeks.

MERETOJA SYNDROME

Mode of inheritance

- Autosomal dominant amyloidotic polyneuropathy mainly affecting patients of Finnish descent.

Ocular features

- Type II lattice corneal dystrophy.
- Open-angle glaucoma.
- Blepharochalasis.

Systemic features

- Cranial neuropathy.
- Multiorgan involvement.

MÖBIUS SYNDROME

Ocular features

- VI and VII nerve palsies, usually bilaterally.
- III and IV nerve palsies occasionally.
- Ptosis occasionally.

Systemic features

- Mask-like facies.
- Small, often hypoplastic tongue.
- Palsies of the V, IX, X, and/or XII nerves.
- Micrognathia.
- High nasal bridge.
- Mental retardation in a minority of cases.

MORQUIO SYNDROME (MPS IV-A, MPS IV-B)

– *see also* **Mucopolysaccharidoses**

Ocular features

- Corneal clouding evident after 5 years of age.
- Glaucoma.

Systemic features

- Mild coarse facies.
- Kyphoscoliosis.
- Knock-knees.
- Metaphyseal widening.
- Hearing loss.
- Flaring of lower ribcage.

Comment

- Onset is at 1 to 3 years. Type IV-A displays a deficiency of N-acetylgalactosamine-6-sulfatase (gene mapped to 16q) and is the more severe type, while type IV-B is due to a deficiency of beta-galactosidase (gene mapped to 3p) and is the milder form. There is urinary excretion of keratan sulfate.

MUCOLIPIDOSES

Formerly, this condition was divided into four subgroups:

1. Type I

Goldberg syndrome, now known as **sialidosis**, is due to deficiency of alpha-neuraminidase.

Mode of inheritance

- Autosomal recessive.

Ocular features

- Subtype I – macular cherry-red spots; some patients have small dot and flake lens opacities.
- Subtype II – macular cherry-red spots, corneal clouding; some patients show punctate lens opacities.

Systemic features

- Subtype I – myoclonus and ataxia, with onset in the second decade.
- Subtype II – coarse facies, myoclonus, ataxia, and abnormal vertebrae.

2. Type II

Now known as Leroy **I-cell disease**, is due to defective processing of lysosomal enzymes.

Mode of inheritance

- Autosomal recessive.

Ocular features

- Pigmentary retinopathy.
- Mild and late corneal clouding.

Systemic features

- Infantile onset of Hurler-like features (facial and skeletal changes with delayed growth and development).
- Congenital dislocation of the hip.
- No urinary mucopolysaccharides.
- Death occurs by 5 years of age.

3. Type III

Now known as **pseudo-Hurler polydystrophy syndrome**, is also due to defective processing of lysosomal enzymes.

Mode of inheritance

- Autosomal recessive.

Ocular features

- Peripheral corneal clouding at, or soon after, birth.

Systemic features

- Milder variant of I-cell syndrome, with symptoms beginning around 4 or 5 years of age with stiff joints.
- No urinary mucopolysaccharides.
- Some patients survive into their twenties.

4. Type IV

Mode of inheritance

- Autosomal recessive.

Ocular features

- Gross congenital corneal clouding.
- Cataract.
- Pigmentary retinopathy.

Systemic features

- Delayed development without skeletal or facial deformities.
- Organomegaly.
- Gross neurologic deficit.

MUCOPOLYSACCHARIDOSES (MPS)

These are a group of related, inherited deficiencies of catabolic glycosidases which are needed for the hydrolysis of mucopolysaccharides. The altered metabolite accumulates in single-membrane vacuoles in the cells of various tissues and organs and is also detectable in the urine. All types are autosomal recessive except Hunter syndrome, which is X-linked recessive. The types are:

Type I	MPS I-H (Hurler)
	MPS I-S (Scheie)
	MPS I-H/S (Hurler–Scheie)
Type II	MPS II (Hunter)
Type III	MPS III-A (Sanfilippo A)
	MPS III-B (Sanfilippo B)
	MPS III-C (Sanfilippo C)
	MPS III-D (Sanfilippo D)
Type IV	MPS IV-A (Morquio – classic)
	MPS IV-B (Morquio-like)
Type VI	MPS VI-A (Maroteaux–Lamy)
	MPS VI-B (Maroteaux–Lamy)
Type VII	MPS VII (Sly)

See under each specific named syndrome for more details.

MULTIPLE NEUROMA SYNDROME
(multiple endocrine neoplasia IIb – MEN IIb)

Mode of inheritance

- Autosomal dominant, with gene mapped to 10q. This is one of three types of MEN II, the other two being MEN IIa and familial medullary thyroid carcinoma (FMTC).

Ocular features

- Prominent corneal nerves.
- Anteverted eyelids.

Systemic features

- Multiple neuroma of the tongue, lips, and intestinal mucosa.
- Medullary thyroid carcinoma.
- Pheochromocytoma.
- Marfanoid habitus.

Comment

- Patients may develop constipation with megacolon before endocrine disorders are diagnosed. MEN IIa is characterized by medullary thyroid carcinoma, parathyroid hyperplasia, and pheochromocytoma, while FMTC displays thyroid carcinoma only.

NANCE–HORAN SYNDROME

Mode of inheritance

- Very rare X-linked recessive disorder.

Ocular features

- Microcornea or microphthalmos.
- Congenital cataract.

Systemic features

- Anteverted and simplex pinnae.
- Dental anomalies.

NEUROFIBROMATOSIS (NF)

There are three main types: NF I, NF II, and segmental NF.

1. NF I (von Recklinghausen's disease)

Mode of inheritance

- Autosomal dominant, with gene mapped to 17q. Half of all patients have a fresh gene mutation.

Ocular features

- Plexiform eyelid neurofibromas giving rise to S-shaped mechanical ptosis.
- Prominent corneal nerves.
- Iris – Lisch nodules, ectropion uveae, mammillations.
- Glaucoma.
- Optic nerve and chiasm glioma.
- Fundus hamartomas.
- Proptosis due to one of the following: optic nerve glioma, other neural tumors, spheno-orbital encephalocele.

Systemic features

- Skin – café-au-lait spots (100% of cases by 4 years of age), axillary freckles, fibroma molluscum, multiple neurofibromata.
- Skeletal defects – congenital bony defects, acquired scoliosis, facial asymmetry, short stature.
- Miscellaneous – malignancies, hypertension, mental impairment.

Comment

- Only 5% of patients with optic nerve glioma (regardless of its location) become symptomatic. All patients have Lisch nodules after 20 years of age (95% after 6 years). Cutaneous neurofibromas usually develop after 6 years of age.

2. NF II (acoustic NF)

Mode of inheritance

- Autosomal dominant, with gene mapped to 22q. Later age of onset.

Ocular features

- Presenile posterior subcapsular cataract.
- Combined hamartoma of the retinal pigment epithelium and retina.

Systemic features

- Bilateral acoustic neuromas – develop over the second and third decades.
- Few café-au-lait spots and cutaneous neurofibroma.

3. Segmental NF

Mode of inheritance

- Thought to be due to somatic mutation.

Ocular features

- Unilateral Lisch nodules if involved segment includes one of the eyes.

Systemic features

- Café-au-lait spots, cutaneous neurofibromas, and internal neurofibromas, all confined to a body segment.

NEURONAL CEROID LIPOFUSCINOSIS

– *see* Batten–Vogt syndrome

NIEMANN–PICK DISEASE

(sphingomyelin lipidosis)

There are five distinct types: classic infantile type (A), visceral type (B), juvenile type (C), Nova Scotia type (D), and adult type (E). Types A, B, and C have deficiency of sphingomyelinase; the abnormality of sphingomyelin storage in types D and E is unclear. All types are autosomal recessive.

1. Type A

Ocular features

- Cherry-red spot.
- Subtle corneal haze.

Systemic features

- Occurs most often in Ashkenazi Jews.
- Hepatosplenomegaly.
- General loss of motor function and mental capabilities early in infancy.
- Death usually occurs by the third year.

2. Type B

Ocular features

- Bull's-eye maculopathy occurs in some patients.

Systemic features

- No CNS involvement.
- Hepatosplenomegaly.
- Patients usually survive into adulthood.

3. Type C

Ocular features

- Some patients display characteristic deficiency of downward gaze.

Systemic features

- Later onset of neurologic abnormalities than in type A.
- Patients appear normal for first 1–2 years and then lose speech, develop ataxia, and start to have fits.

4. Types D and E

Ocular features

- Deficiency of downward gaze.

Systemic features

- Ataxia.
- Athetosis.

NOONAN SYNDROME (Turner-like syndrome)

Mode of inheritance

- Autosomal dominant.

Ocular features

- Strabismus.
- Epicanthic folds and downsloping palpebral apertures.
- Ptosis.
- Hypertelorism.
- Keratoconus.
- Myopia.
- Optic disc hypoplasia.

Systemic features

- Abnormal facies.
- Webbing of the neck.
- Pectus excavatum.
- Short stature.
- Cryptorchidism.
- Congenital heart disease.
- One-third of patients have bleeding disorders.

NORRIE DISEASE

Mode of inheritance

- X-linked recessive.

Ocular features

- Bilateral total retinal detachments presenting as retrolental masses at birth.

Systemic features

- Hearing loss.
- Mental deficiency.
- CNS degenerative changes.

Comment

- A recent histopathologic study of an 11 weeks' gestation fetus suggests that there is no retinal dysplasia as previously thought and that the detachments occur as a result of a primary abnormality of vascular proliferation, perhaps related to persistent hyperplastic vitreous.

OCULODENTODIGITAL SYNDROME

Mode of inheritance

- Autosomal dominant, but most cases are fresh mutations.

Ocular features

- Microphthalmos.
- Open-angle glaucoma, which may be a late complication and therefore these patients should have ophthalmic review.
- Microcornea.
- Epicanthic folds.
- Cataract.
- Iris anomaly.

Systemic features

- Camptodactyly of the fifth finger.
- Enamel hypoplasia.

OLIVOPONTOCEREBELLAR ATROPHY

Mode of inheritance

- There are five types: all are autosomal dominant except type II, which is autosomal recessive.

Ocular features

- Only type III is associated with a bull's-eye maculopathy.

Systemic features

- Onset of ataxia, tremor, involuntary movement, usually in middle age.

OSTEOGENESIS IMPERFECTA

There are four types: I, II, III, and IV, all of which are autosomal dominant.

1. Type I

Ocular features

- Blue sclera.
- Megalocornea.

Systemic features

- Fractures.
- Hyperextensible joints.
- Hearing loss.
- Triangular facial appearance.
- Easily bruised.
- Growth deficiency (postnatal).
- Dental hypoplasia.

2. Type II

Ocular features

- Deep-blue sclera.
- Shallow orbits.

Systemic features

- Multiple fractures.
- Short, thick long-bones.
- Prenatal growth deficiency.
- Patients normally die in early infancy.

3. Type III

Ocular features

- Blue sclera in infancy but not in adulthood.

Systemic features

- Multiple fractures at birth.
- Anomalous dentition.
- Hearing loss.
- Prenatal growth deficiency.

4. Type IV

Ocular features

- Normal sclera.

Systemic features

- Normal or moderately short stature.
- Bone deformity.
- Femoral bowing at birth.

OSTEOPETROSIS

Mode of inheritance

- There are two main types: one is autosomal recessive (AR) and severe, while the other is autosomal dominant (AD) and mild.

1. AR osteopetrosis

Ocular features

- Optic nerve compression.
- Strabismus.
- Shallow orbits.

Systemic features

- Dense, fragile bone.
- Marrow compression causes pancytopenia.
- Cranial nerve compression.
- Failure of dental eruption.
- One-third survival at 6 years.

2. AD osteopetrosis

Ocular features

- Ocular involvement is infrequent.

Systemic features

- Mild, delayed features.
- Mild skeletal changes.
- Bone pain.
- Normal lifespan.

PAGET DISEASE

Ocular features

- Optic atrophy.
- Cranial nerve palsies.

Systemic features

- Uncontrolled bone turnover with excessive osteoclastic and osteoblastic activity resulting in excessive new bone formation that is structurally abnormal. Bones commonly involved are the femur, pelvis, tibia, skull, and lumbosacral spine.

Comment

- Look for X-ray changes and raised serum alkaline phosphatase.

PAGON SYNDROME (C.H.A.R.G.E.)

Ocular features/Systemic features

- Coloboma – varies from iris/retinal coloboma to microphthalmos/anophthalmos.
- Heart defects.
- Atresia choanae – bony and /or soft tissue.
- Retarded growth – postnatal.
- Genital anomalies.
- Ear anomalies – and/or deafness.

PARRY–ROMBERG SYNDROME
(progressive hemifacial atrophy)

Ocular features

- Congenital iris hypochromia.
- Horner syndrome.
- Progressive enophthalmos.
- Pseudoptosis.
- Restrictive strabismus.

Systemic features

- Progressive facial atrophy, usually unilateral.

PATAU SYNDROME

Mode of inheritance

- Trisomy 13.

Ocular features

- Coloboma.
- Microphthalmos.
- Retinal dysplasia.
- Anophthalmos.
- Cyclopia.
- Cataract.
- Optic nerve dysplasia.

Systemic features

- Forebrain defects.
- Hare lip and cleft palate.
- Posterior scalp skin defects.
- Polydactyly.
- Hyperconvex narrow nails.
- Cardiac defects.

Comment

- Only 5% of patients survive beyond 6 months and those that do are severely mentally retarded.

PEMPHIGUS VULGARIS

Onset is between 40 and 60 years of age; there is an association with HLA-DR4.

Ocular features

- Cicatricial conjunctivitis.

Systemic features

- Early in disease, mainly oral mucosal involvement.
- Later, widespread cutaneous thin-walled blisters.

PERONEAL MUSCULAR ATROPHY
– see Charcot–Marie–Tooth disease

PETERS-PLUS SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Peters anomaly.
- Glaucoma.
- Nystagmus.

Systemic features

- Mental retardation.
- Prenatal and postnatal growth deficiency.
- Cardiac defects.
- Prominent forehead.
- Significantly delayed developmental milestones.

PIERRE ROBIN SYNDROME
– see Robin sequence

POLYARTERITIS NODOSA (PAN)

There are two forms of this: the classic form and microscopic polyarteritis.

1. Classic PAN

Ocular features

- Peripheral corneal ulceration.
- Scleritis.
- Orbital pseudotumor.
- Retinal arteritis.

Systemic features

- Myalgia, arthralgia, and pyrexia.
- Renal dysfunction.
- Polyneuropathy (mononeuritis multiplex).
- Hypertension.
- Myocardial infarction and/or heart failure.
- No lung involvement.
- Affects elderly males.
- p-ANCA negative.

2. Microscopic polyarteritis

Ocular features

- Ocular involvement is unusual.

Systemic features

- Renal dysfunction.
- Lung involvement.
- p-ANCA positive.

POLYCYTHEMIA RUBRA VERA

This is due to excess proliferation of erythroid, myeloid, and megakaryocytic progenitor cells. Clinical features usually present in patients over the age of 60 years and are due to hyperviscosity and increased bone marrow activity.

Ocular features

- Slow-flow retinopathy (cotton-wool spots, flame-shaped hemorrhages).
- Conjunctival telangiectasia.

Systemic features

- Severe itching after bathing.
- Thrombosis and hemorrhage.
- Gout.
- Hypertension and angina.

Comment

- Look for raised hemoglobin, packed cell volume, leukocyte alkaline phosphatase score, and serum uric acid.

POLYMYOSITIS AND DERMATOMYOSITIS

The etiology of these conditions is unknown, but dermatomyositis is associated with an increased risk of carcinoma of the bronchus (males) and carcinoma of the ovary (females). The two conditions are identical except for the fact that dermatomyositis is accompanied by a rash.

Ocular features

- Keratoconjunctivitis sicca.
- Scleritis.
- Retinal vasculitis.

Systemic features

- Proximal muscle weakness and wasting.
- Arthralgia/arthritis.
- Dysphagia.
- Raynaud phenomenon.
- Sjögren syndrome.

Comment

- Look for raised serum creatine phosphokinase and aldolase, distinctive EMG, and muscle histology showing necrosis of fibers with swelling of muscle cells.

PORPHYRIA CUTANEA TARDA

This is due to an abnormality in hepatic uroporphyrinogen decarboxylase.

Ocular features

- Cicatricial conjunctivitis.
- Scleritis.

Systemic features

- Bullous eruption of skin on exposure to sunlight which heals with scarring.

Comment

- Look for raised urinary uroporphyrin.

PRADER–WILLI SYNDROME

Mode of inheritance

- Sporadic deletion of 15q.

Ocular features

- Upslanting palpebral fissures.
- Congenital ectropion uveae.
- Strabismus.

Systemic features

- Obesity.
- Hypotonia.
- Small hands/feet.
- Mild mental retardation.
- Cryptorchidism.
- Behavioral problems.

PRIMARY BILIARY CIRRHOSIS

This is a presumed autoimmune disorder affecting females in the fifth decade and causing damage to bile ducts.

Ocular features

- Keratoconjunctivitis sicca in 70% of cases.

Systemic features

- Hepatosplenomegaly.
- Pruritus, which usually precedes jaundice.
- Pigmented xanthelasma due to secondary hypercholesterolemia.

Comment

- Look for anti-mitochondrial and anti-smooth-muscle antibody, raised alkaline phosphatase, and raised cholesterol.

RENDU–OSLER–WEBER SYNDROME

(hereditary hemorrhagic telangiectasia or Osler hemorrhagic telangiectasia syndrome)

Mode of inheritance

- Autosomal dominant, with gene mapped to 9q.

Ocular features

- Conjunctival telangiectasia.
- Retinal hemorrhages.

Systemic features

- Telangiectasia of lips, skin, and tongue.
- AV malformation of gastrointestinal (GI) system and/or lung.
- Epistaxis, which is the commonest form of bleeding, followed by GI bleed.

REFSUM DISEASE

(heredopathia atactica polyneuritiformis)

Mode of inheritance

- Autosomal recessive condition due to phytanic acid accumulation throughout the body secondary to phytanic acid alpha-hydrolase deficiency.

Ocular features

- Nyctalopia.
- Retinitis pigmentosa.
- Optic nerve atrophy.
- Miotic, poorly dilating pupils.

Systemic features

- Peripheral neuropathy.
- Cerebellar ataxia.
- Increased CSF protein in the absence of increased cells.
- Neural deafness.
- Ichthyosis.
- Baldness.
- Cardiac arrhythmias.
- Skeletal abnormalities.

Comment

- Signs and symptoms are often present before 10 years of age, and early detection and mangement (with a diet low in phytanic acid) can arrest disease progression.

RHINOSPORIDIOSIS

This is caused by the fungus *Rhinosporidium seberi*; is mainly seen in south India.

Ocular features

- Very rarely affects the conjunctiva.

Systemic features

- Polyp-like delicate vascular lesions of the mucosa of the nose, nasopharynx, and soft palate.

RIEGER SYNDROME

Mode of inheritance

- Autosomal dominant.

Ocular features

- Posterior embryotoxon.
- Iris hypoplasia.
- Polycoria.
- Corectopia.
- Ectropion uveae.
- Glaucoma.
- Iris mammillations.

Systemic features

- Facial anomalies.
- Dental anomalies.
- Rudimentary paraumbilical skin.
- Defect in the region of the pituitary gland.

RILEY–DAY SYNDROME
(familial dysautonomia)

Mode of inheritance

- Autosomal recessive condition usually found in Ashkenazi Jews in which there is autonomic instability.

Ocular features

- Alacrima.
- Corneal ulceration.

Systemic features

- Excessive sweating.
- Loss of vasomotor control.
- Labile hypertension.
- Impaired taste.
- Decreased pain and temperature sensation, which may result in neuropathic (Charcot) joints.
- Dysphagia.
- Hyporeflexia.

ROBERTS SYNDROME
(Roberts-SC phocomelia)

Mode of inheritance

- Autosomal recessive.

Ocular features

- Shallow orbits.
- Hypertelorism.
- Corneal clouding.

Systemic features

- Hypomelia.
- Growth deficiency (prenatal).
- Midfacial cleft.
- Microcephaly.

ROBIN SEQUENCE **(Pierre Robin syndrome)**

This usually occurs in otherwise healthy individuals but may be seen in Edward syndrome (trisomy 18) or in association with Stickler syndrome.

Systemic features

- Micrognathia.
- Glossoptosis.
- Cleft soft palate.

Comment

- The sequence itself is due to mandibular hypoplasia and has no ocular associations *per se*.

ROTHMUND–THOMSON SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Juvenile zonular cataracts, usually apparent between 2 and 7 years of age.
- Corneal dystrophy.

Systemic features

- Poikiloderma (irregular pigmentation and depigmentation, scarring and atrophy of skin).
- Short stature.
- Sparse hair.
- Microdontia and/or anodontia.
- Dystrophic nails.

RUBINSTEIN–TAYBI SYNDROME

Ocular features

- Downslanting palpebral fissures.
- Long eyelashes.
- Ptosis.
- Epicanthic folds.
- Strabismus.
- Uveal coloboma.
- Congenital glaucoma.

Systemic features

- Broad thumbs and toes.
- Maxillary hypoplasia.
- Speech difficulties.
- Stiff, unsteady gait.
- Postnatal growth deficiency.
- Cryptorchidism.
- Low IQ.

Comment

- Features can be very variable, even among members of the same family, but almost all cases have broad big toes and maxillary hypoplasia.

RUSSELL–SILVER SYNDROME

Ocular features

- Blue sclera.

Systemic features

- Short stature.
- Small triangular face.
- Tendency for fasting hypoglycemia in early infancy.
- Limb asymmetry.

SANDHOFF DISEASE
(G_{M2} gangliosidosis, type II)

Mode of inheritance

- Autosomal recessive due to deficiency of hexosaminidase A and B.

Ocular features

- Cherry-red spot.
- Early blindness.

Systemic features

- Neural and non-neural tissue involvement, with symptoms evident within the first 6 months.
- Occasionally, bony deformities.

Comment

- The clinical features of this condition are very similar to those of Tay–Sachs disease.

SANFILIPPO SYNDROME (MPS III-A, -B, -C, and -D) – *see also* Mucopolysaccharidoses

Mode of inheritance

- Autosomal recessive.The different types of Sanfilippo syndrome are clinically identical and in all four types there is excess urinary heparan sulfate. They are differentiated by deficiencies in different enzymes.

Ocular features

- Progressive rod–cone dystrophy.
- Glaucoma.
- Optic atrophy.
- Synophrys.

Systemic features

- Moderately coarse facies.
- Mildly stiff joints.
- Mental deficiency.
- Hepatosplenomegaly.
- Cardiac problems.

Comment

- A significant number of patients die by the second decade.

SCHEIE SYNDROME (MPS I-S)
– *see also* Mucopolysaccharidoses

Mode of inheritance

- Autosomal recessive condition with deficiency of α-L-iduronidase, leading to excess urinary dermatan sulfate. The gene is located on chromosome 4p.

Ocular features

- Early corneal clouding.
- Pigmentary retinopathy.
- Optic atrophy.

Systemic features

- Normal intelligence.
- Broad mouth.
- Stiff joints, especially of the hands.
- Truncal hirsutism.
- Short neck.
- Increased incidence of aortic valve disease.

Comment

- Symptoms usually start after 5 years of age, with a fairly normal lifespan.

SCHILDER DISEASE

Ocular features

- Bilateral optic neuritis without subsequent visual improvement.

Systemic features

- Relentless progressive widespread demyelination indistinguishable histologically from multiple sclerosis.
- Onset before 10 years of age and fatal within 1 to 2 years.

SIALIDOSIS
(formerly mucolipidosis I) – *see* Mucolipidoses

SJÖGREN–LARSSON SYNDROME

Mode of inheritance

- Autosomal recessive.

Ocular features

- Colobomatous microphthalmos.
- Congenital cataract.
- Fundus crystals.

Systemic features

- Spastic diplegia.
- Congenital ichthyosis.
- Mental handicap.
- Short stature.
- Speech defects.
- Short fingers and toes.

SLY SYNDROME (MPS VII)
– *see also* Mucopolysaccharidoses

Mode of inheritance

- Autosomal recessive in which there is a deficiency of β-glucuronidase. Gene mapped to chromosome 7.

Ocular features

- Corneal clouding.
- Pigmentary retinopathy.
- Glaucoma.

Systemic features

- Mental deficiency.
- Coarse facies.
- Flared lower ribcage.
- Hepatosplenomegaly.
- Macrocephaly.

Comment

- There is wide range of clinical severity, from mild with survival into adolescence, to severe with death in the neonatal period.

SPHINGOLIPIDOSES
These are a group of metabolic disorders caused by defects in sphingolipid metabolism. Sphingolipids commonly occur in cell membranes and myelin. This group includes:
- Gangliosidoses – G_{M1} and G_{M2} (Tay–Sachs and Sandhoff disease).
- Fabry disease.
- Gaucher disease.
- Niemann–Pick disease.

See individual conditions for details. Three other conditions which are also sphingolipidoses but have not been detailed in this section are metachromatic leukodystrophy, Krabbe disease, and Farber lipogranulomatosis.

SPOROTRICHOSIS
This is caused by the fungus *Sporothrix schenckii*.

Ocular features

- Parinaud oculoglandular syndrome.

Systemic features

- Erythematous, maculopapular lesion at the site of skin inoculation.
- Rarely, lung involvement.

STICKLER SYNDROME

Mode of inheritance

- Autosomal dominant. Mutations of type II collagen gene COL2AI are thought to be responsible, on chromosome 12q.

Ocular features

- High myopia.
- Lens subluxation.
- Glaucoma.
- Optically 'empty' vitreous.
- Retinal detachment.

Systemic features

- Midfacial hypoplasia.
- Robin sequence (Pierre Robin syndrome).
- Arthropathy.
- Cleft palate.
- Mitral valve prolapse.
- Deafness.

Comment

- Retinal detachment usually occurs after the second decade but may occur in childhood. Three-quarters of patients develop myopia by the age of 20 years.

STURGE–WEBER SYNDROME

Ocular features

- Glaucoma.
- Iris heterochromia.
- Hemangiomas of the episclera, iris, ciliary body, and choroid.

Systemic features

- Nevus flammeus.
- Ipsilateral leptomeningeal hemangioma with seizures.

Comment

- Nevus flammeus may occur in isolation, but the risk of neurologic and ocular complications rises if the area affected involves the ophthalmic division of the trigeminal nerve.

SULFITE OXIDASE DEFICIENCY

Mode of inheritance

- Autosomal recessive.

Ocular features

- Lens subluxation.

Systemic features

- Progressive muscular rigidity.
- Decerebrate posturing.
- Mental retardation.

Comment

- Most patients die by the age of 5 years.

SYSTEMIC LUPUS ERYTHEMATOSUS (SLE)

This is an autoimmune connective tissue disorder characterized by a widespread vasculitis.

Ocular features

- Keratoconjunctivitis sicca.
- Peripheral corneal ulceration.
- Retinopathy.

Systemic features

- Pyrexia.
- Painful but clinically normal-looking joints.
- 'Butterfly' facial rash.
- Cardiac involvement – pericarditis.
- Renal dysfunction.
- CNS involvement.

Comment

- ANA positive: look for anti-dsDNA, which is specific for SLE.

SYSTEMIC SCLEROSIS

Ocular features

- Enophthalmos.
- Cicatrizing conjunctivitis.

Systemic features

- Raynaud phenomenon.
- Scleroderma – thickening of the skin, with a waxy appearance.
- Fibrotic changes under the skin, causing binding-down of facial skin and tapering of digits.
- Cutaneous calcinosis.

TANGIER DISEASE

Mode of inheritance

- Autosomal recessive condition in which there is an absence of HDL (high-density lipoprotein) and an accumulation of cholesterol esters in the tissues.

Ocular features

- Stromal corneal crystals.
- Vortex keratopathy.

Systemic features

- Enlarged, orange tonsils.
- Splenomegaly.
- Neuropathy.
- Facial diplegia.

Comment

- Corneal findings are usually evident between 20 and 40 years of age.

TAKAYASU DISEASE

This is a life-threatening, idiopathic, large-vessel vasculitis which mainly affects young Asian women.

Ocular features

- Slow-flow retinopathy.
- Ocular ischemic syndrome.

Systemic features

- Involvement of the aorta and its major branches – hence, this condition is also called 'pulseless disease'.

TAY–SACHS DISEASE, CLASSIC
(gangliosidosis G_{M2}, type I)

Mode of inheritance

- Autosomal recessive caused by severe deficiency of hexosaminidase A which is almost exclusively seen in Ashkenazi Jews.

Ocular features

- Cherry-red spot at the macula in virtually all patients.
- Optic atrophy.

Systemic features

- Motor weakness evident within the first 6 months of life.
- Progressive symptoms of deafness, blindness, and seizures after the age of 18 months.

Comment

- Death usually occurs by the age of 2–3 years. Optic atrophy is usually seen after 1 year of age, but cherry-red spot may be seen within 2 months of life.

TAY–SACHS DISEASE, JUVENILE
(gangliosidosis G_{M2}, type III)

Mode of inheritance

- Autosomal recessive with only partial deficiency of hexosaminidase A.

Ocular features/Systemic features

- Onset is later than in the classic form, occurring between 2 and 6 years of age, and is followed by progressive blindness. Death usually occurs by the second decade. Features are similar to those of the classic-type disease.

Comment

- There is an adult-onset form of hexosaminidase A deficiency in which there is progressive deterioration of gait and posture; vision and ocular fundi remain normal.

TEMPLE–AL GAZALI SYNDROME
(Midas syndrome)

Ocular features

- Microphthalmos.
- Sclerocornea.

Systemic features

- Linear irregular areas of dermal aplasia, especially of the head and neck.

TERSON SYNDROME
This is the association of spontaneous subarachnoid hemorrhage and subhyaloid hemorrhage.

TORRE SYNDROME

Ocular features

- Multiple sebaceous adenomas on the eyelids.
- Other multiple eyelid lesions, e.g. inclusion cysts.

Systemic features

- Visceral carcinomas, especially of the colon.
- Cutaneous sebaceous adenomas and other lesions, e.g. squamous cell carcinoma, keratoacanthoma.

TOXIC EPIDERMAL NECROLYSIS
(Lyell disease)
This is a staphylococcal infection in which exfoliatin toxin is released causing separation of epidermal cells.

Ocular features

- Cicatrizing conjunctivitis.

Systemic features

- Skin lesions resemble 'scalded skin'.

TREACHER COLLINS SYNDROME

Mode of inheritance

- Autosomal dominant with most cases probably caused by fresh mutations of a gene on chromosome 5q.

Ocular features

- Coloboma of outer third of lower lid.
- Downslanting palpebral fissures.
- Partial or total absence of lower eyelashes.
- Strabismus and/or amblyopia.

Systemic features

- External ear anomalies.
- Malar hypoplasia with or without zygomatic arch hypoplasia.
- Mandibular hypoplasia.
- Cleft palate.
- Conductive deafness.

TRISOMY 13 – *see* Patau syndrome

TRISOMY 18 – *see* Edward syndrome

TUBEROUS SCLEROSIS (Bourneville disease)

Mode of inheritance

- Autosomal dominant, but over three-quarters of cases are caused by fresh mutations. Two genetic loci have been identified, on 9q and 16p, respectively.

Ocular features

- Retinal astrocytomas in over 50% of cases.

Systemic features

- Adenoma sebaceum (fibroangiomatous skin lesions), seen in 50% of cases by 5 years of age.
- Ash-leaf spots (or other-shaped white macules), usually present at birth.
- Café-au-lait spots.
- Hamartomas of the brain and seizures.
- Cystic bone lesions.
- Other hamartomas – subungual, visceral.

TULAREMIA

This is a zoonosis due to the Gram-negative organism *Francisella tularensis*. The infection enters via inoculation through the skin or conjunctivae.

Ocular features

- Parinaud oculoglandular fever.

Systemic features

- Ulcerating cutaneous papule with tender suppurative lymphadenopathy.
- Pneumonia.
- Septicemia with shock.

TURNER SYNDROME

Mode of inheritance

- This occurs in an XO patient with 45 chromosomes.

Ocular features

- Ptosis.
- Epicanthic folds.
- Microcornea.
- Keratoconus.
- Blue sclera.
- Cataract.
- Conjunctival lymphangiectasia.

Systemic features

- Short female.
- Ovarian dysgenesis.
- Congenital lymphedema.
- Broad chest with widely spaced nipples.
- Short neck with posterior webbing.
- Multiple pigmented nevi.
- Renal anomalies, e.g. horseshoe kidney.

TYROSINEMIA TYPE II (essential tyrosinemia, Richner–Hanhart syndrome)

Mode of inheritance

- Autosomal recessive condition due to deficiency of soluble tyrosine aminotransferase, giving rise to raised serum tyrosine levels with excretion of tyrosine in the urine.

Ocular features

- Corneal pseudodendrites with crytalline edges.
- Conjunctival thickening.
- Vortex-like keratopathy.
- Nystagmus.
- Strabismus.

Systemic features

- Palmar keratosis.
- Severe psychomotor retardation.

Comment

- Type I tyrosinemia is usually fatal due to hepatic cirrhosis and renal tubular failure, unless there is early intervention with dietary restriction of tyrosine and phenylalanine. There are no ocular abnormalities in this type.

USHER SYNDROME

This term encompasses a group of genetic disorders predisposing to visual loss and deafness. There are several types:

1. Usher I

Ocular features

- Retinitis-pigmentosa-like retinopathy.

Systemic features

- Congenital, severe, profound hearing loss with absent vestibular function.
- Three subtypes.

2. Usher II

Ocular features

- Retinitis-pigmentosa-like retinopathy.

Systemic features

- Congenital, moderate to severe hearing loss with normal vestibular function.
- Two subtypes.

3. Usher III

Ocular features

- Retinitis-pigmentosa-like retinopathy.

Systemic features

- Progressive hearing loss.

VON HIPPEL–LINDAU SYNDROME

Mode of inheritance

- Autosomal dominant, with locus for gene defect on 3q.

Ocular features

- Retinal capillary haemangiomas, usually not evident until the third decade.

Systemic features

- Cerebellar and spinal cord hemangioblastoma.
- Pheochromocytoma.
- Hypernephroma.
- Visceral cysts.
- Polycythemia.

WAARDENBURG SYNDROME

There are three types of this syndrome. Types I and II are autosomal dominant, but the mode of inheritance of type III is unclear. The gene defect for type I is on 2q, while that for type II is on 3q.

Ocular features

- Telecanthus.
- Hypochromic irides.
- Heterochromia irides in some cases.
- Glaucoma.
- Synophrys.
- Poliosis.
- Fundus hypopigmentation.
- Microcornea.
- Short palpebral fissures.

Systemic features

- Deafness.
- Partial albinism – white forelock with premature graying.

Comment

- Types I and III are similar, but type III also includes upper limb defects, flexion contractures, and syndactyly. Types I and II differ in that only the former has telecanthus.

WALDENSTRÖM MACROGLOBULINEMIA

Ocular features

- Slow-flow retinopathy.
- Cotton-wool spots.
- Bilateral orbital pseudotumor.

Systemic features

- Lymphadenopathy.
- Hyperviscosity syndrome.
- Bone marrow infiltration symptoms, e.g. anemia, easily bruised.

Comment

- Most frequently affects elderly men.
- Look for lymphoplasmacytoid cells on bone marrow biopsy, IgM paraproteinemia, pancytopenia, or raised ESR.

WARBURG SYNDROME
(Walker–Warburg syndrome)

Mode of inheritance

- Autosomal recessive.

Ocular features

- Microphthalmos.
- Cataract.
- Retinal dysplasia.
- Uveal coloboma.
- Corneal clouding secondary to Peters anomaly.

Systemic features

- Deafness.
- Klippel–Feil anomaly.
- Preauricular skin tags.
- Low hairline.

WILSON DISEASE
(hepatolenticular degeneration)

Mode of inheritance

- Autosomal recessive disorder located to 13q.

Ocular features

- Kayser–Fleischer corneal ring, especially seen in patients with neurologic complications.
- Sunflower cataract.

Systemic features

- Chronic liver disease with cirrhosis.
- Neurologic signs caused by basal ganglia involvement, e.g. dysarthria, tremor, involuntary movements.
- Premature dementia.

Comment

- Look for reduced serum copper and ceruloplasmin, with increased urinary copper.

WOLFRAM SYNDROME (D.I.D.M.O.A.D.)

Mode of inheritance

- Autosomal recessive disorder localized to chromosome 4.

Ocular features/Systemic features

- Diabetes Insipidus.
- Diabetes Mellitus.
- Optic Atrophy.
- Deafness.

XERODERMA PIGMENTOSUM

Mode of inheritance

- Autosomal recessive condition in which there is defective repair mechanism of DNA damaged by UV light.

Ocular features/Systemic features

- Increased incidence of cutaneous basal cell carcinoma, squamous cell carcinoma, and melanoma.

ZELLWEGER SYNDROME
(hepatocerebrorenal syndrome)

Mode of inheritance

- Autosomal recessive condition in which the cellular hallmark is failure to assemble normal peroxisomes. Two loci have been identified: one at 7q and the other at 1p.

Ocular features

- Congenital cataract.
- Optic disc hypoplasia and pallor.
- Pigmentary retinopathy.
- Corneal clouding.

Systemic features

- CNS abnormalities.
- Hepatomegaly.
- Large fontanelles.
- Renal cysts.
- Dysmorphic features.
- Absent liver peroxisomes.

Comment

- The majority of affected children die by the end of the first year.

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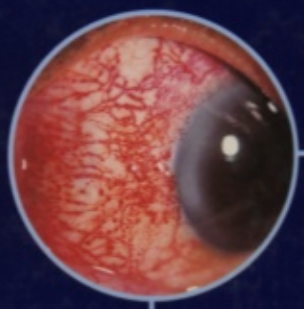
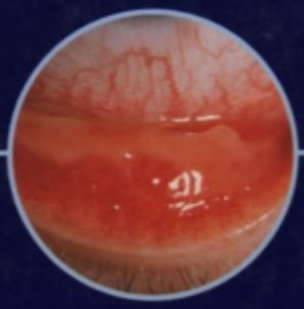
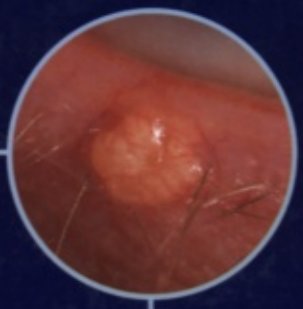
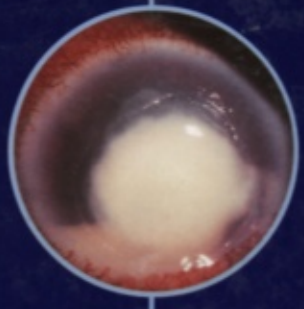


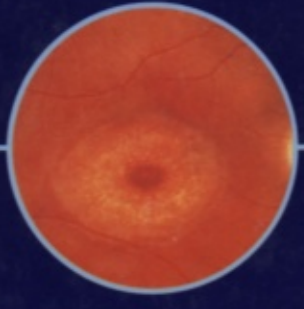
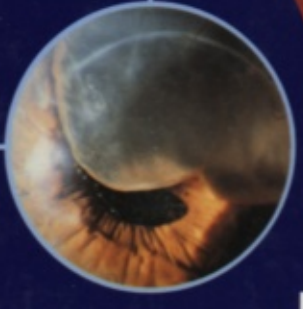

Ophthalmology: Clinical Signs and Differential Diagnosis presents a comprehensive, fully illustrated visual catalog of clinical signs in ophthalmology, organized by region.

The book is lavishly illustrated with over 1500 superb colour photographs and 50 angiograms of the highest quality. The text is concise and focuses on the information needed to make a quick and accurate diagnosis: the signs, ocular and systemic associations, causes and other pertinent information. The book has been carefully structured to provide immediate access to key information. Summary tables list systemic associations and explain the diagnostic differences between clinical signs. Additionally an appendix summarizes the salient genetic and clinical features of all the uncommon systemic disorders mentioned in the text.

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