CONGENITAL ANOMALIES IN THE EYE

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3rd YEAR 6th SEMESTER

TODAY WE ARE DISCUSSING ANOMALIES OF……..

- EYELID
- CORNEA
- LENS
- UVEAL TRACT
- RETINA AND CHOROID
- LACRIMAL APPARTUS
- ORBIT

EYELID

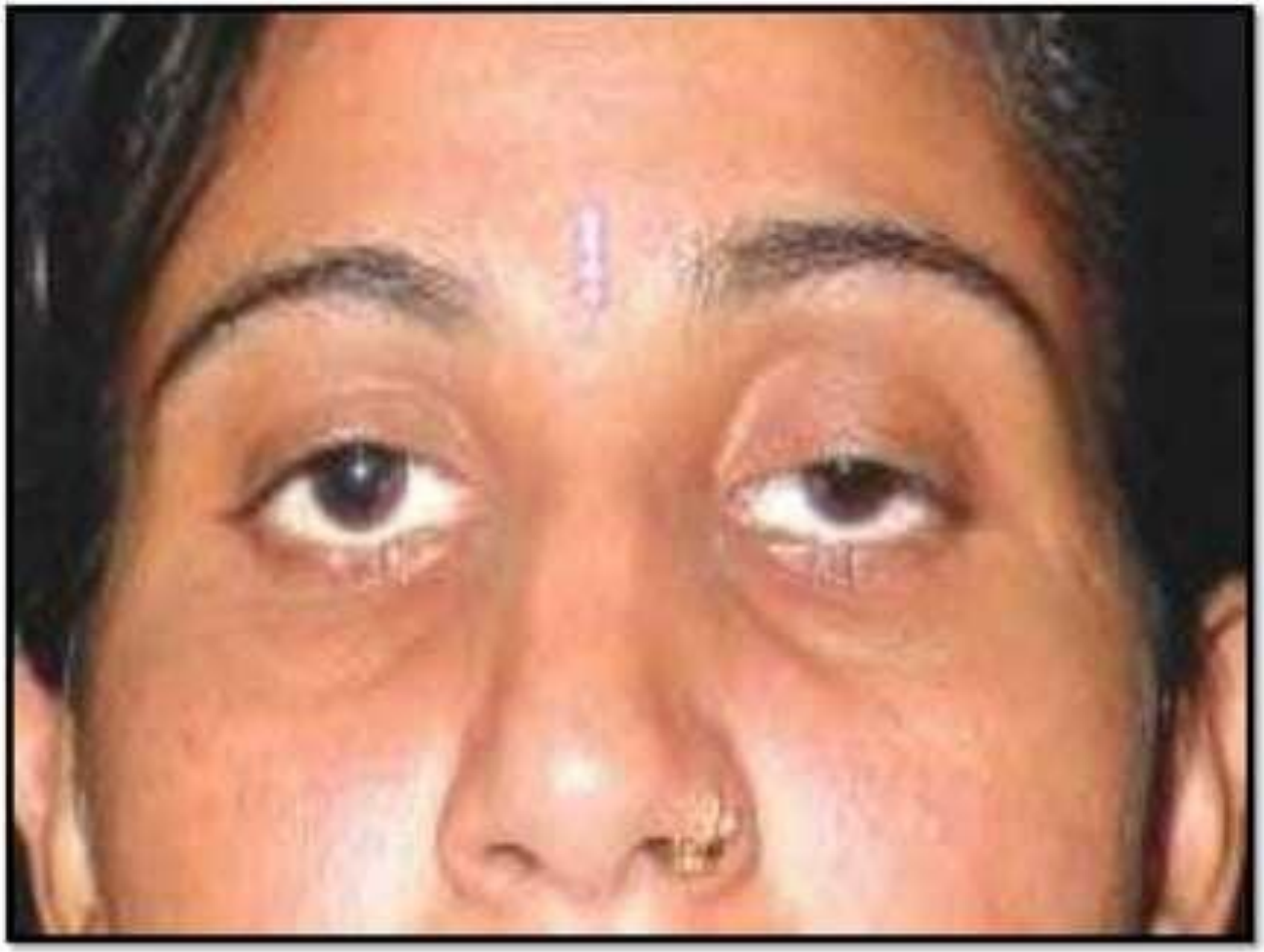
- Congenital ptosis
- Congenital coloboma
- Epicanthus
- Telecanthus
- Epiblepharon
- Microblepharon
- Distichiasis
- euryblepharon

CONGENITAL PTOSIS

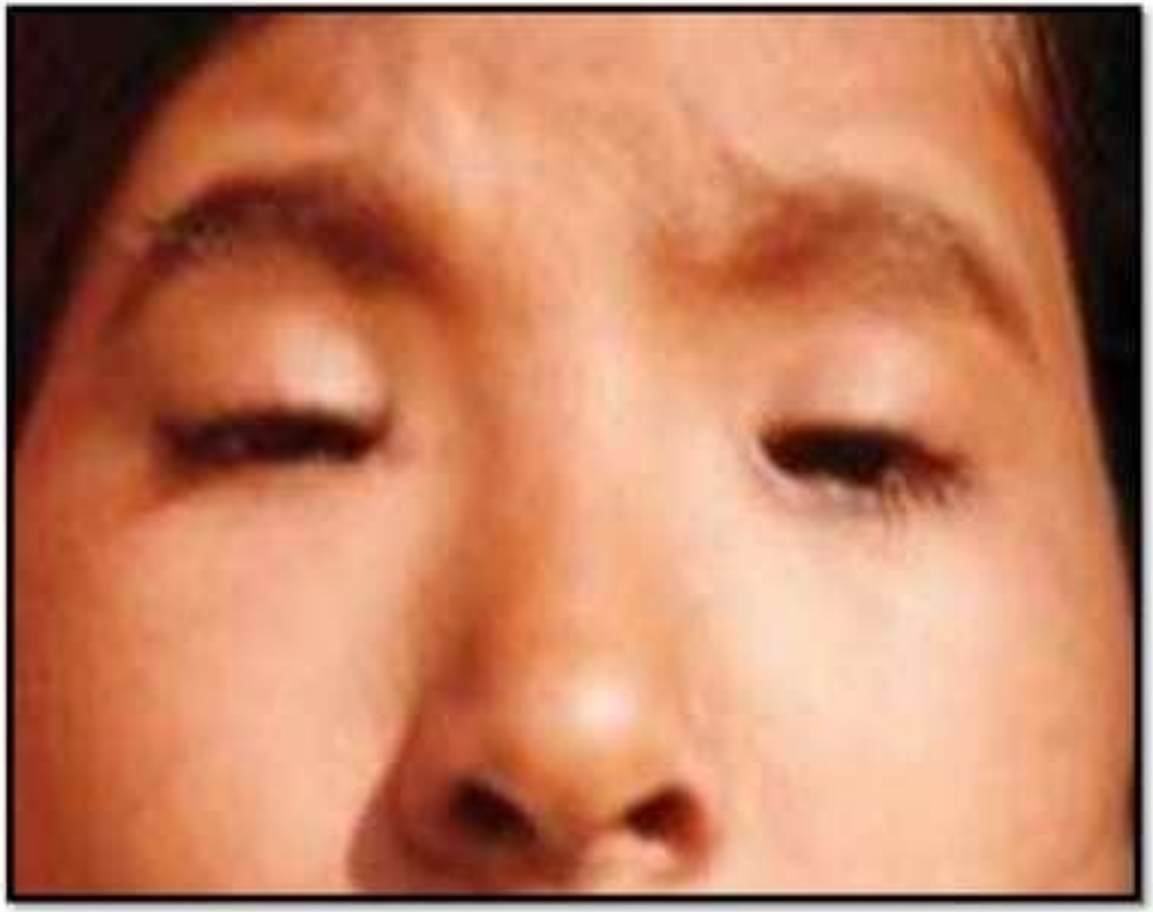
- It is a common congenital anomaly.
- It is associated with congenital weakness (maldevelopment) of the levator palpebrae superioris (LPS).

- ❑ *Simple congenital ptosis (not associated with* any other anomaly).
- ❑ Congenital ptosis with associated weakness of superior rectus muscle.
- ❑ As a part of *blepharophimosis syndrome, which* comprises congenital ptosis, blepharophimosis, telecanthus and Epicanthus inversus.
- ❑ *Congenital synkinetic ptosis (Marcus Gunn jawwinking* ptosis).
- In this condition there occurs retraction of the ptotic lid with jaw movements,

SIMPLE CONGENITAL PTOSIS

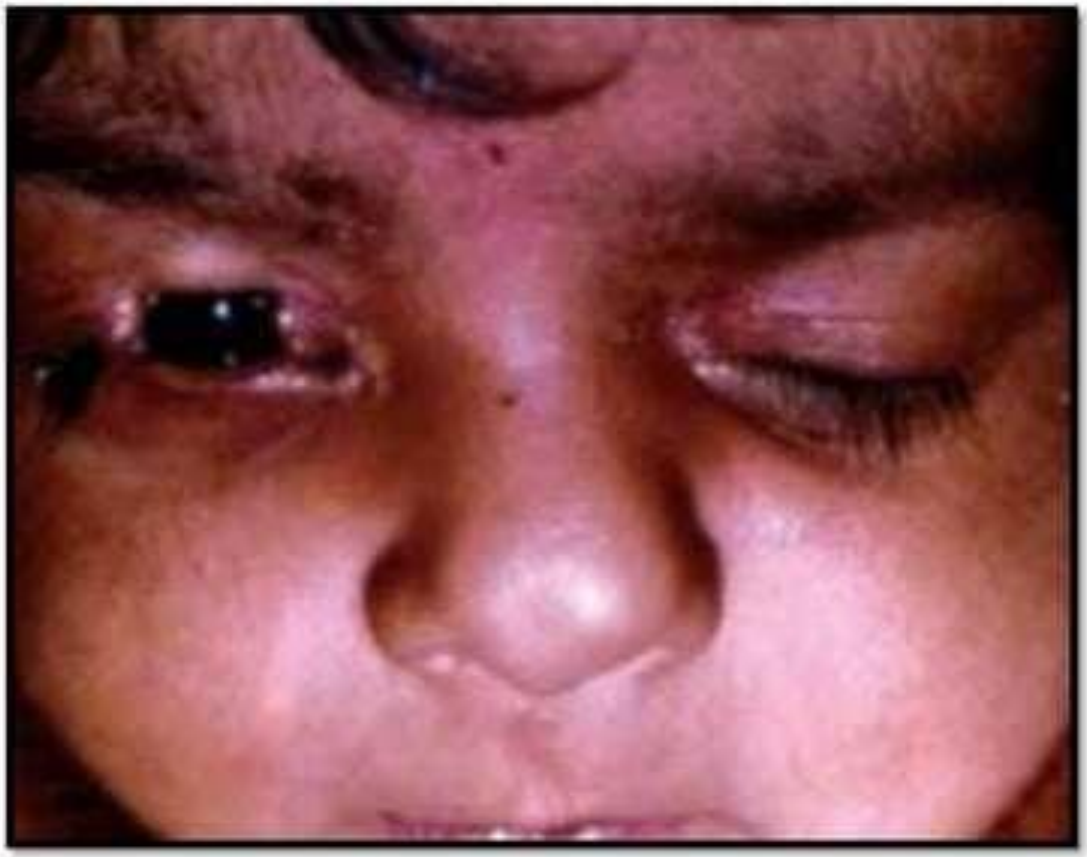


BLEPHAROPHIMOSIS SYNDROME



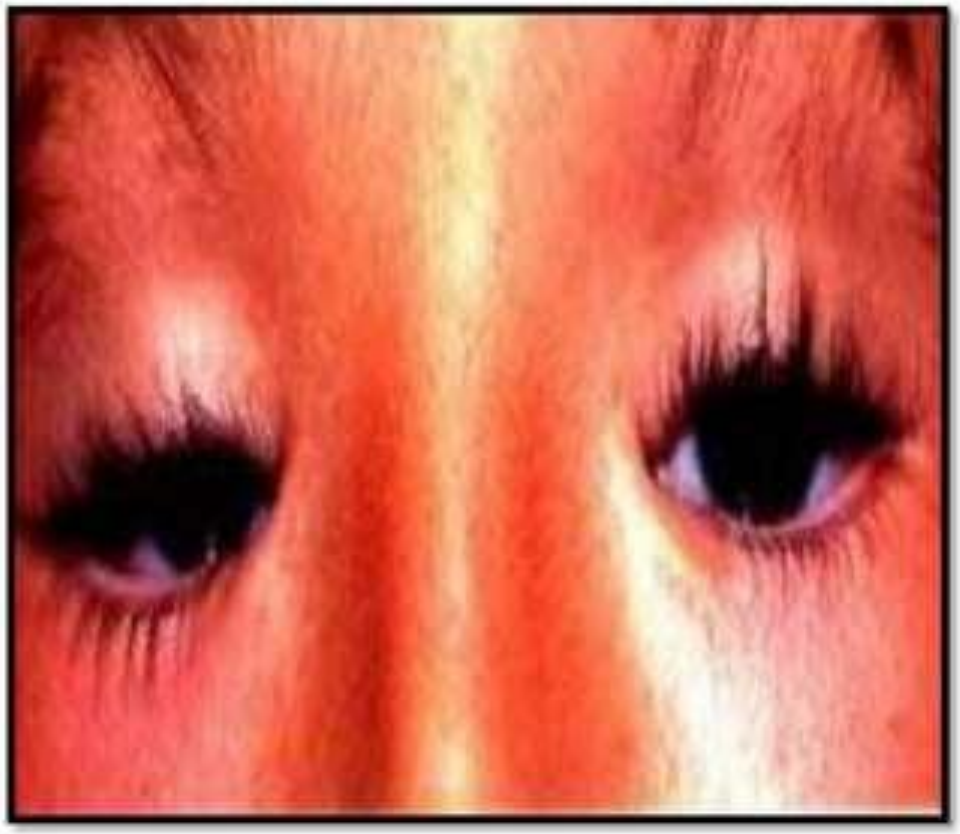
CONGENITAL COLOBOMA

- It is a rare condition characterised by a full thickness triangular gap in the tissues of the lids.
- The anomaly usually occurs near the nasal side and involves the upper lid more frequently than the lower lid.



EPICANTHUS

- It is a semicircular fold of skin which covers the medial canthus.
- It is a bilateral condition and may disappear with the development of nose.
- It is a normal facial feature in Mongolian races.
- It is the most common congenital anomaly of the lids.
- THEY MAY GIVE RISE TO PSEUDO ESOTROPIA..

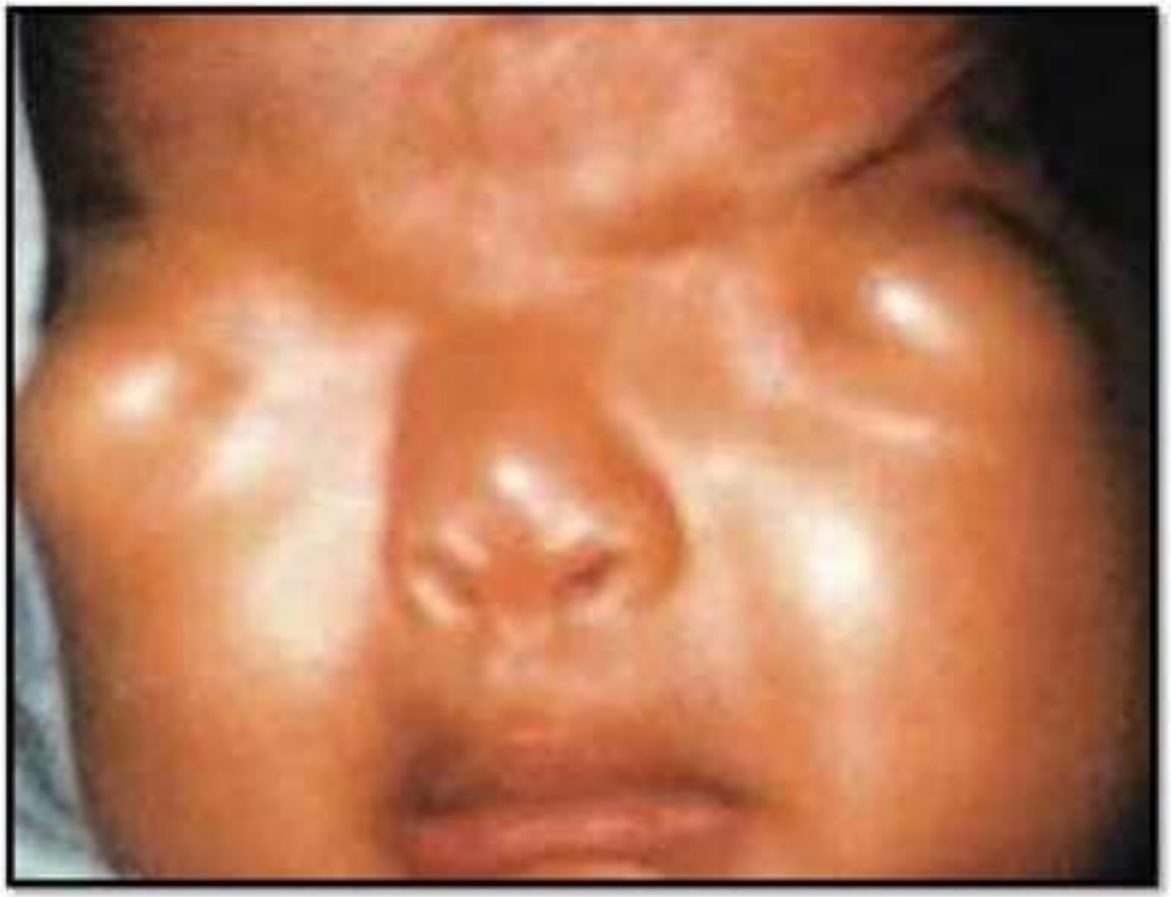


DISTICHIASIS

- *Congenital distichiasis is a rare* anomaly in which an extra row of cilia occupies the position of Meibomian glands which open into their follicles as ordinary sebaceous glands.
- These cilia are usually directed backwards and when rubbing the cornea.

CRYPTOPHTHALMOS

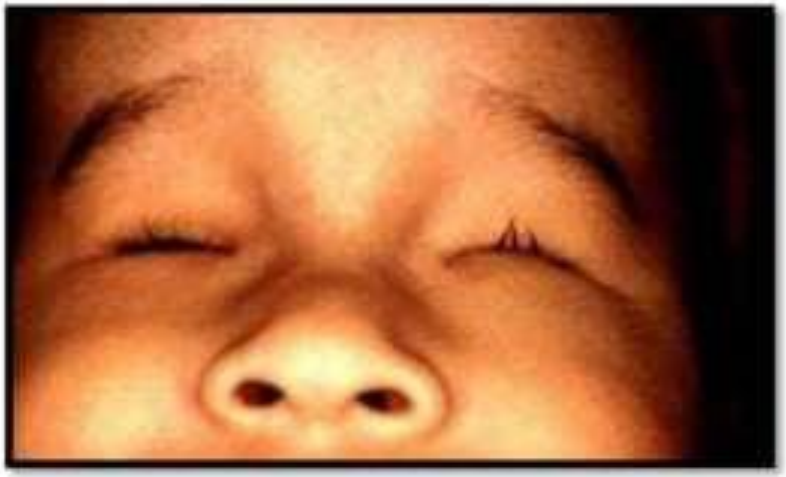
- It is a very rare anomaly in which lids fail to develop and the skin passes continuously from the eyebrow to the cheek hiding the eyeball.



MICROBLEPHARON

- In this condition, eyelids are abnormally small.
- It is usually associated with microphthalmos or anophthalmos.
- Occasionally the lids may be very small or virtually absent and the condition is called *ablepharon*.

ANOPHTHALMOS



TELECANTHUS

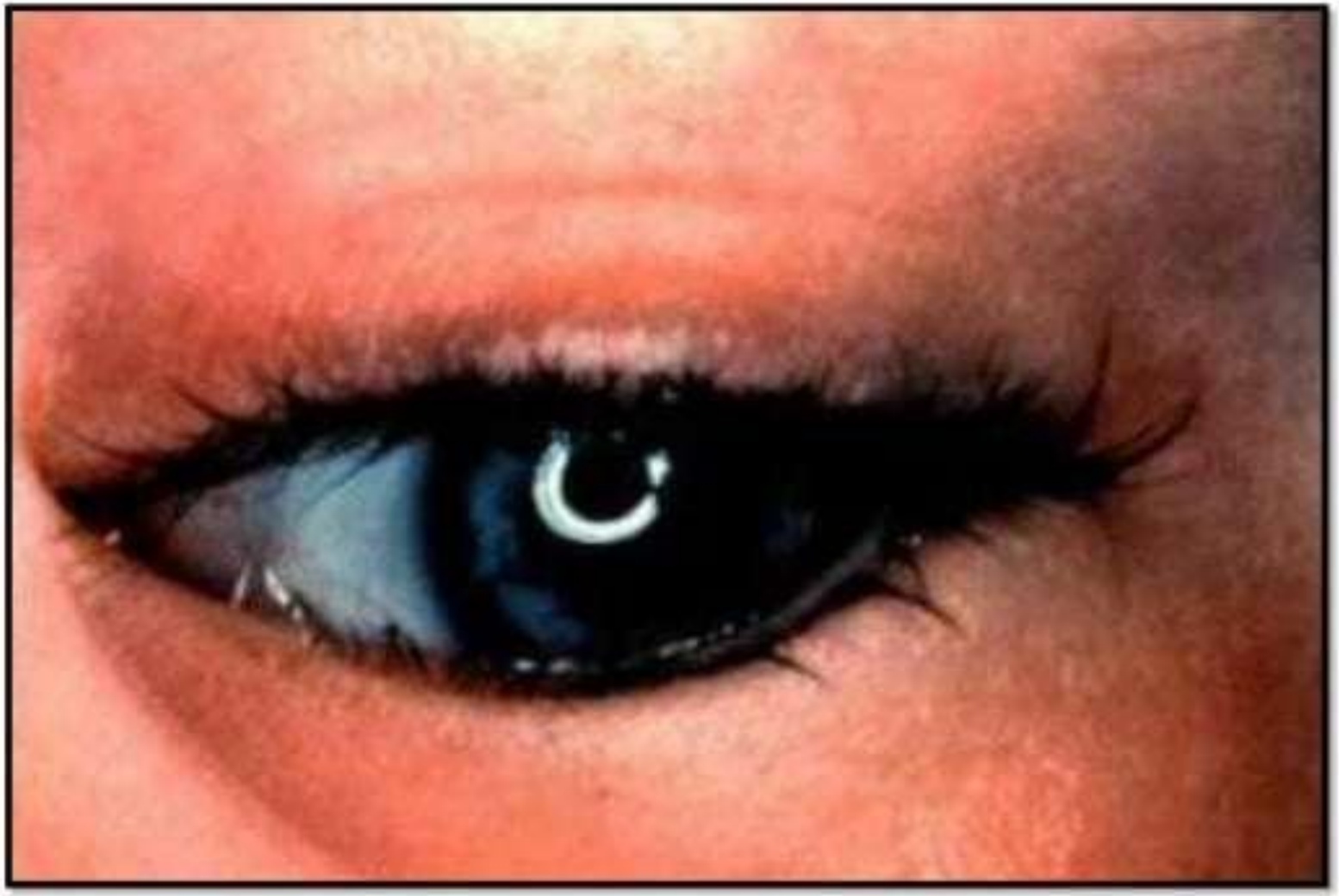
- It is an uncommon condition that may occur in isolation or an association with blepharophimosis syndrome.

EPIBLEPHARON

- Refers to a congenital anomaly in which a horizontal fold of tissue rides above the lower lid margin.
- It usually disappear with the growth of face and needs no surgical correction.

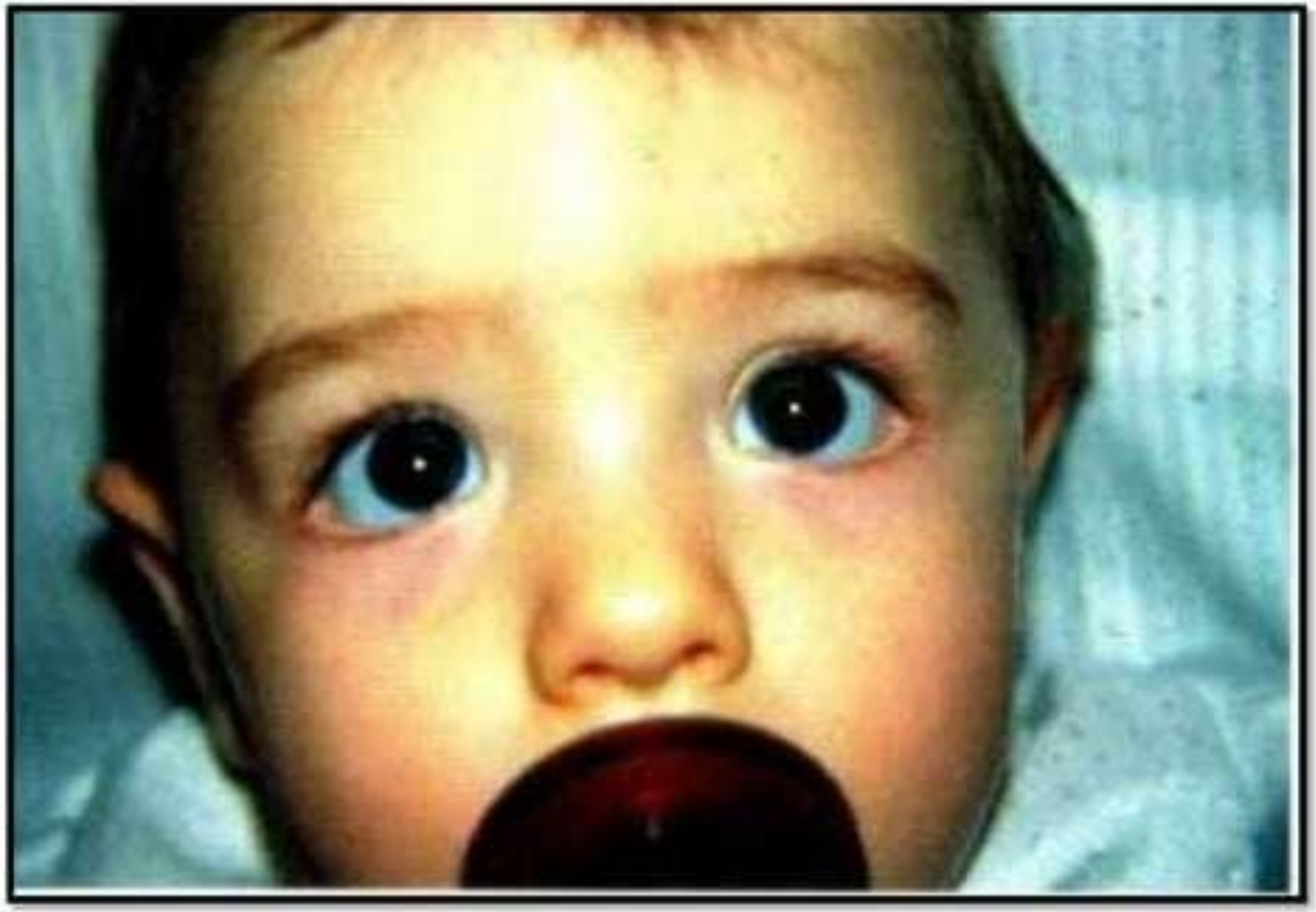
CONGENITAL ENTROPION

- It is a rare condition seen since birth.
- It may be associated with microphthalmos.



EURYBLEPHARON

- It refers to unilateral or bilateral horizontal widening of palpebral fissure.
- Usually associated with lateral canthal malposition and lateral ectropion.



CORNEA

- ✓Megalocornea
- ✓Microcornea
- ✓Corneal plana
- ✓Cloudy cornea

MEGALOCORNEA

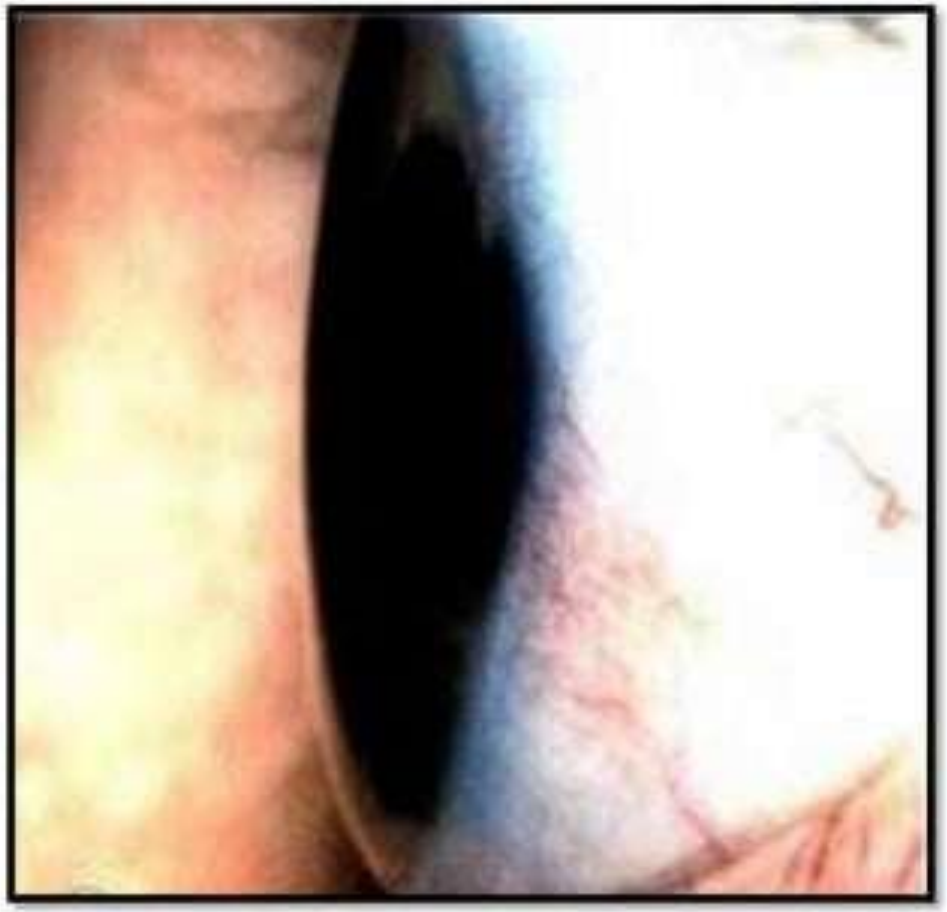
- Horizontal diameter of cornea at birth is about 10 mm and the adult size of about 11.7 mm is attained by the age of 2 years.
- Megalocornea is labelled when the horizontal diameter of cornea is of adult size at birth or 13 mm or greater after the age of 2 years.
- The cornea is usually clear with normal thickness and vision.
- The condition is not progressive.

MICROCORNEA

- In microcornea, the horizontal diameter is less than 10 mm since birth.
- The condition may occur as an isolated anomaly (rarely) or in association with *nanophthalmos (normal small eyeball) or microphthalmos (abnormal small eyeball).*

CORNEA PLANA

- This is a rare anomaly in which bilaterally cornea is comparatively flat since birth. It may be associated with microcornea.
- Cornea plana usually results in marked astigmatic refractive error.

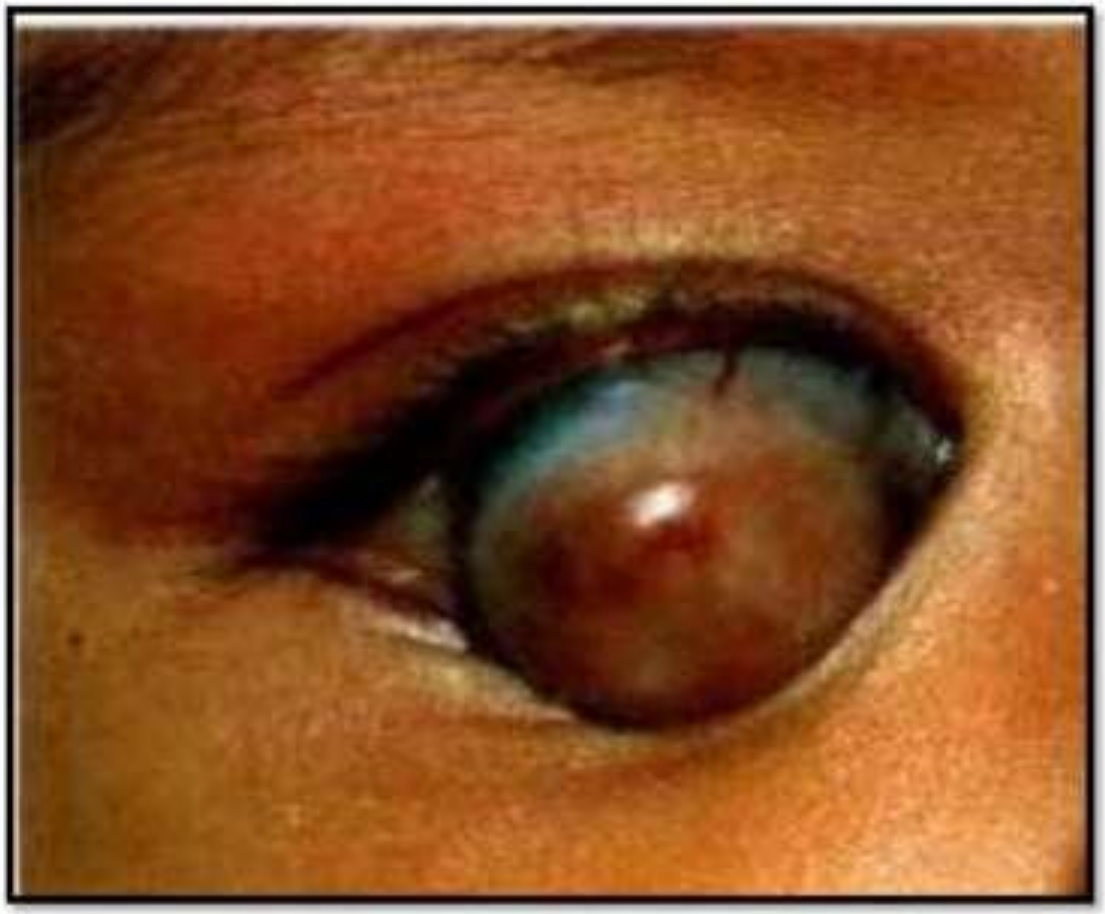


CONGENITAL CLOUDY CORNEA

- Sclerocornea
- Tears in Descemet's membrane
- Ulcer
- Metabolic conditions
- Posterior corneal defect
- Endothelial dystrophy
- Dermoid

KERACTASIA

- It is a very rare usually unilateral condition thought to be the result of intrauterine keratitis and perforation.
- It is often associated with high IOP.



UVEAL TRACT

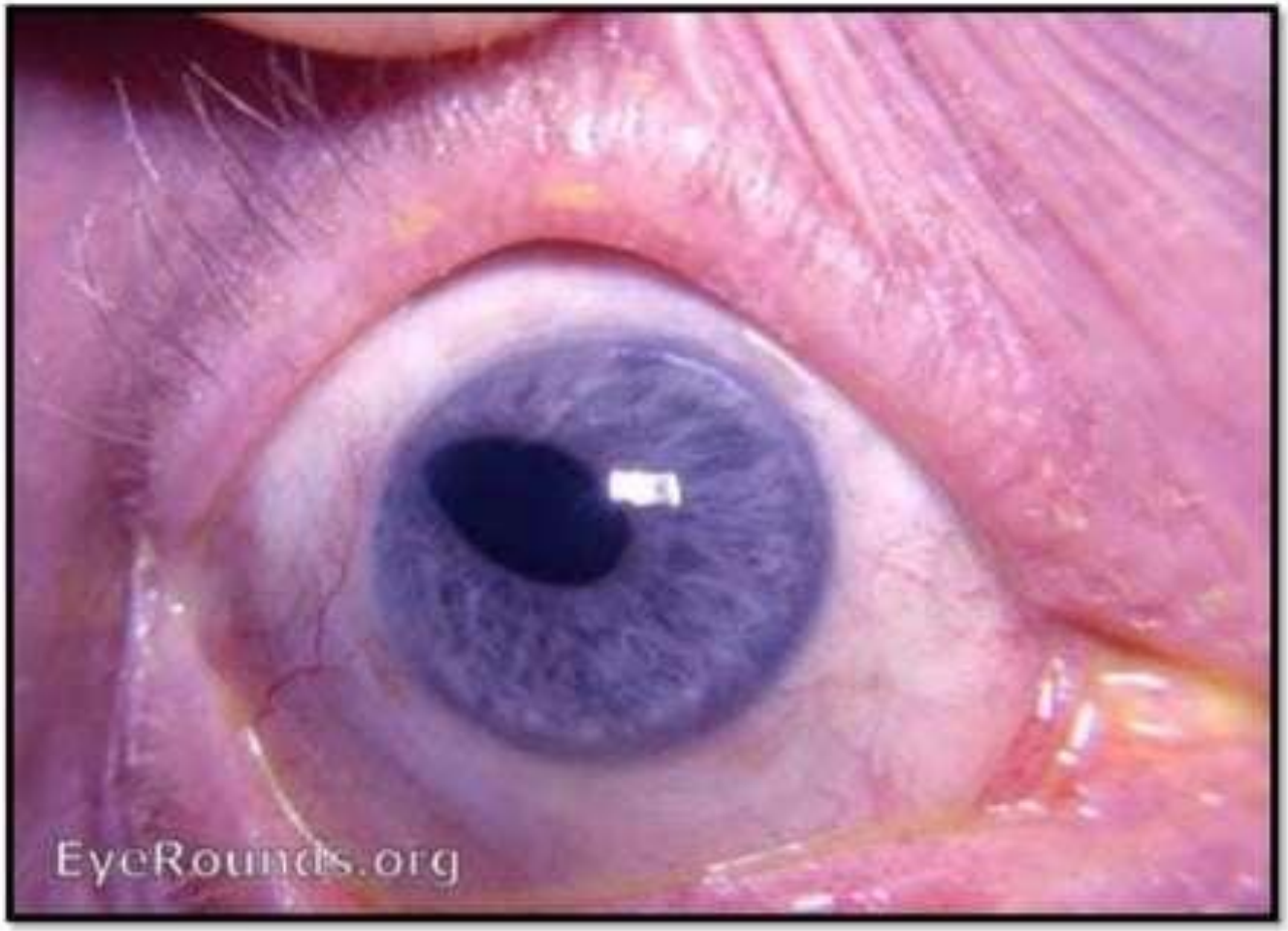
- CORECTOPIA
- ANIRIDIA
- POLYCORIA
- PERSISTENT PUPILLARY MEMBRANE
- CONGENITAL COLOBOMA OF THE UVEAL TRACT

HETEROCHROMIA OF IRIS

- It refers to variations in the iris colour and is a common congenital anomaly.
- In *heterochromia iridium colour* of one iris differs from the other.
- Sometimes, one sector of the iris may differ from the remainder of iris; such a condition is called *heterochromia iridis*

CORECTOPIA

- It refers to abnormally eccentric placed pupil.
- Normally pupil is placed slightly nasal to the centre.

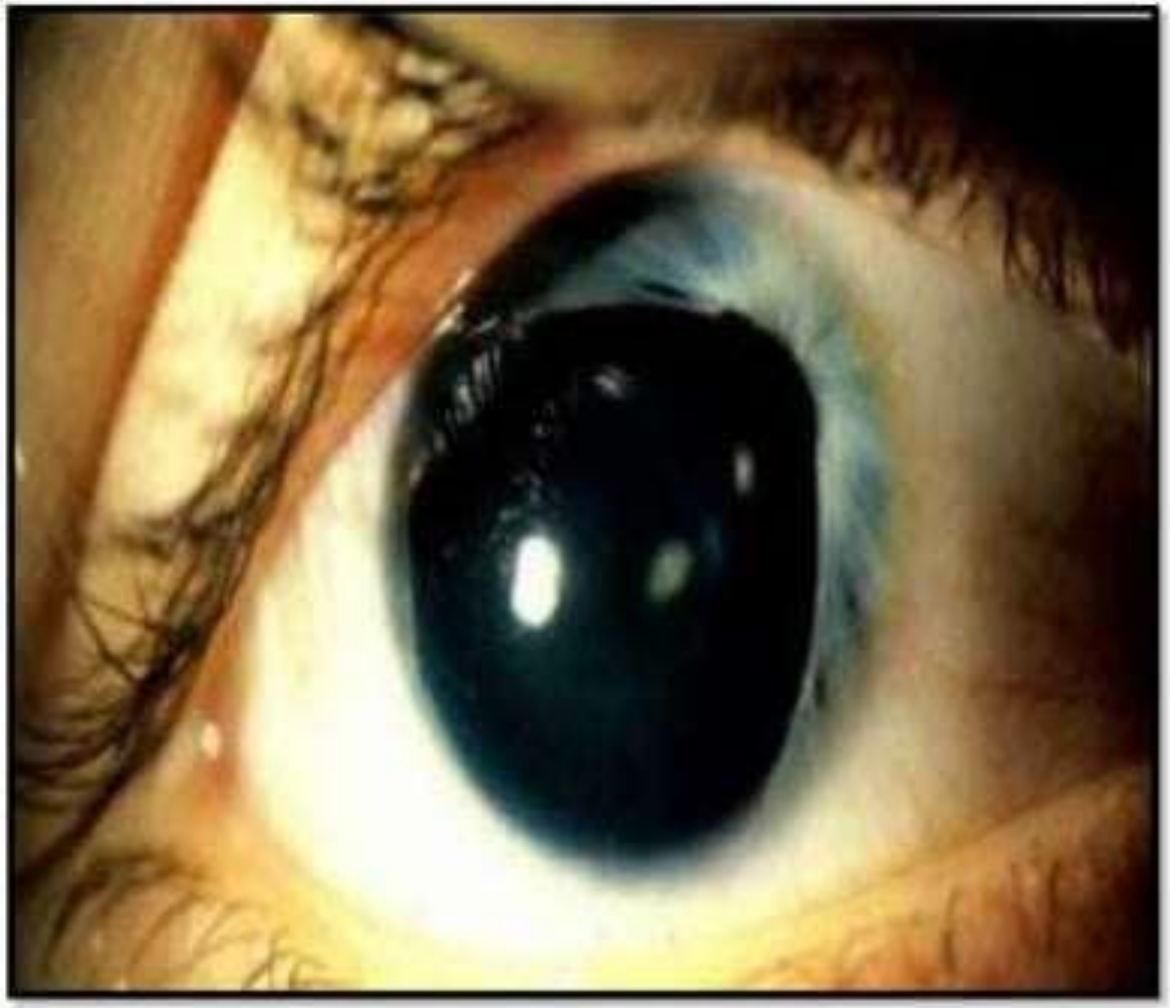

EyeRounds.org

POLYCORIA

- In this condition, there are more than one pupil.

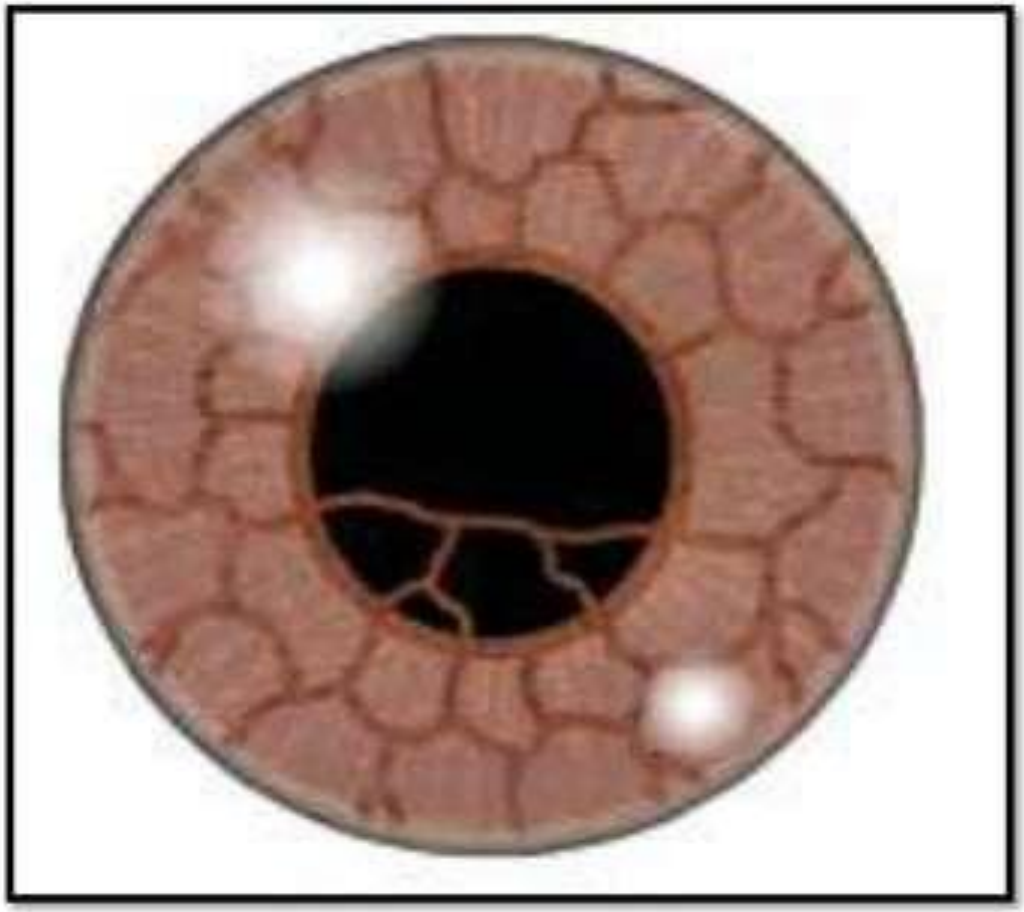
CONGENITAL ANIRIDIA

- It refers to congenital absence of iris. *True aniridia,* i.e., complete absence of the iris is extremely rare.
- Usually, a peripheral rim of iris is present and this condition is called '*Clinical aniridia*'.



PERSISTENT PUPILLARY MEMBRANE

- It represents the remnants of the vascular sheath of the lens.
- It is characterised by stellate-shaped shreds of the pigmented tissue coming from anterior surface of the iris (attached at collarette)

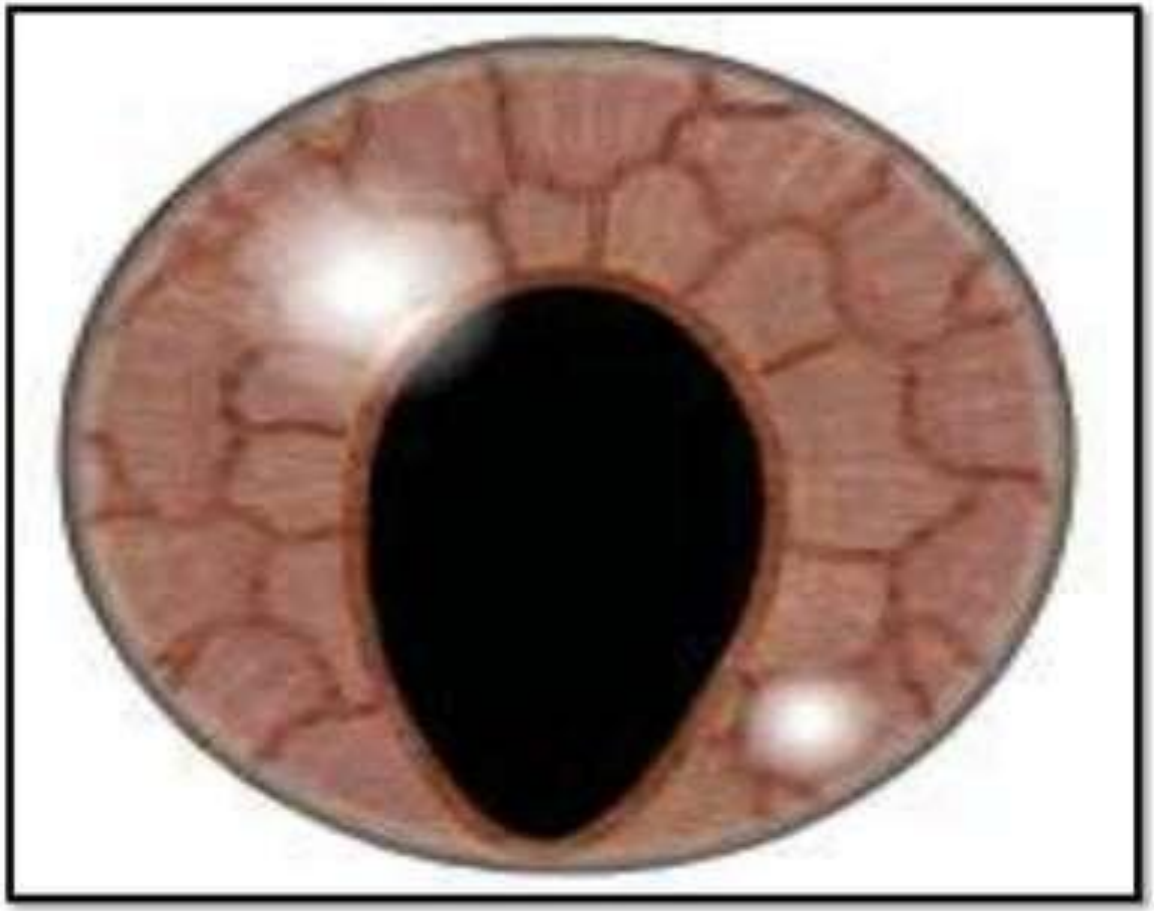


CONGENITAL COLOBOMA OF THE UVEAL TRACT

- Congenital coloboma (absence of tissue) of iris, ciliary body and choroid may be seen in association or independently.
- Coloboma may be typical or atypical

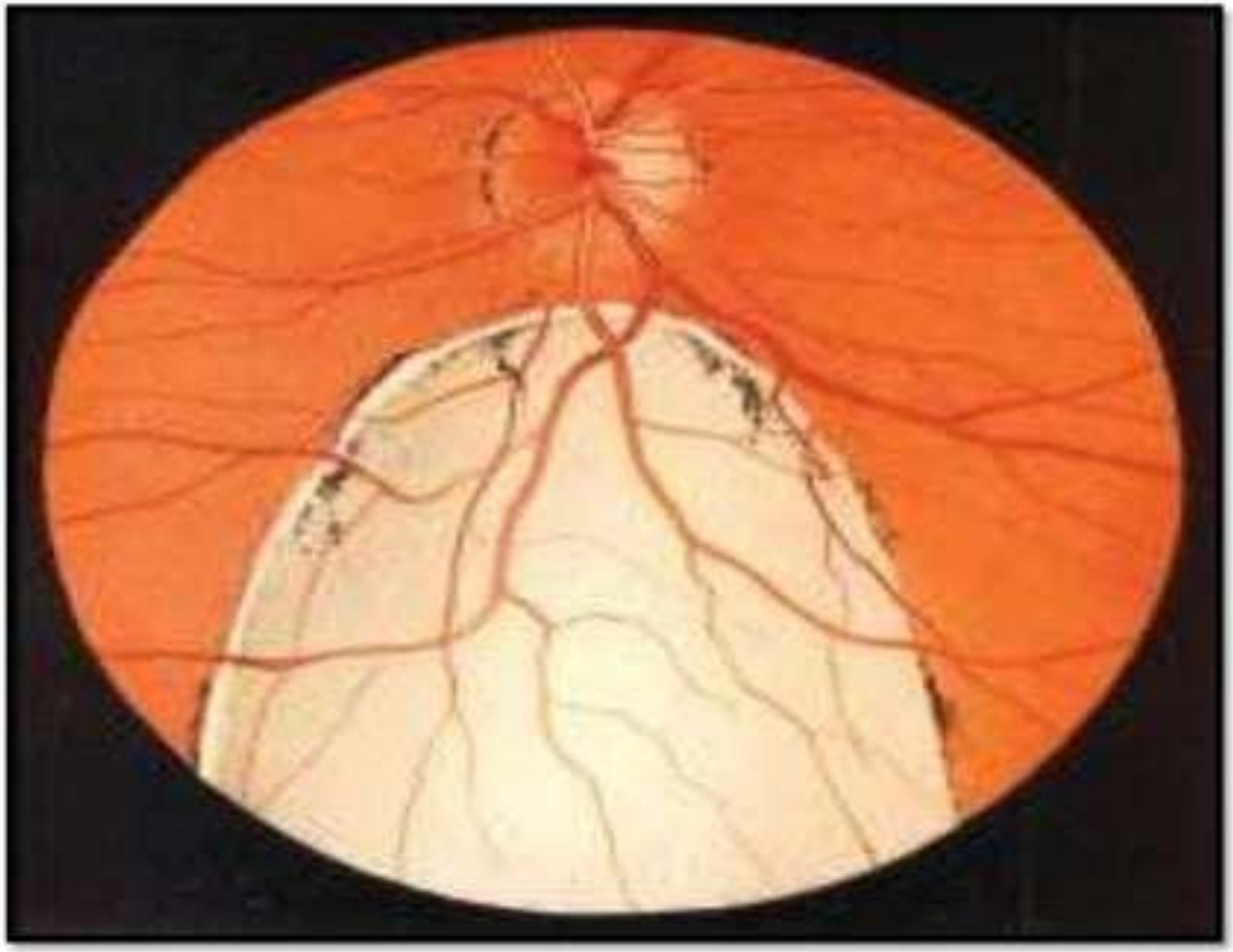
TYPICAL COLOBOMA

- *Typical coloboma is seen in the inferonasal* quadrant and occurs due to defective closure of the embryonic fissure.



ATYPICAL COLOBOMA

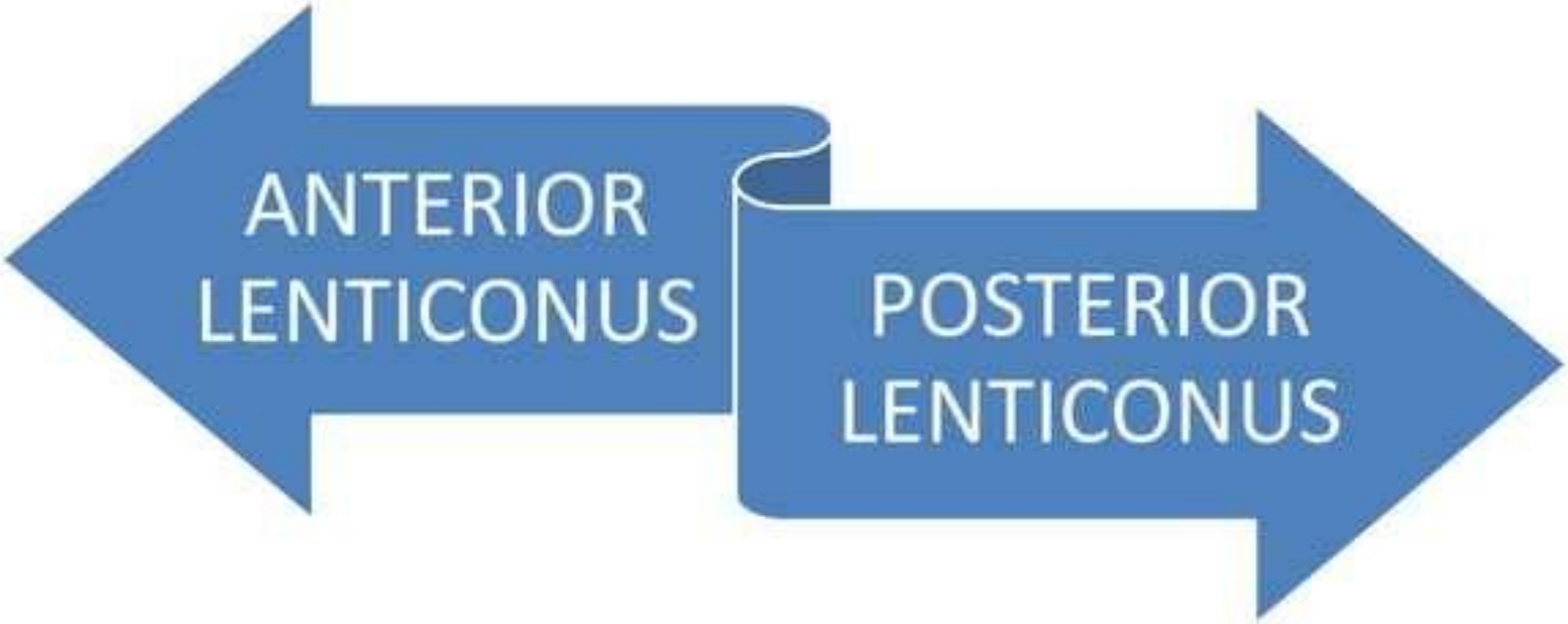
- It is occasionally found in other position.
- It is usually incomplete.



CRYSTALLINE LENS

- LENTICONUS
- LENTIGLOBUS
- COLOBOMA

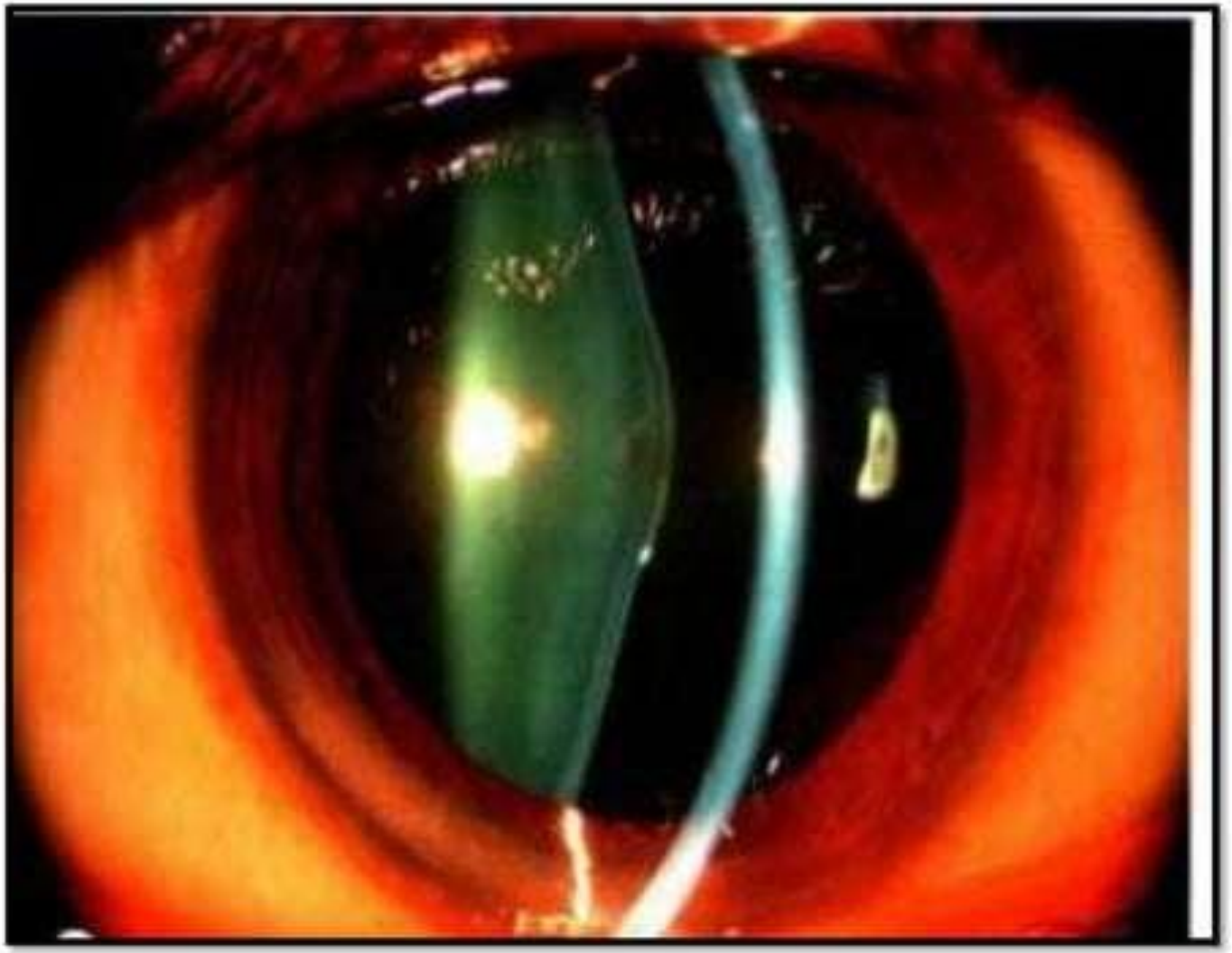
LENTICONUS



ANTERIOR LENTICONUS

1. SIGN : Bilateral axial projection of the anterior surface of lens into the anterior chamber.
2. ASSOCIATION. About 90% of patient have Alport syndrome which may be associated with cataract.

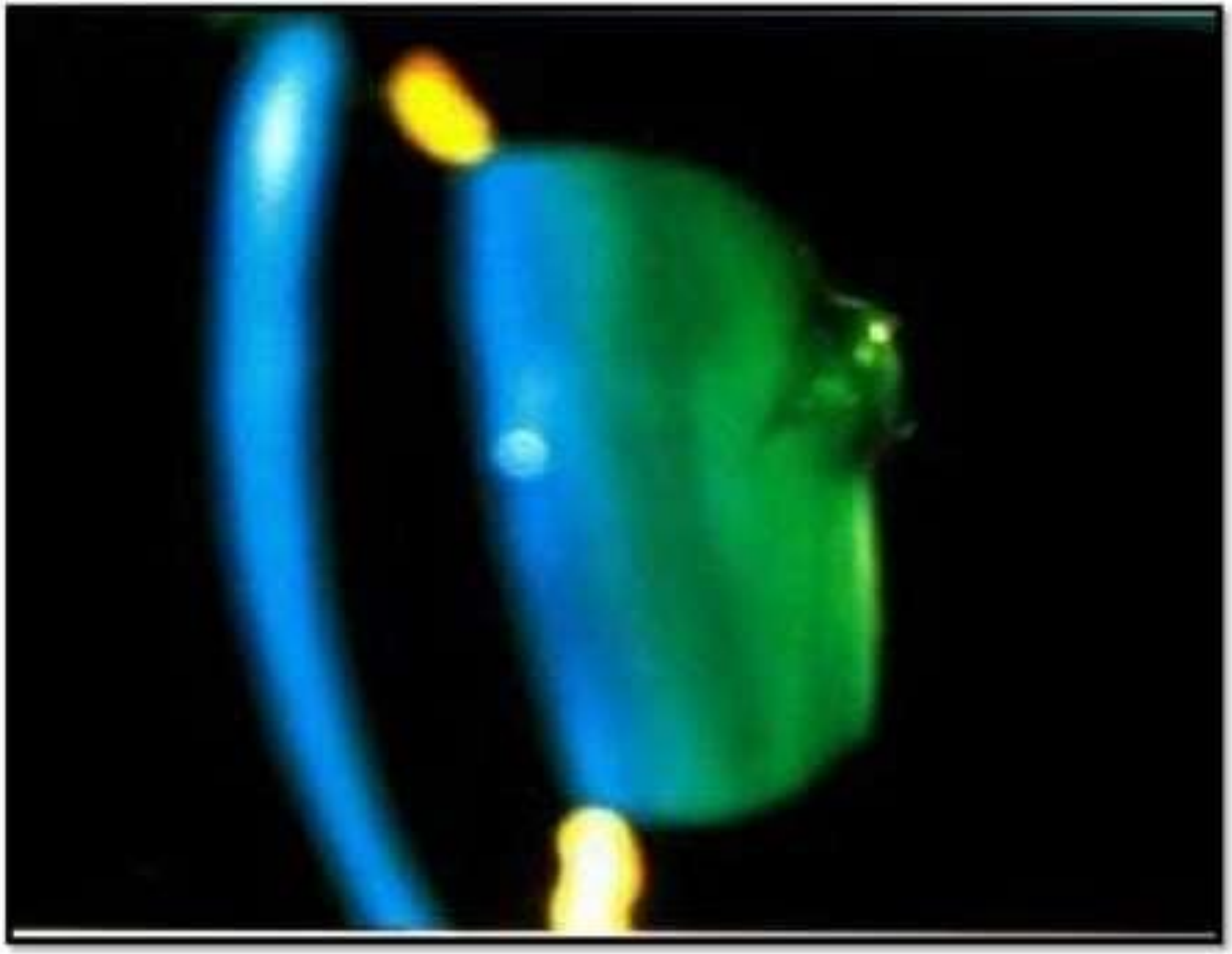
   Retinal flecks & posterior polymorphous corneal dystrophy.



POSTERIOR LENTICONUS

1. INHERITANCE : Most are unilateral, sporadic & not associate with syndrome abnormalities. Rarely bilateral cases, may be familial.
2. SIGNS : A round or conical bulge of the posterior axial zone of the lens into the vitreous associate with local thinning of absence of the capsule.

- It may be associated with opacification of the posterior capsule & hyaloids remnants.



LENTIGLOBUS

- Very rare,
- Usually unilateral, generalized hemispherical deformity of the lens which may be associated with posterior polar lens opacity.

COLOBOMA

- A coloboma is characterized by notching at the inferior equator with corresponding abesence of zonular fibers. It is not true coloboma as there is no focal absences of a tissue layer due to failure of closure of the optic fissure.

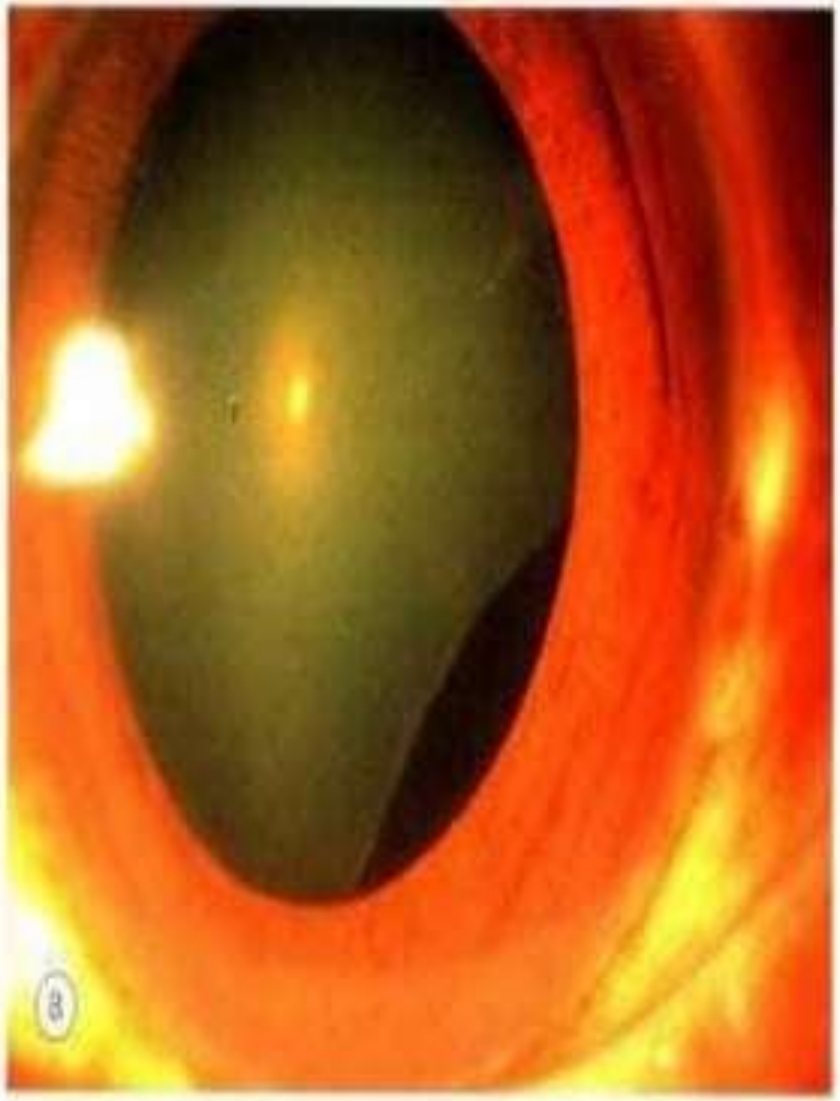


Fig. 3.24
(a) Lens coloboma; **(b)** coloboma of the lens and iris in a microphthalmic eye (Courtesy of R Foyla – fig. a; N Rogers – fig. b)

MICROSPHEROPHAKIA

I. SIGNS: the lens is small & spherical

II. CAUSES : include AD micro spherophakia which is not associated with syndrome defects, Marfan syndrome, well-Marchesani syndrome, hyperlysinaemia & congenital rubella.

- ocular association- include peter anomaly and familial ectopia lentis pupillae
- complication- include lenticular myopia, subluxation and total dislocation in to anterior chamber.

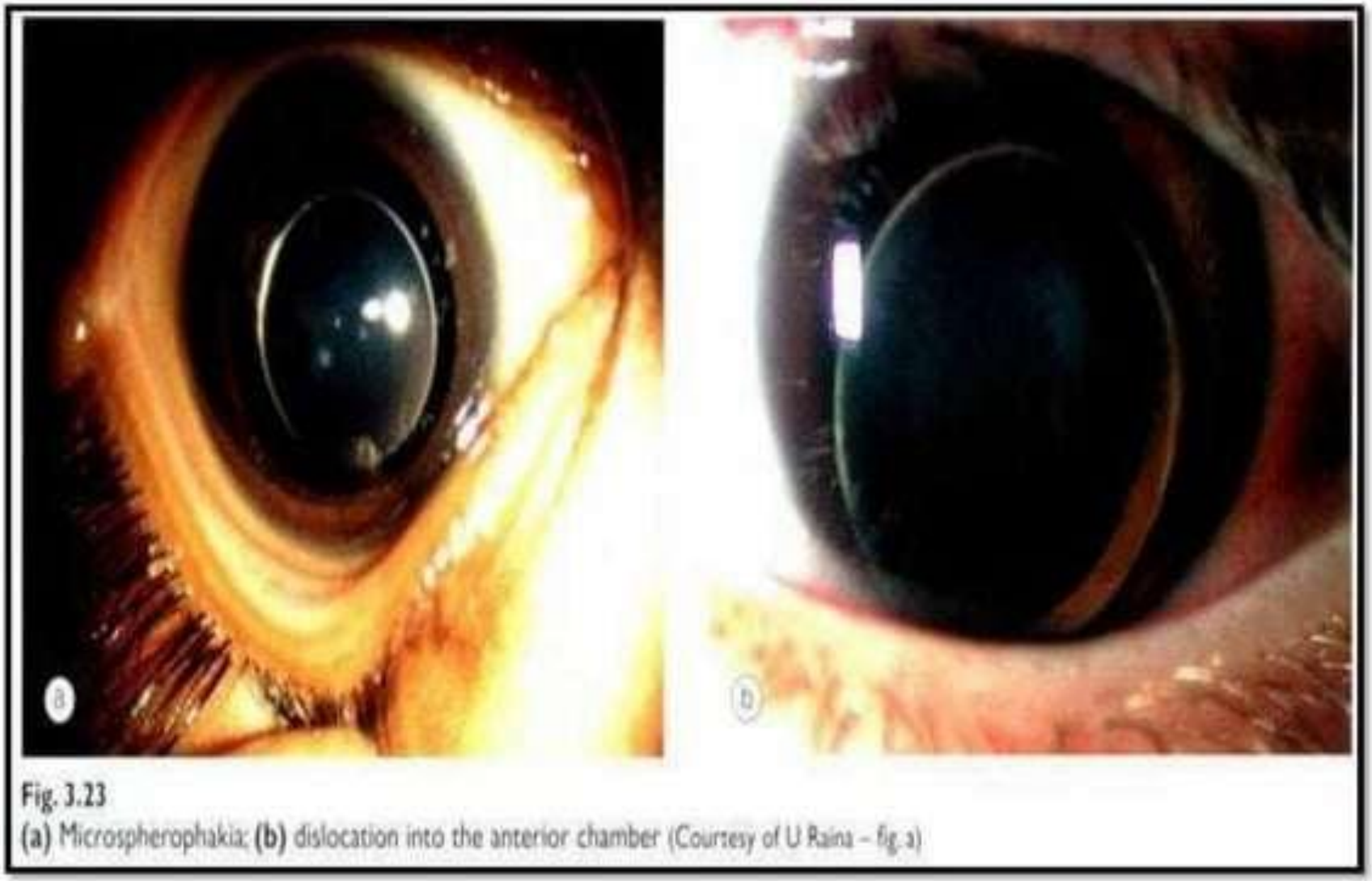
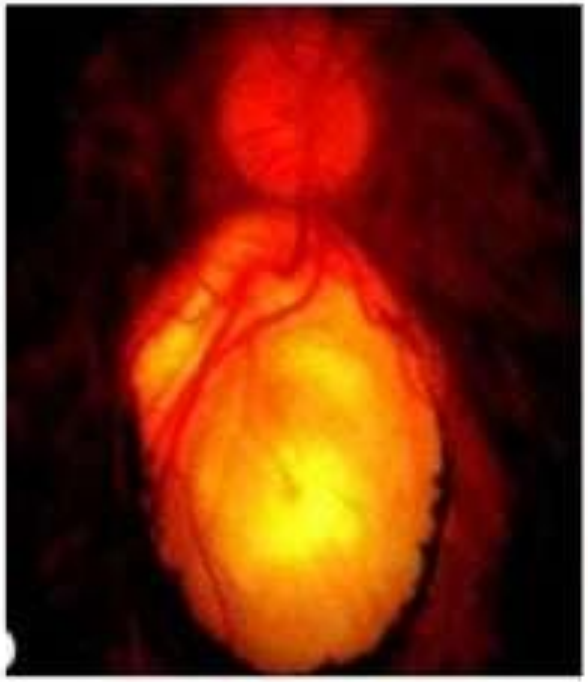
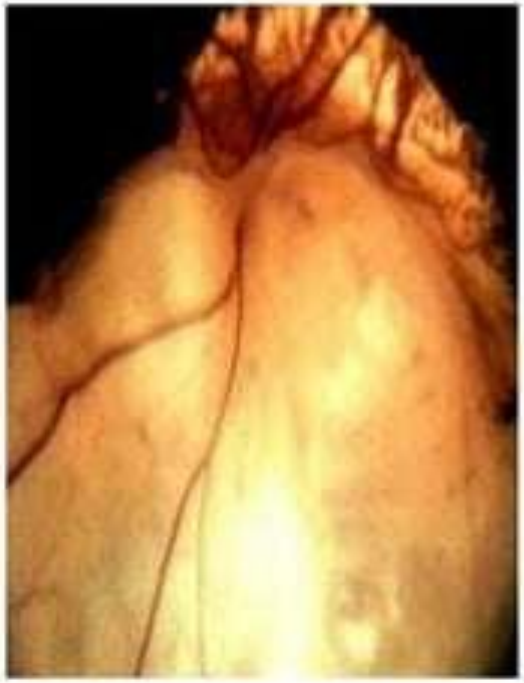
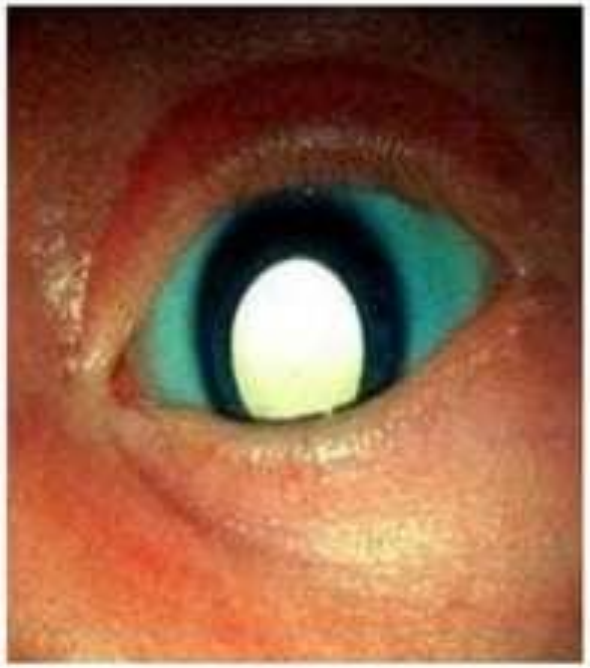


Fig. 3.23
(a) Microspherophakia; (b) dislocation into the anterior chamber (Courtesy of U Raina – fig. a)

RETINA & CHOROID

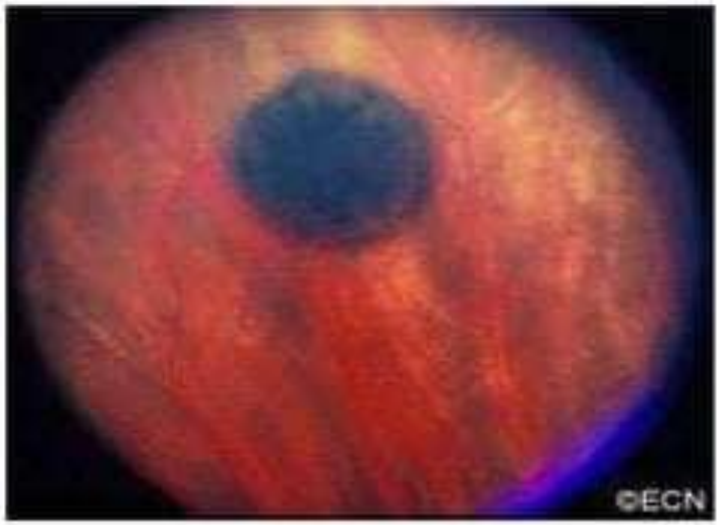
CHOROIDAL COLOBOMA

- A coloboma is the absence of part of an ocular structure as a result of incomplete closure of the embryonic fissure that may involve the entire length of the fissure (complete coloboma)or only part .
- A chorioretinal coloboma may be unilateral or bilateral & usually occurs sporadically in otherwise normal individuals.

ANOMALIES OF THE OPTIC DISC

- crescents,
- situs inversus,
- congénital pigmentation,
- coloboma,
- drusen
- hypoplasia of the optic disc.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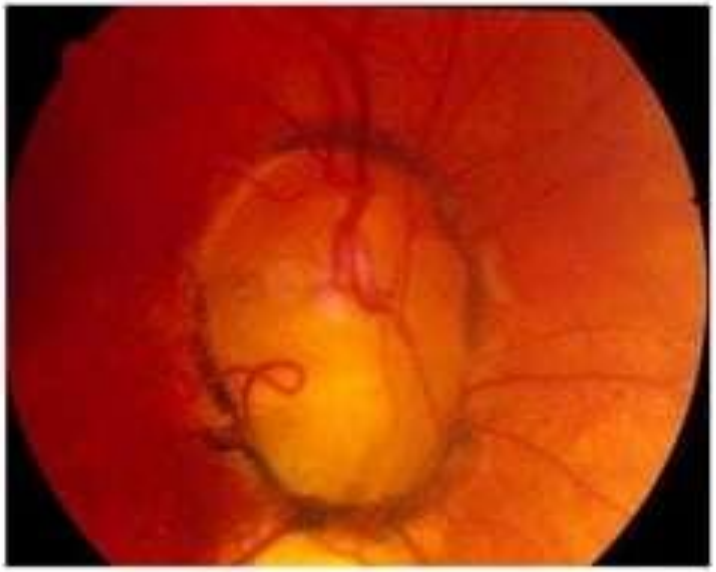
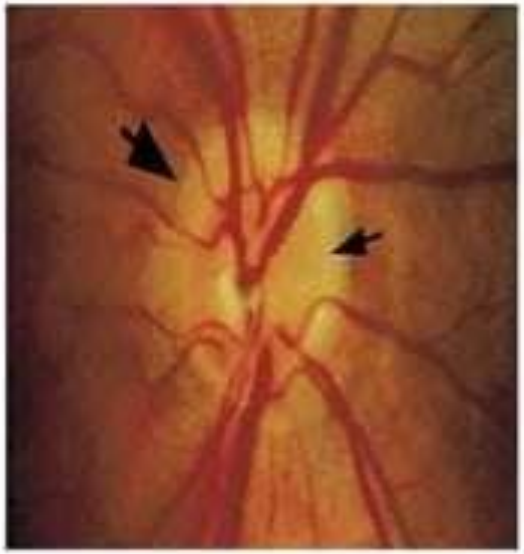
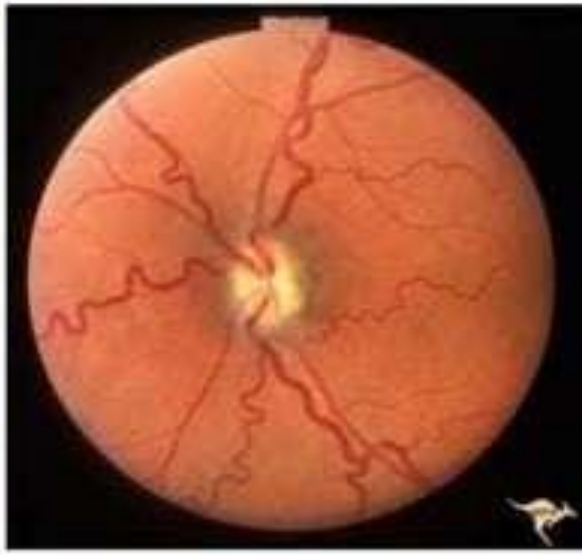
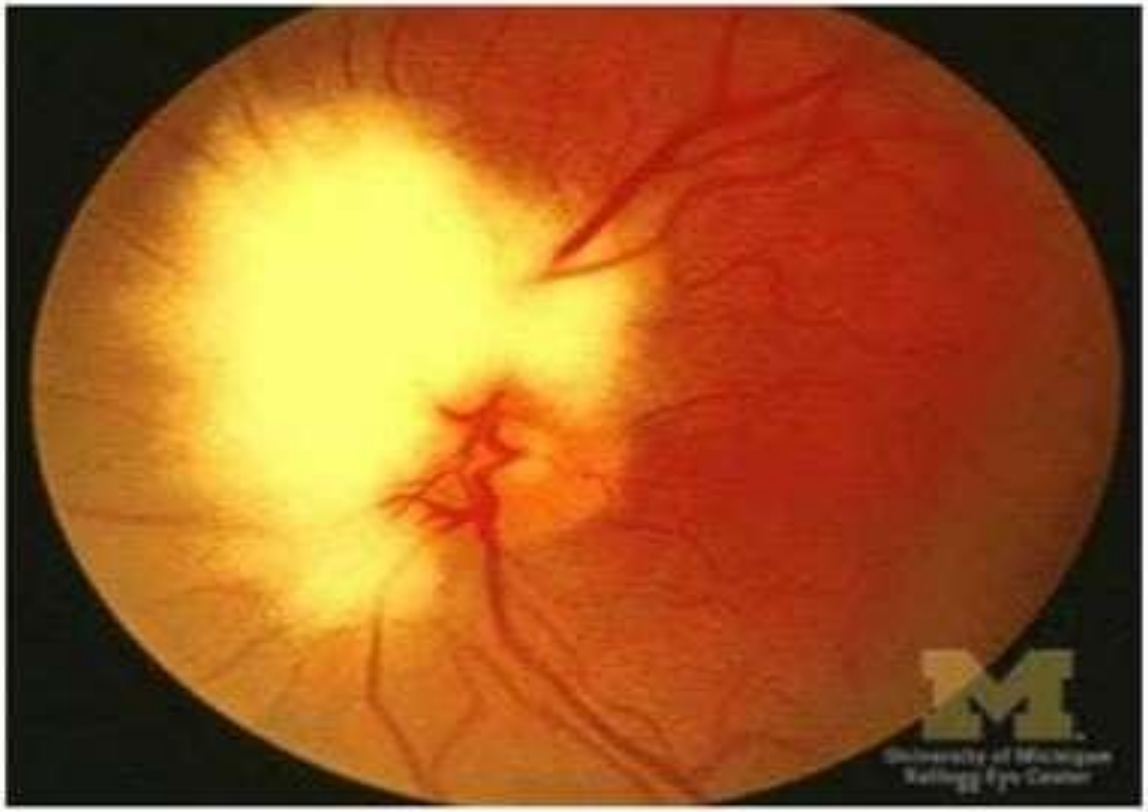






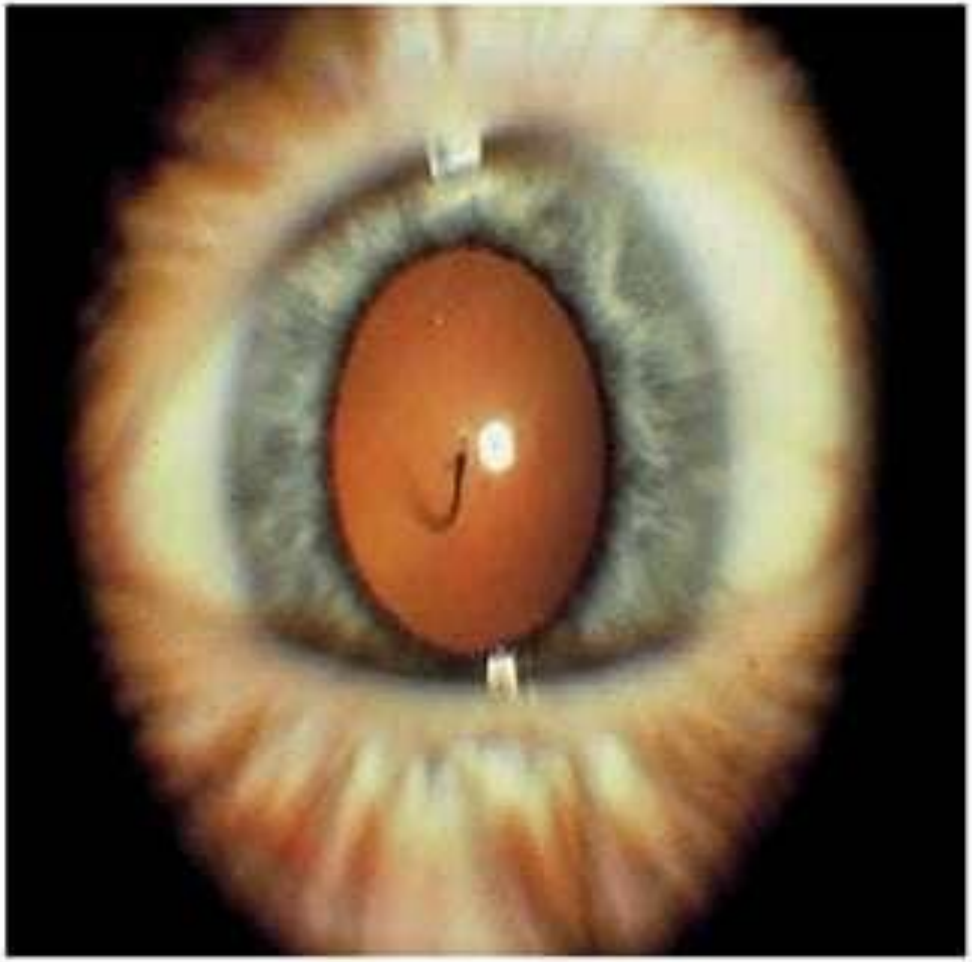
ANOMALIES OF THE NERVE FIBRES

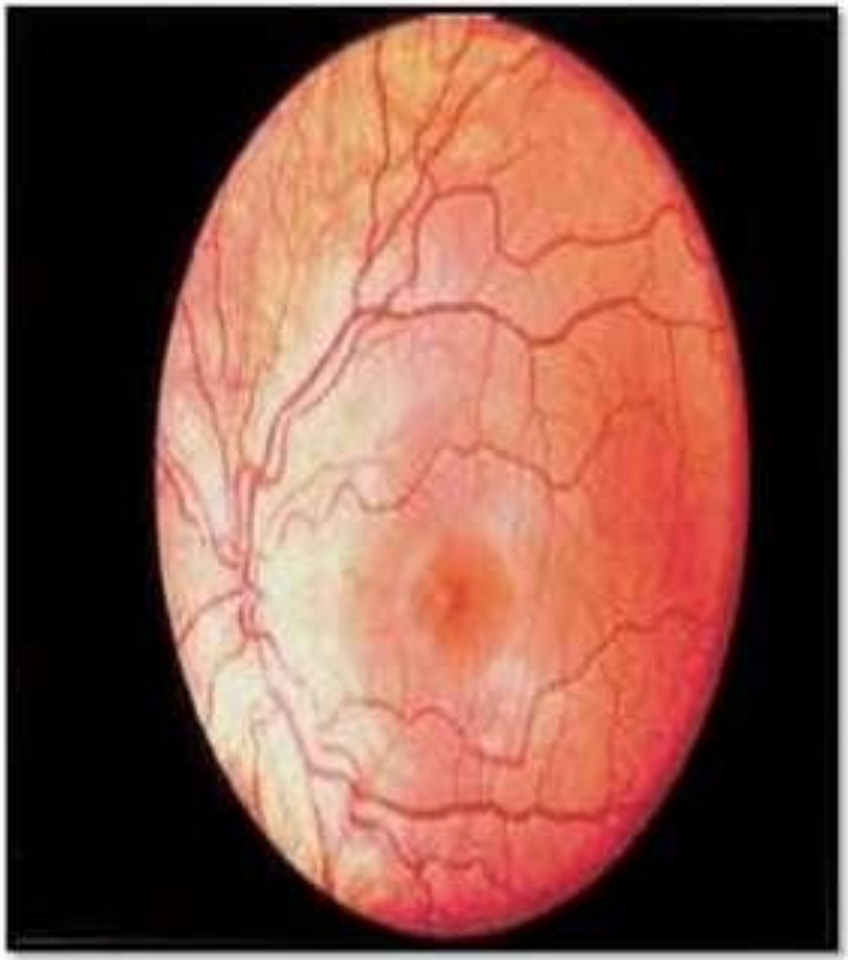
- Medullated (opaque) nerve fibres.



ANOMALIES OF VASCULAR ELEMENTS

- persistent hyaloid artery
- congenital tortuosity of retinal vessels.

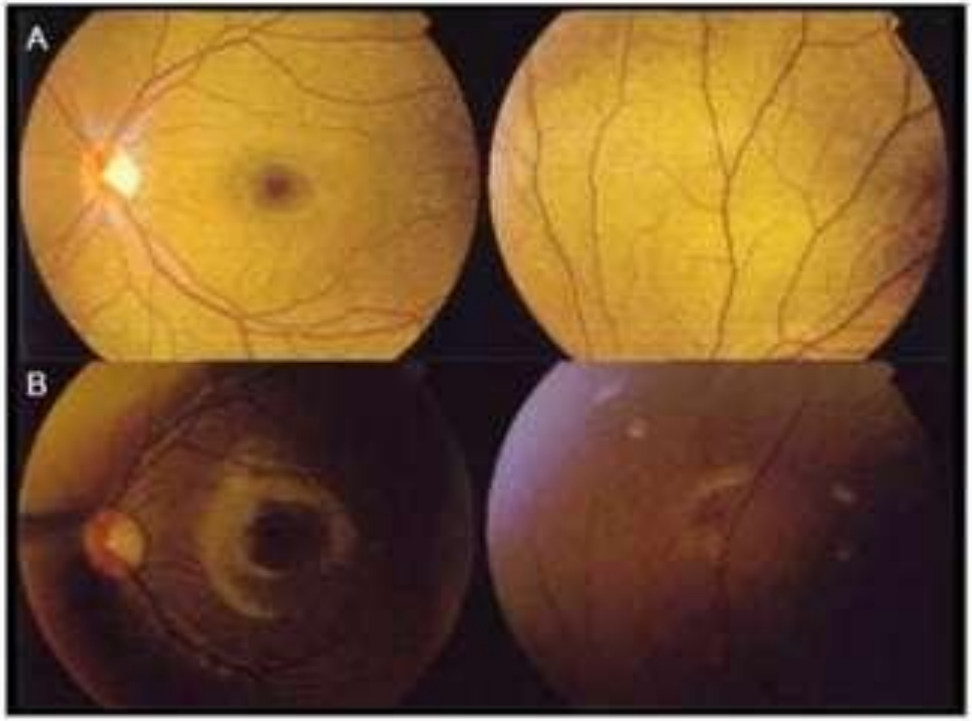




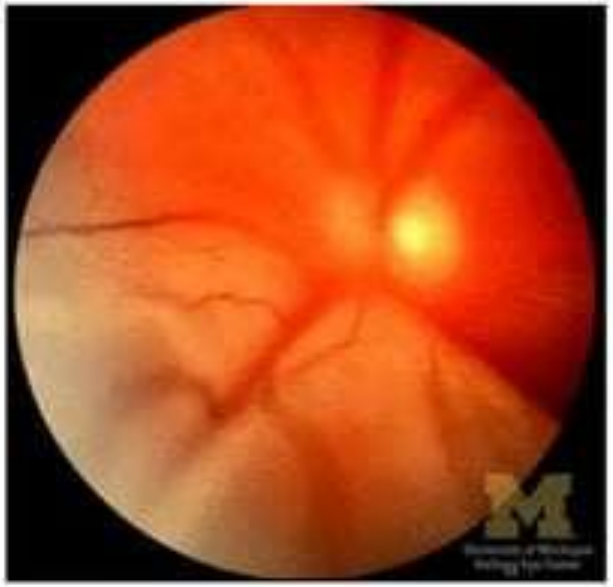
ANOMALIES OF RETINA PROPER

- Albinism
- Congenital night blindness
- Congenital day blindness
- Oguchi's disease
- Congenital retinal cyst
- congenital retinal detachment
- coloboma of the fundus.

OGUCHI'S DISEASE



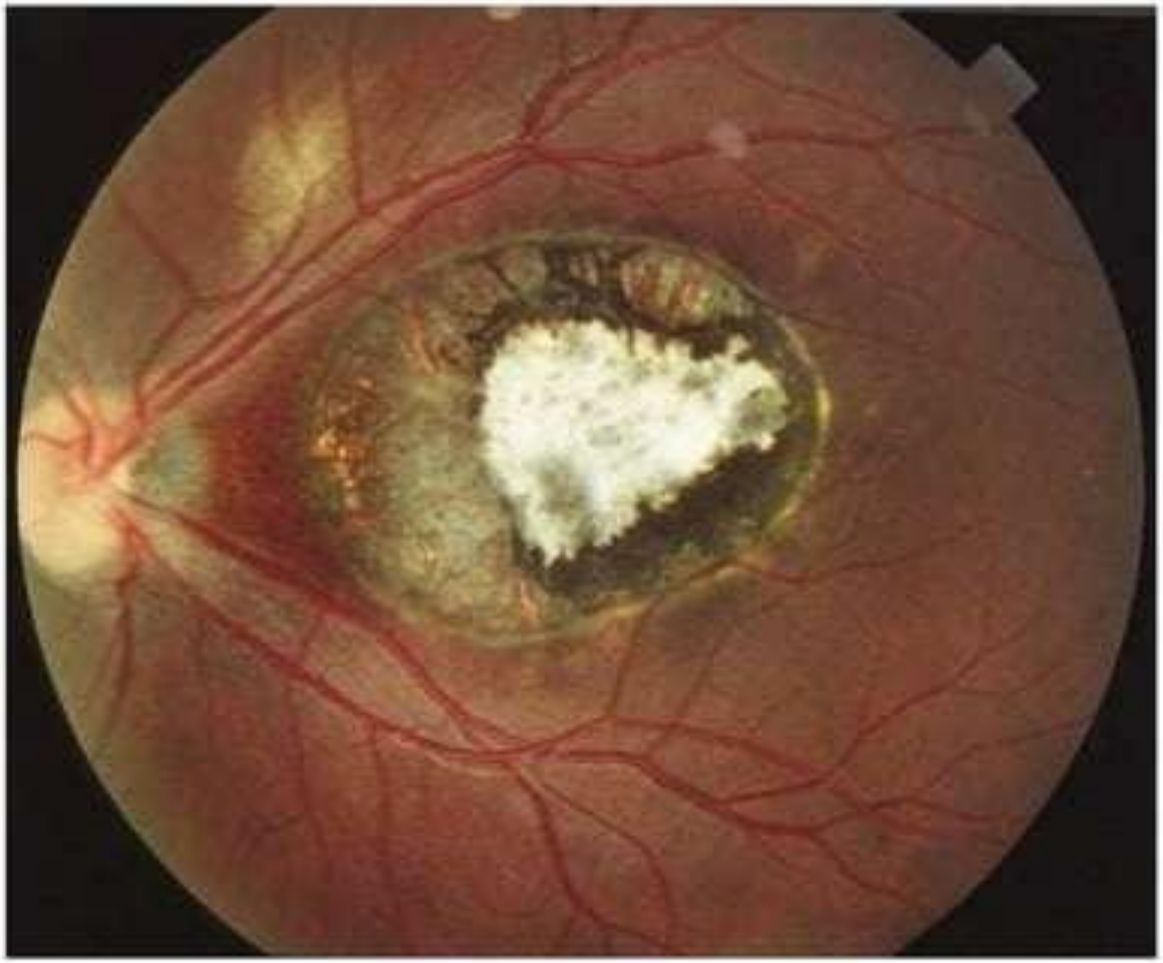
CONGENITAL RETINAL DETACHMENT



ANOMALIES OF THE MACULA

- aplasia,
- hypoplasia
- coloboma

MACULAR COLOBOMA



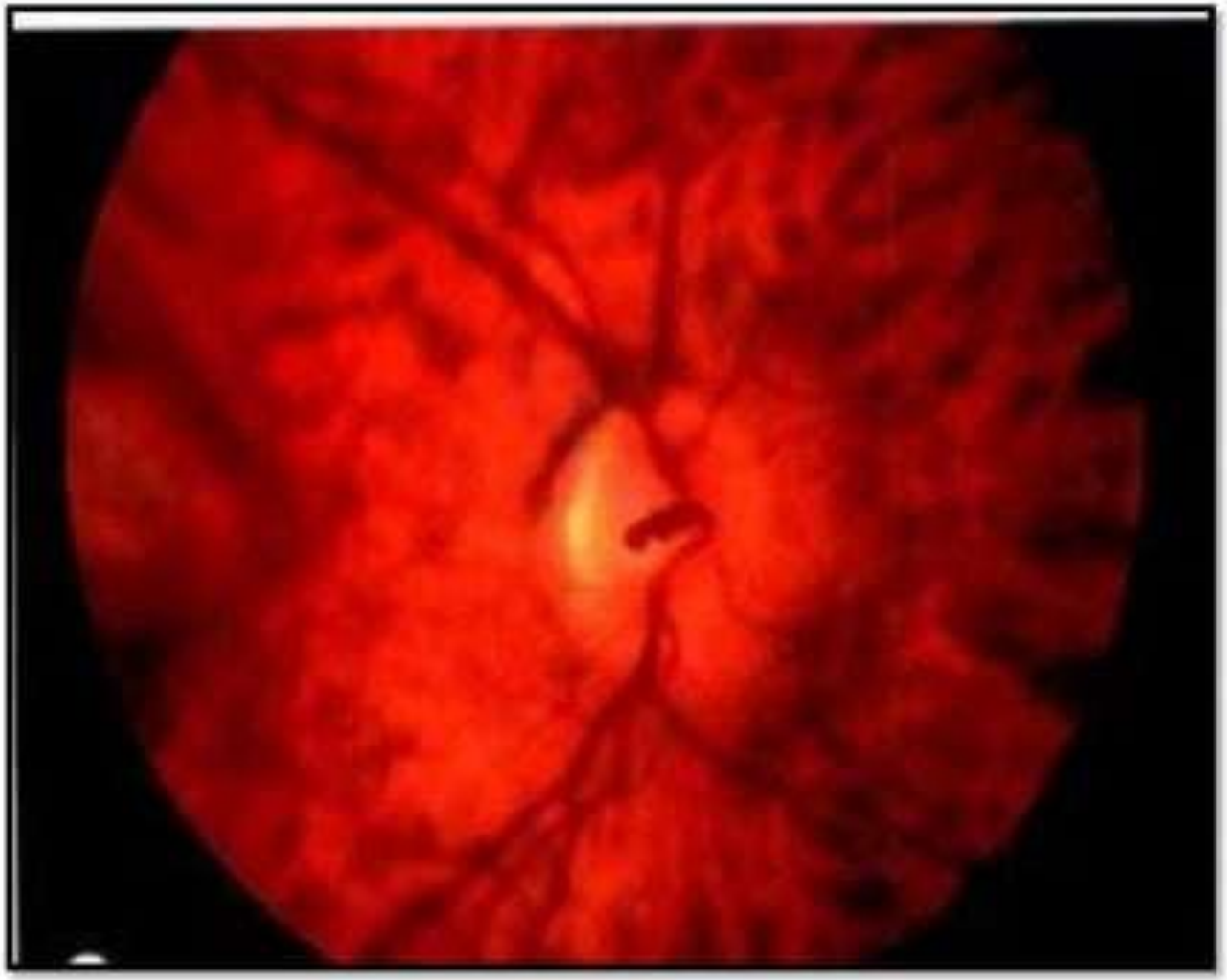
OPTIC NERVE

- PREPAPILLARY LOOP
- BERGMESTER PAPILLA
- OPTIC DISC COLOBOMA
- OPTIC NERVE HYPOPLASIA
- OPTIC DISC PIT
- CONGENITAL TILTED DISC SYNDROME
- MEGALOPAPILLA

PREPAPILLART LOOP

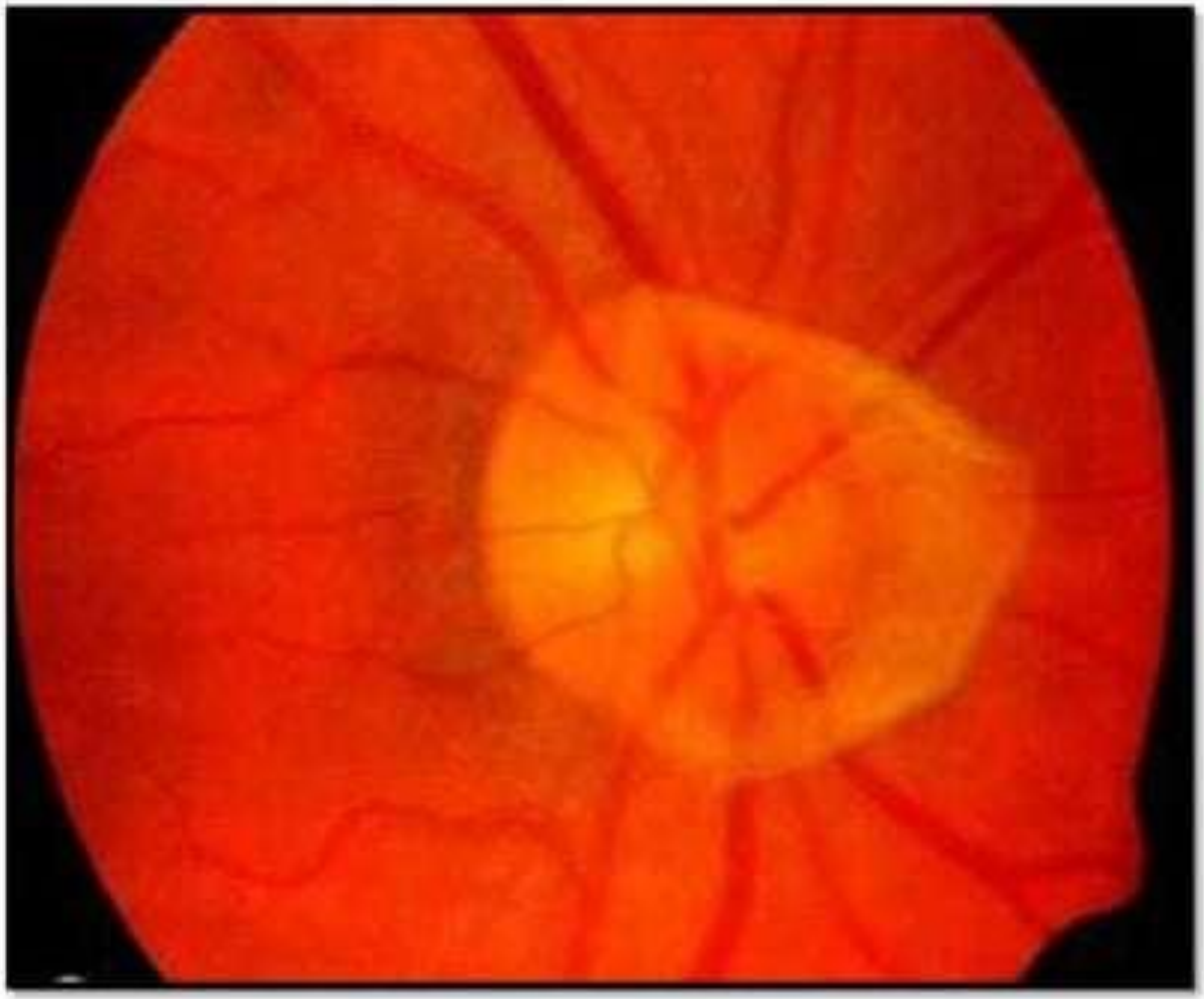
- sign- A unilateral vascular loop extending from the disc in to the vitreous cavity
- complication- obstruction in the distribution of the retinal artery supplying the loop occur in 10% cases

  vitreous hemorrhage are rare



BERGMEISTER PAPILLA

- Bergmeister papilla is an uncommon unilateral anomaly that is derived from avascular remnants of the hyaloids system & is characterized by raised glial tissue on the disc surface



OPTIC NERVE HYPOPLASIA

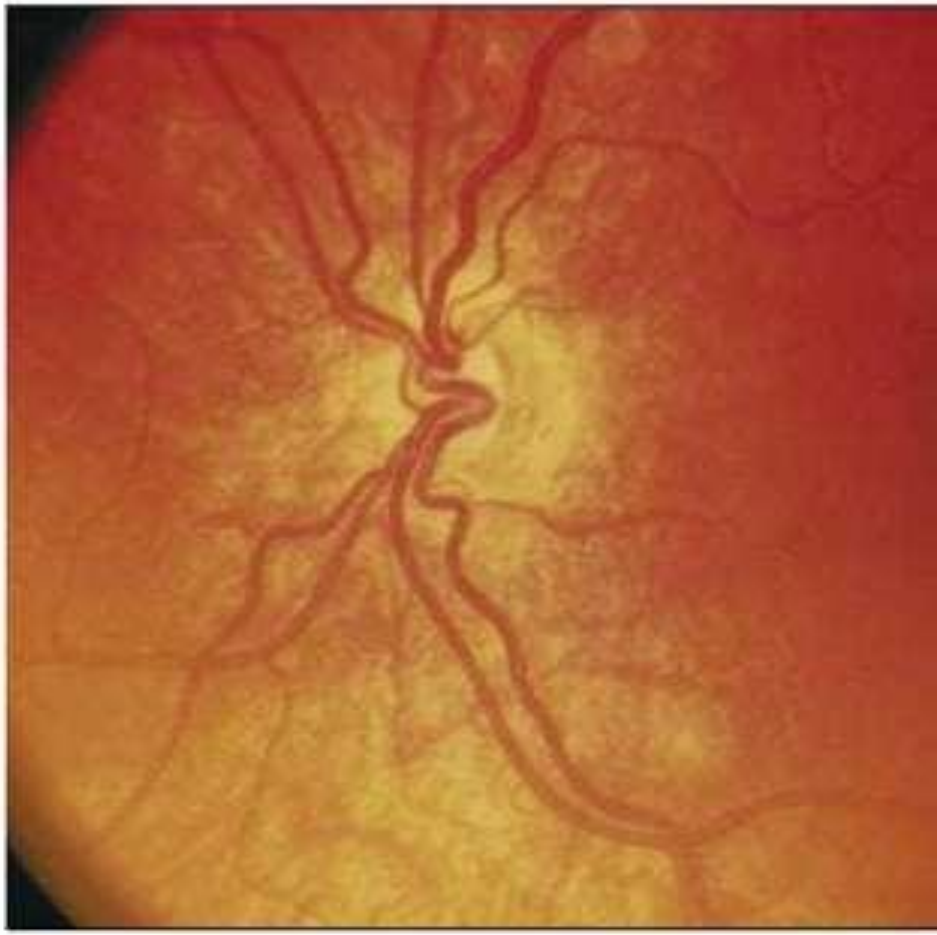
Optic nerve hypoplasia (ONH) is the most common optic disc anomaly and is the third leading cause of blindness in children in the western world after cerebral damage and retinopathy of prematurity.

Risk factors for ONH include

- young maternal age,
- maternal smoking,
- preterm birth and its complications.

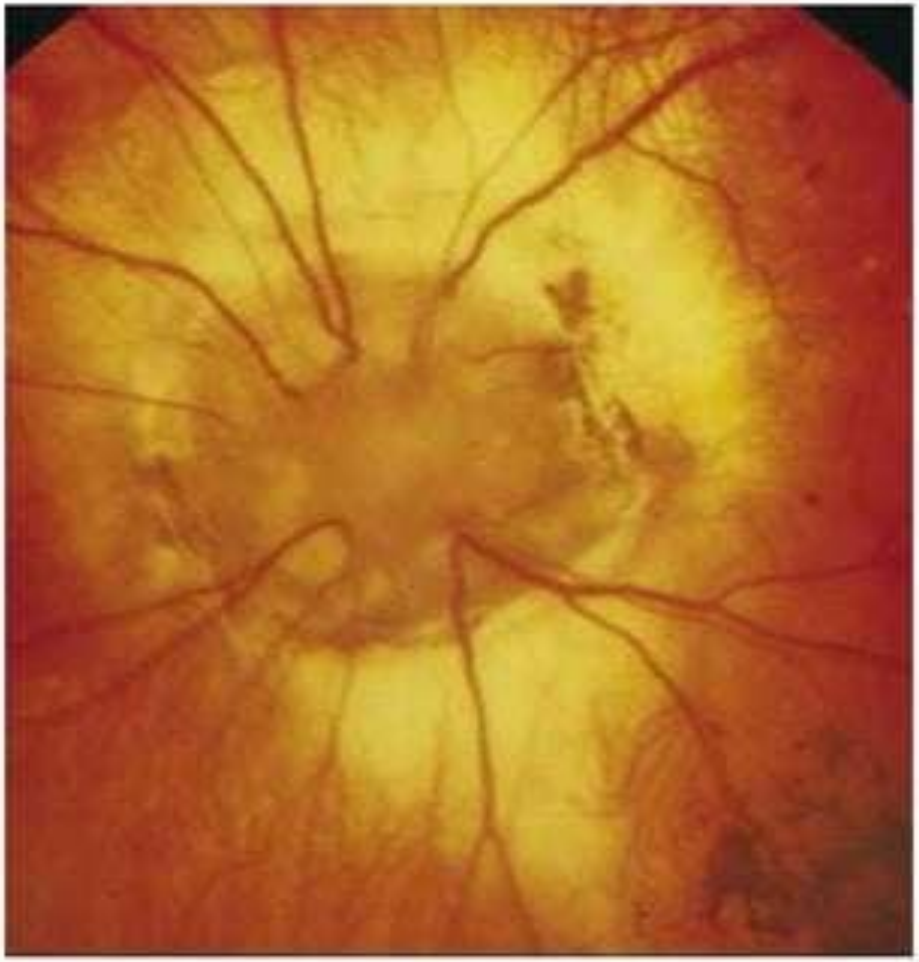
Clinical features

- An abnormally small optic nerve head.
- Double-ring sign:
- The optic nerve is pale surrounded by a yellowish peripapillary ring of sclera and an outer concentric ring of hypopigmentation.
- *outer ring of normal junction between sclera and lamina cribrosa*
- *inner ring denoting extension of retina and RPE over lamina cribrosa*



MORNING GLORY DISC ANOMALY

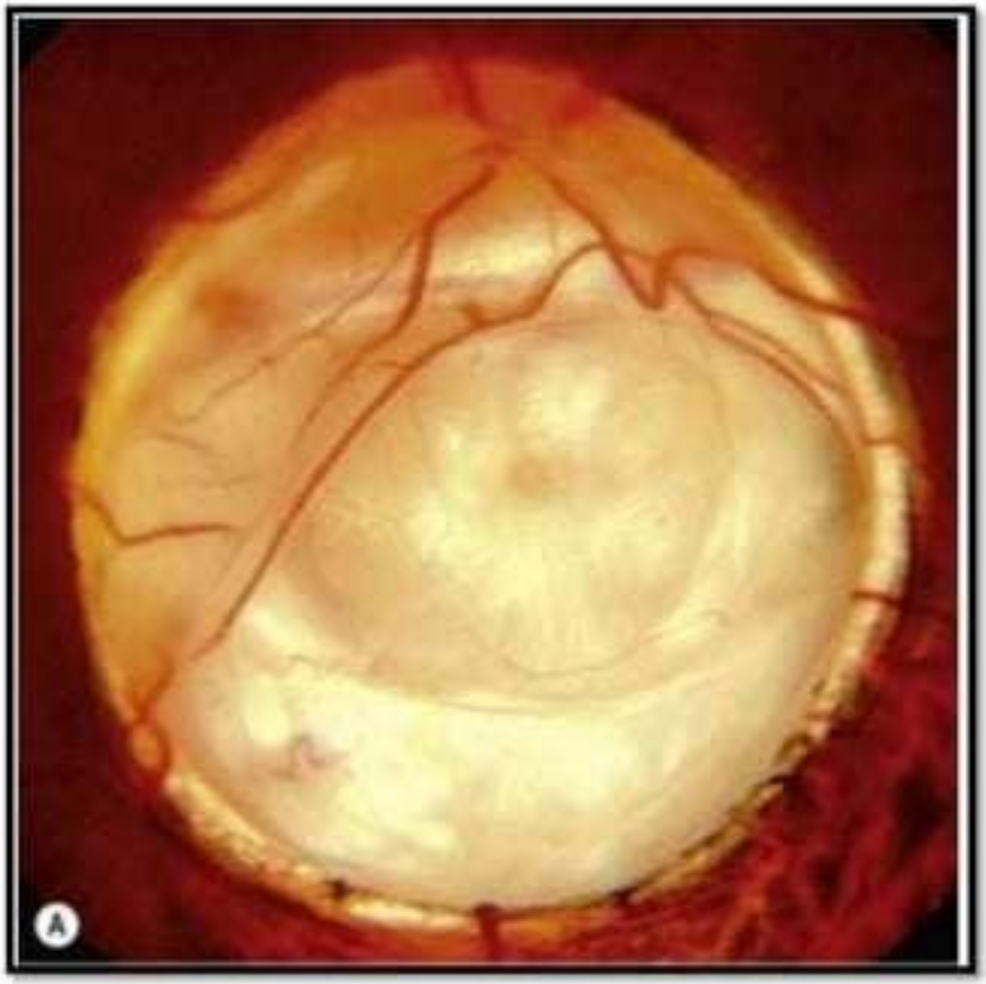
- The morning glory disc anomaly is a congenital excavation of posterior globe that involves the optic disc.
- The term reflects the morphological similarity to the flower of the morning glory plant.
- Morning glory syndrome is ostensibly a sporadic condition.
- It usually occurs as a unilateral condition, though bilateral lesions have been reported.
- Morning glory discs are more common in females (2:1)



OPTIC DISC COLOBOMA

- Optic disc coloboma can be present in one or both eyes.
- They may arise sporadically or be inherited in autosomal dominant fashion.
- It has recently been shown to be associated with PAX2 gene mutations as part of the renal-coloboma syndrome.

- **Pathogenesis:** It is caused by an incomplete or abnormal apposition of the proximal ends of the embryonic fissure.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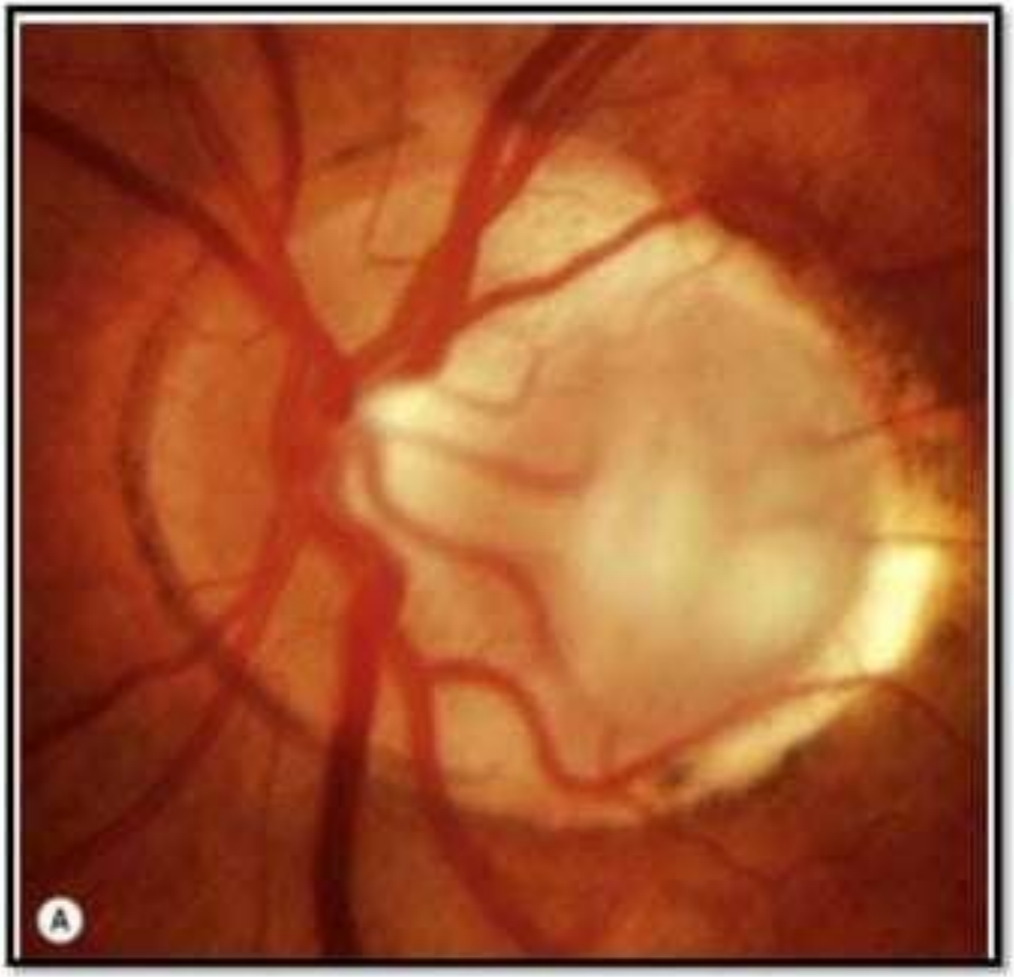
OPTIC PIT

- Optic nerve pits are rare congenital anomalies that are part of a spectrum of congenital cavitary optic disc anomalies that may be associated with juxtapapillary retinal detachments.

- They occur in less than 1 in 10,000 patients seen in an ophthalmic setting and are bilateral in 10% to 15% of cases.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MEGALOPAPILLA

- Megalopapilla is a generic term that connotes an abnormally large optic disc that lacks the inferior excavation of optic disc coloboma or the numerous anomalous features of the morning glory disc anomaly.
- This condition is usually bilateral and often associated with a large cup-to-disc ratio.
- Patients who have Megalopapilla are often suspected to have glaucoma.
- Unlike the situation in glaucoma, however, the optic cup is usually round or horizontally oval with no vertical notch or encroachment .

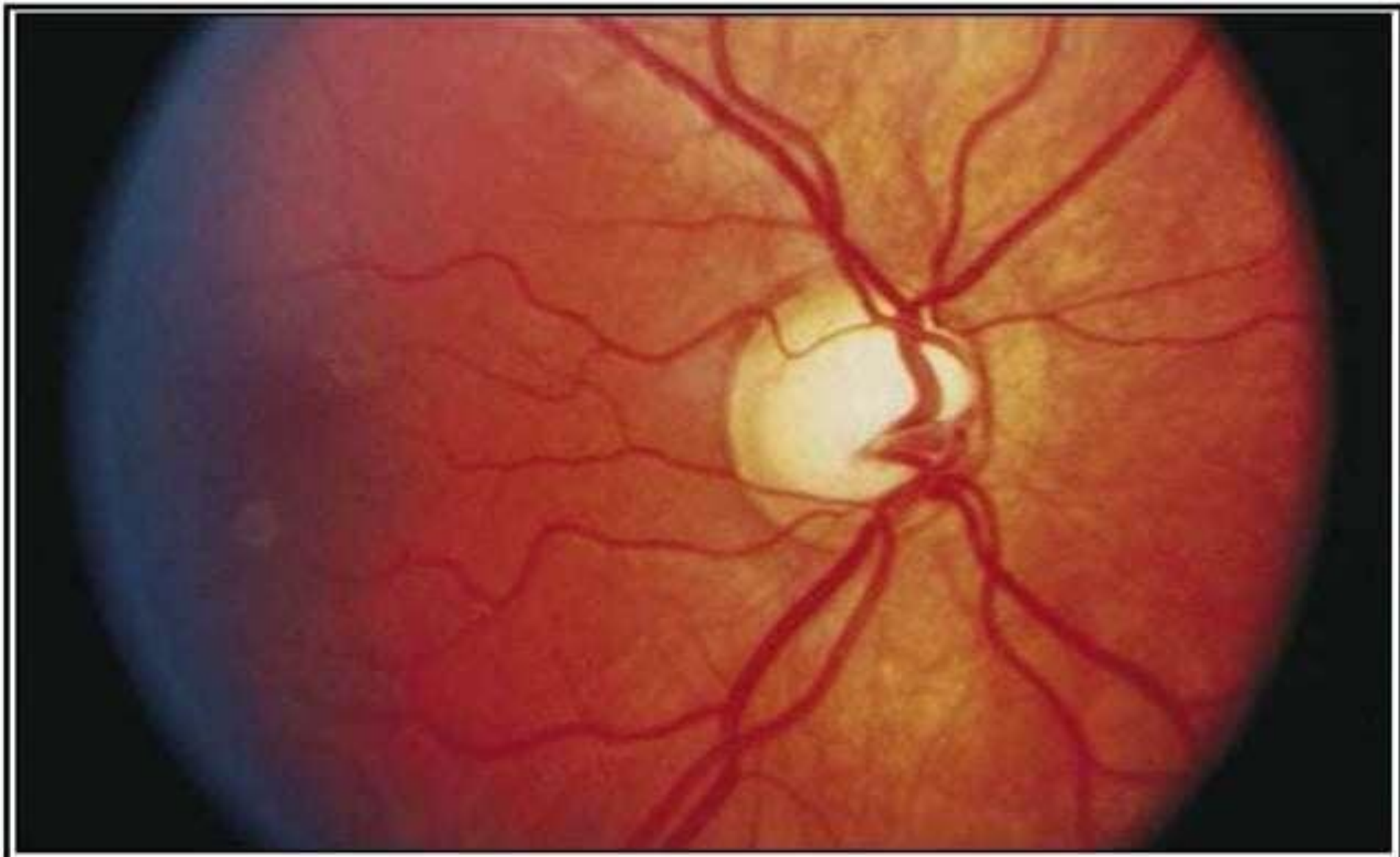
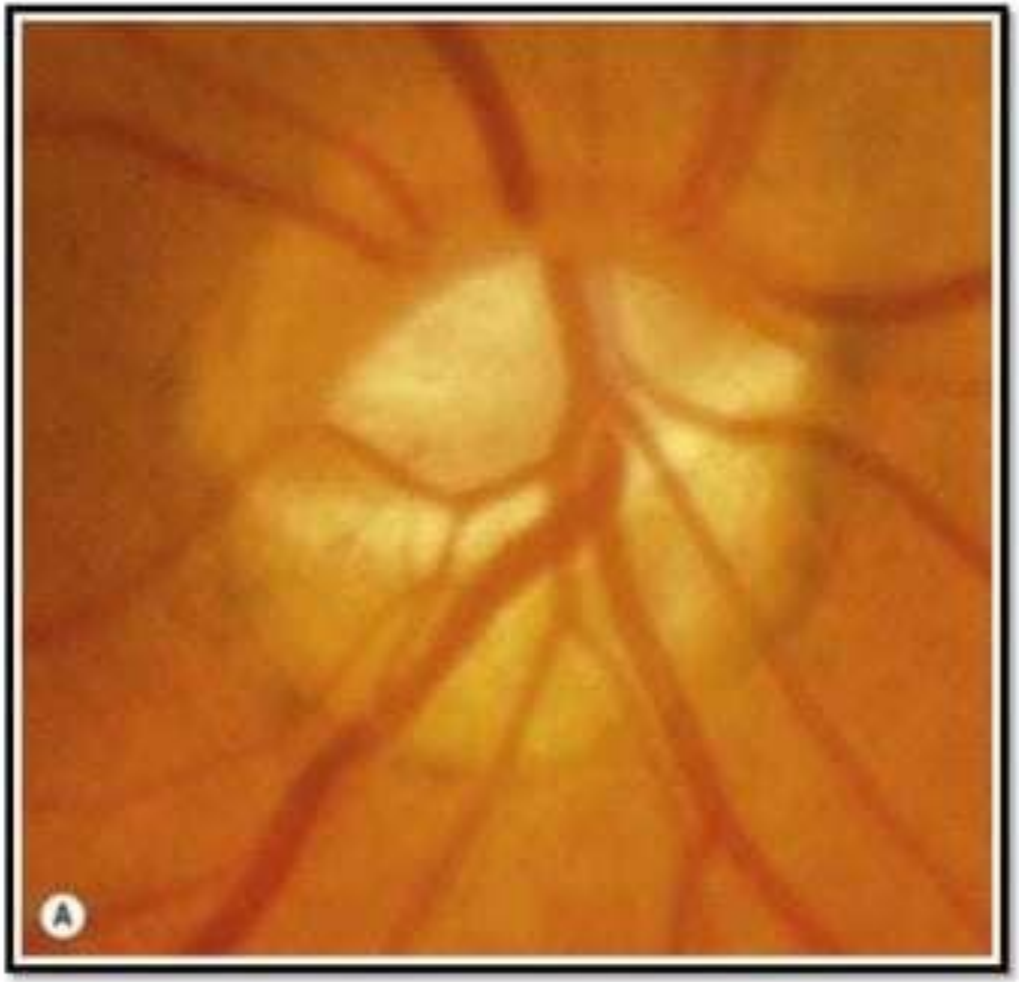


Fig. 9-4-6 Megalopapilla.

CONGENITAL TILTED DISC SYNDROME

- The tilted disc syndrome is a nonhereditary bilateral condition.
- The supero temporal optic disc is elevated and the inferonasal disc is displaced posteriorly, which results in an optic disc of oval appearance with its long axis obliquely orientated.
- This configuration is accompanied by situs inversus of the retinal vessels, congenital inferonasal conus, thinning of the inferonasal retinal pigment epithelium and choroid, and myopic astigmatism.
- These features presumably result from a generalized ectasia of the inferonasal fundus that involves the corresponding sector of the optic disc.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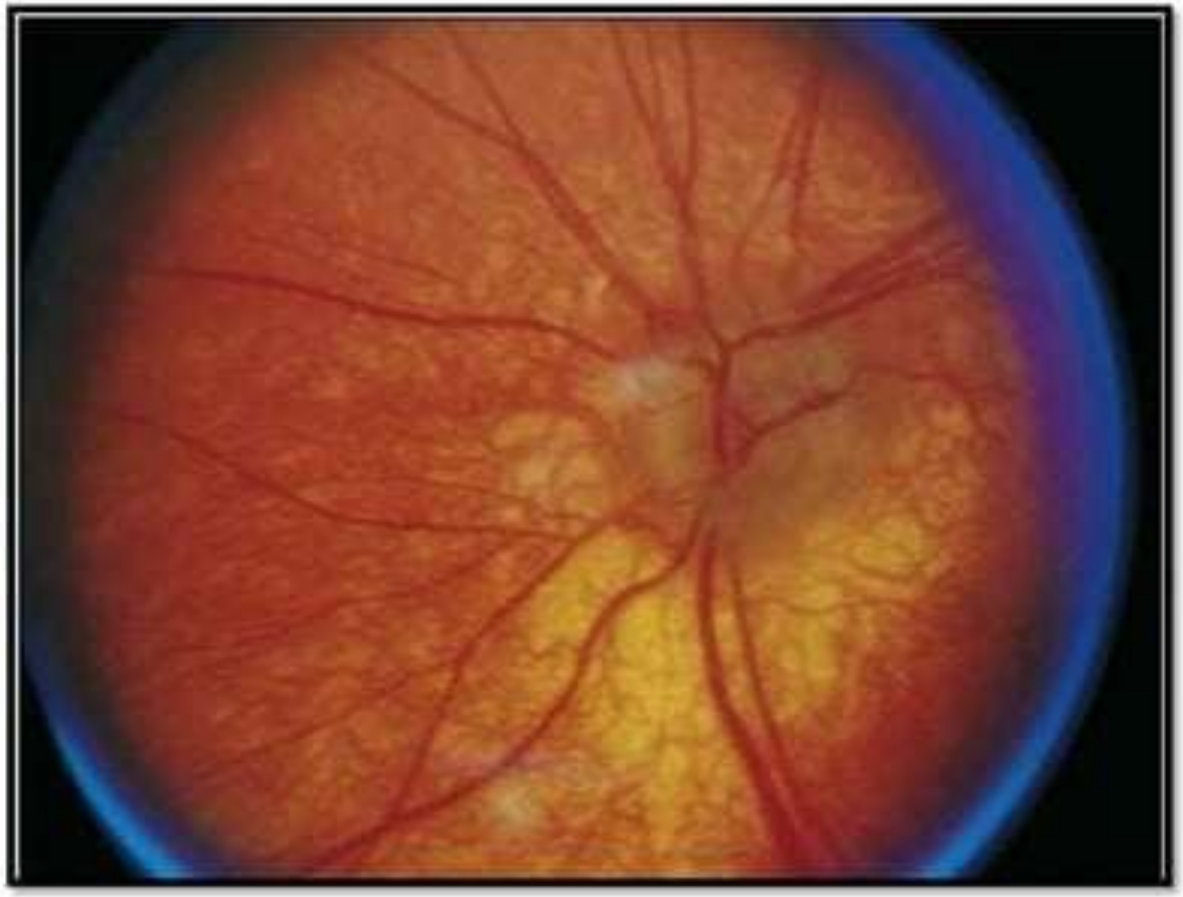
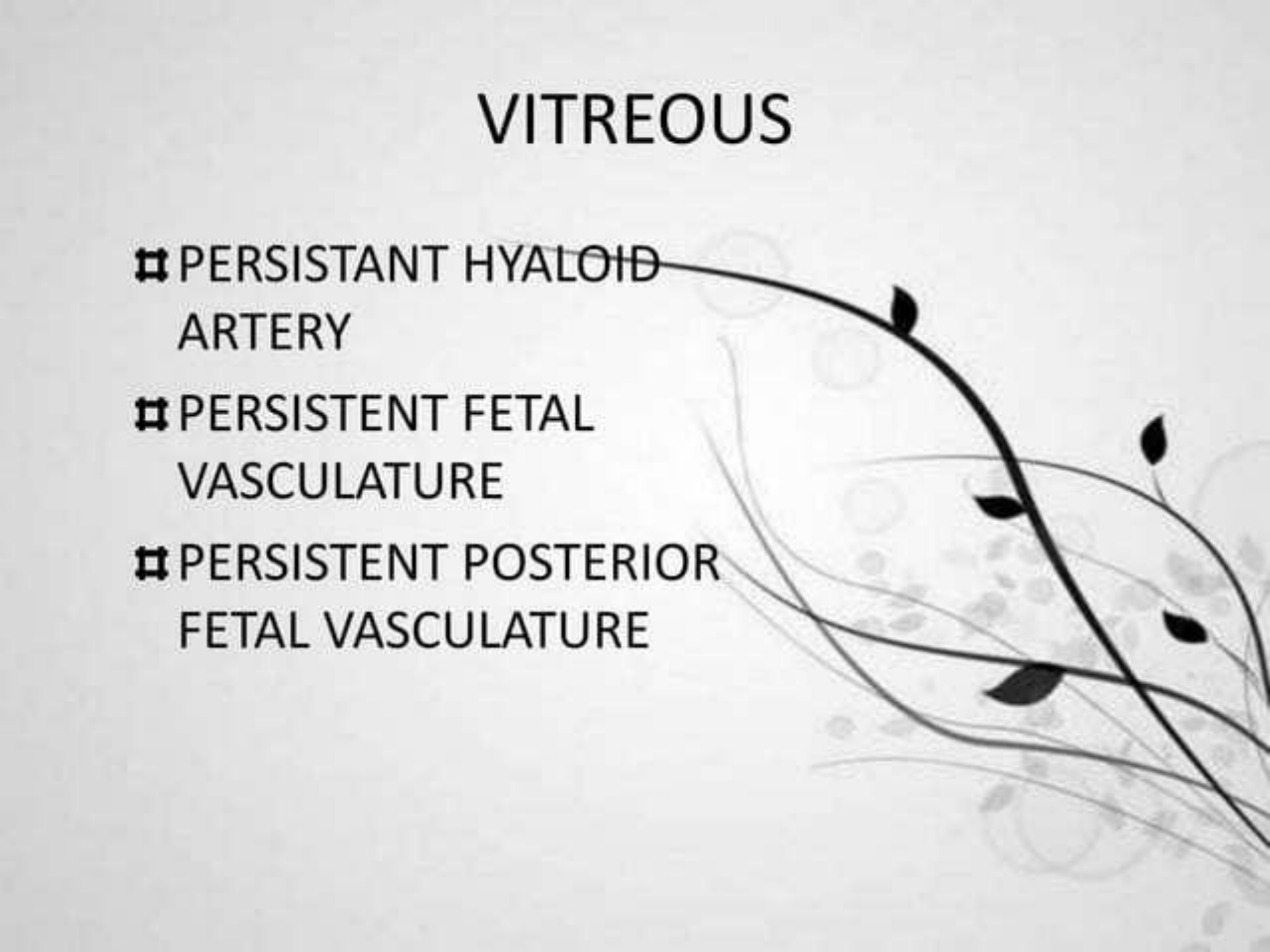
CONGENITAL OPTIC DISC PIGMENTATION

- Congenital optic disc pigmentation is a condition in which melanin anterior to or within the lamina cribrosa imparts a gray appearance to the disc.



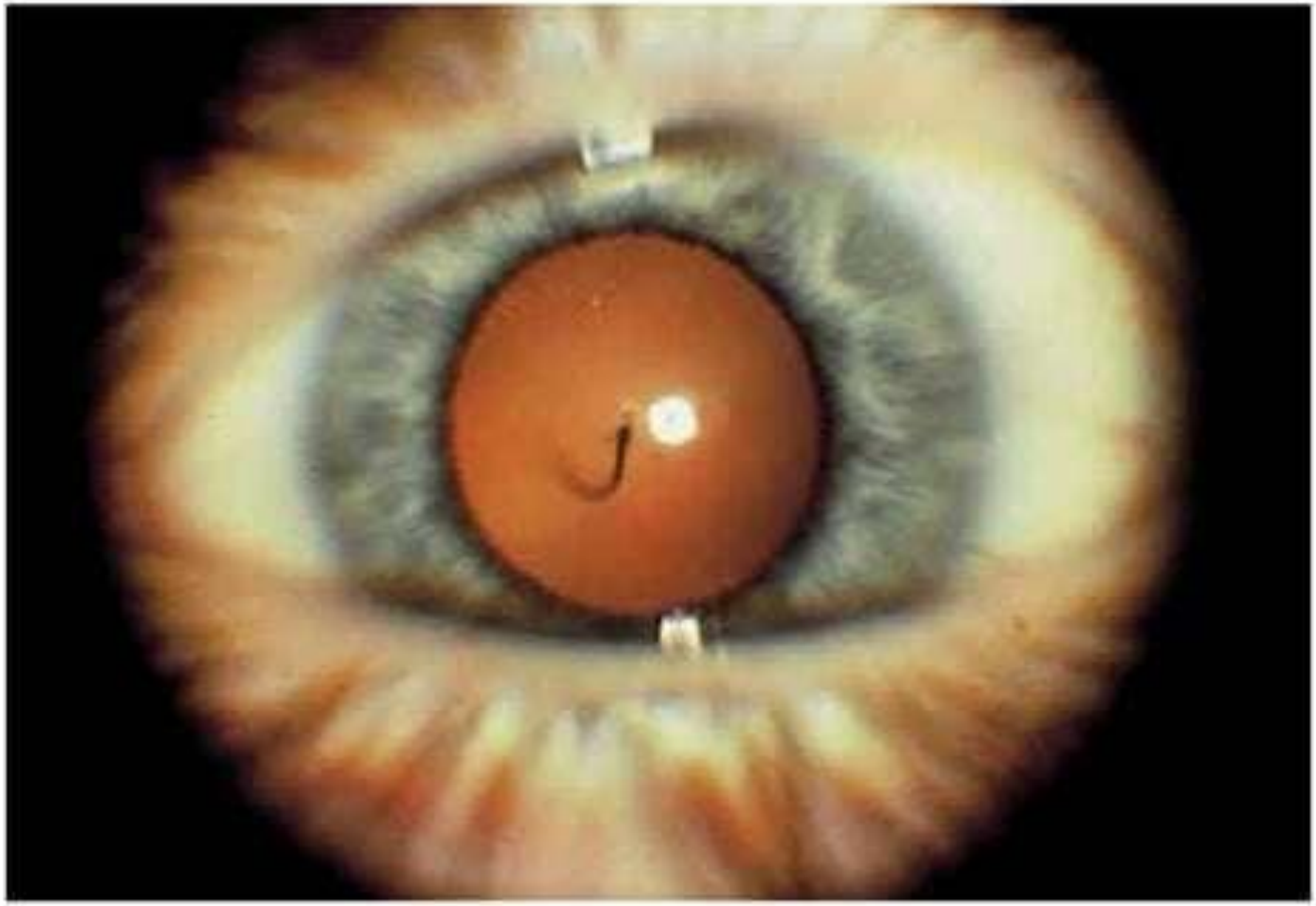
VITREOUS

- PERSISTANT HYALOID ARTERY
- PERSISTENT FETAL VASCULATURE
- PERSISTENT POSTERIOR FETAL VASCULATURE



PERSISTENT HYALOID ARTERY

- Persistent hyaloid artery is unilateral condition seen in 95% of the premature infants but rarely seen in adult.

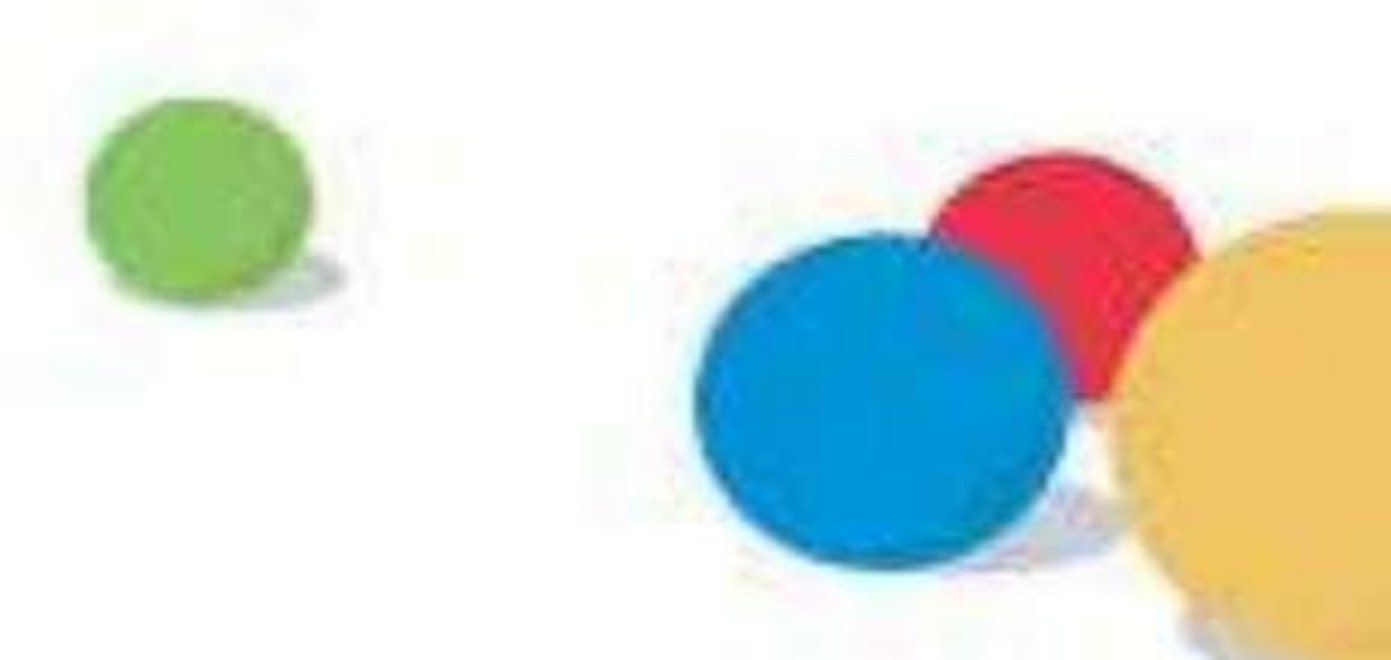


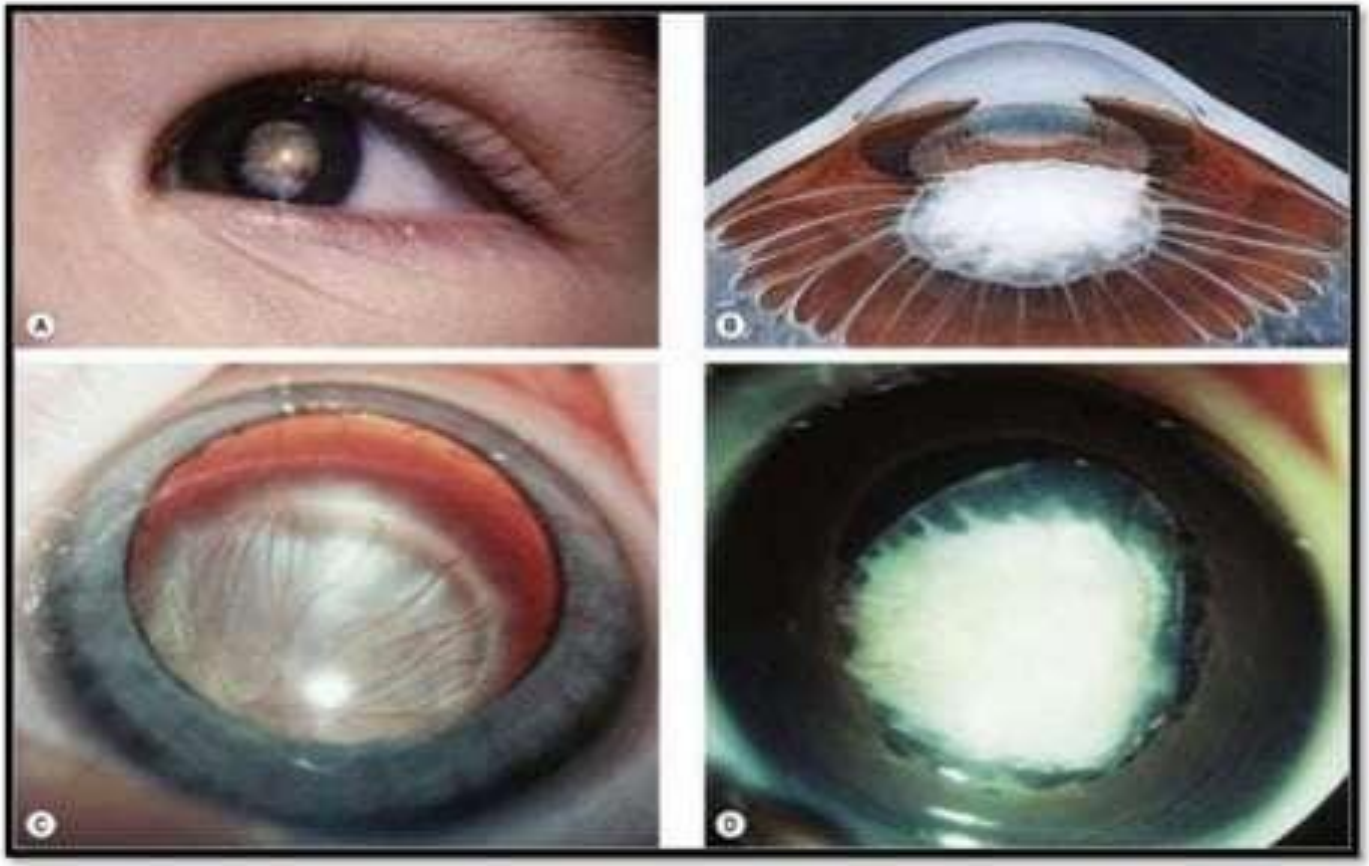
PERSISTENT FETAL VASCULATURE

- It is an uncommon , sporadic , unilateral condition.
- Caused by failure of regression of the primary vitreous.
- It is typically associated with mild microphthalmos.
- Patient is present with leukocoria.
- There are two types.

PERSISTENT ANTERIOR FETAL VASCULATURE

- Abnormally confined with anterior segment and often involve the lens.

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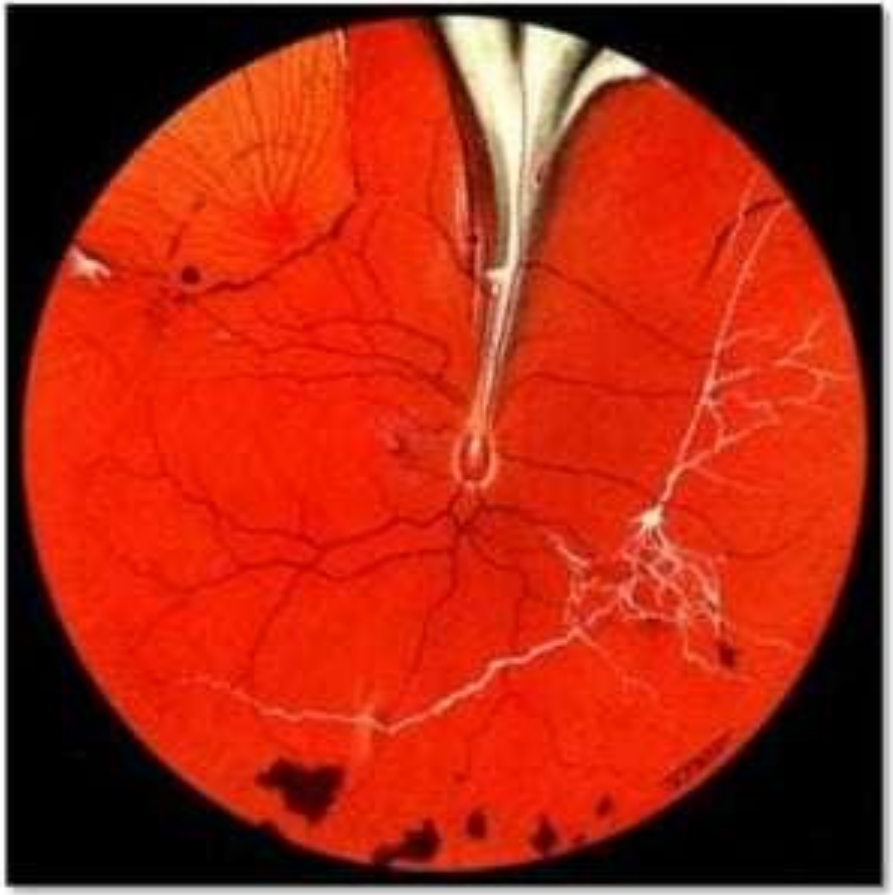
PERSISTENT POSTERIOR FETAL VASCULATURE

- The abnormality is confined to the posterior segment and lens is usually clear.
- Patient is present with leukocoria , strabismus and nystagmus.

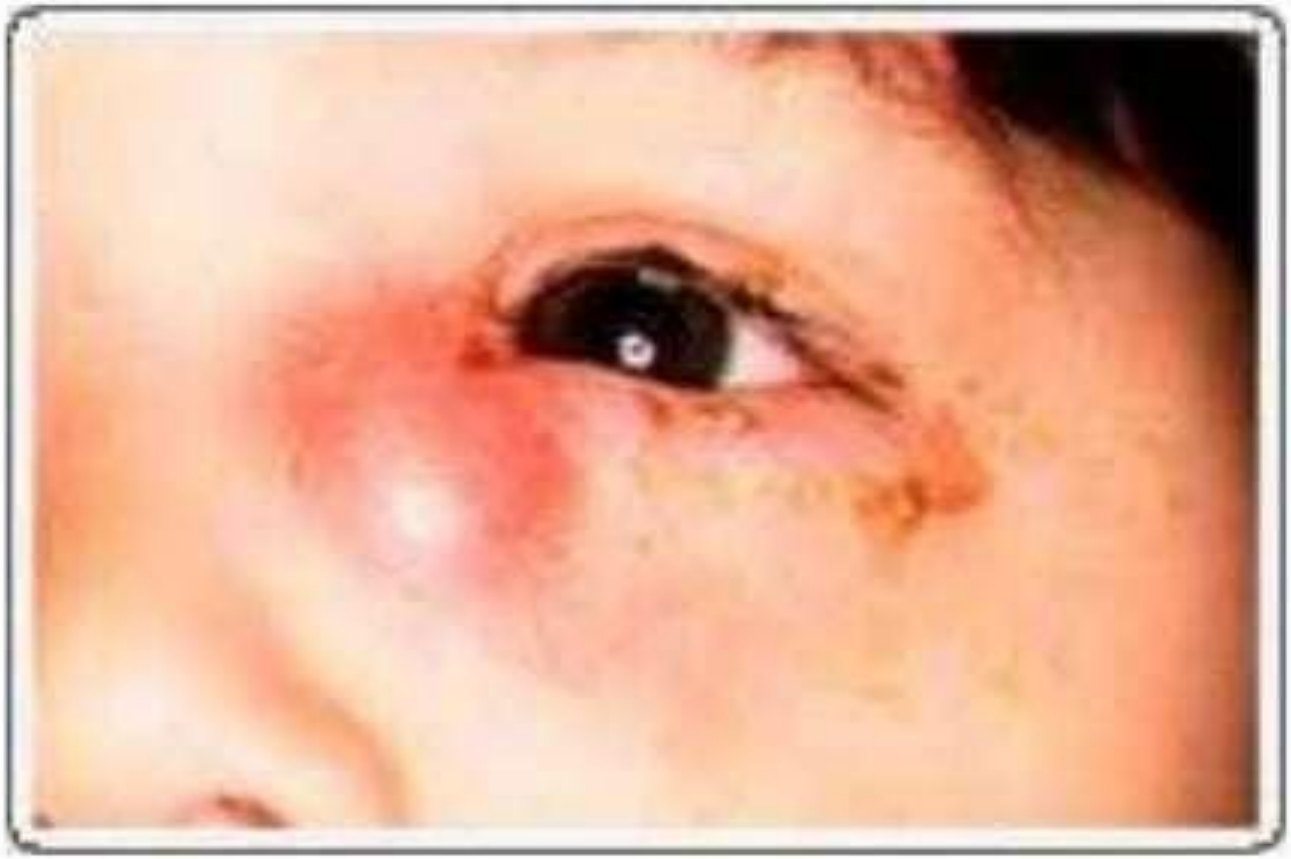


LACRIMAL SYSTEM

≠ CONGENITAL DACRYOCYTITIS

CONGENITAL DACRYOCYTITIS

- It is an inflammation of the lacrimal sac occurring in newborn infants; and thus also known as *dacryocystitis neonatorum*.



ORBIT

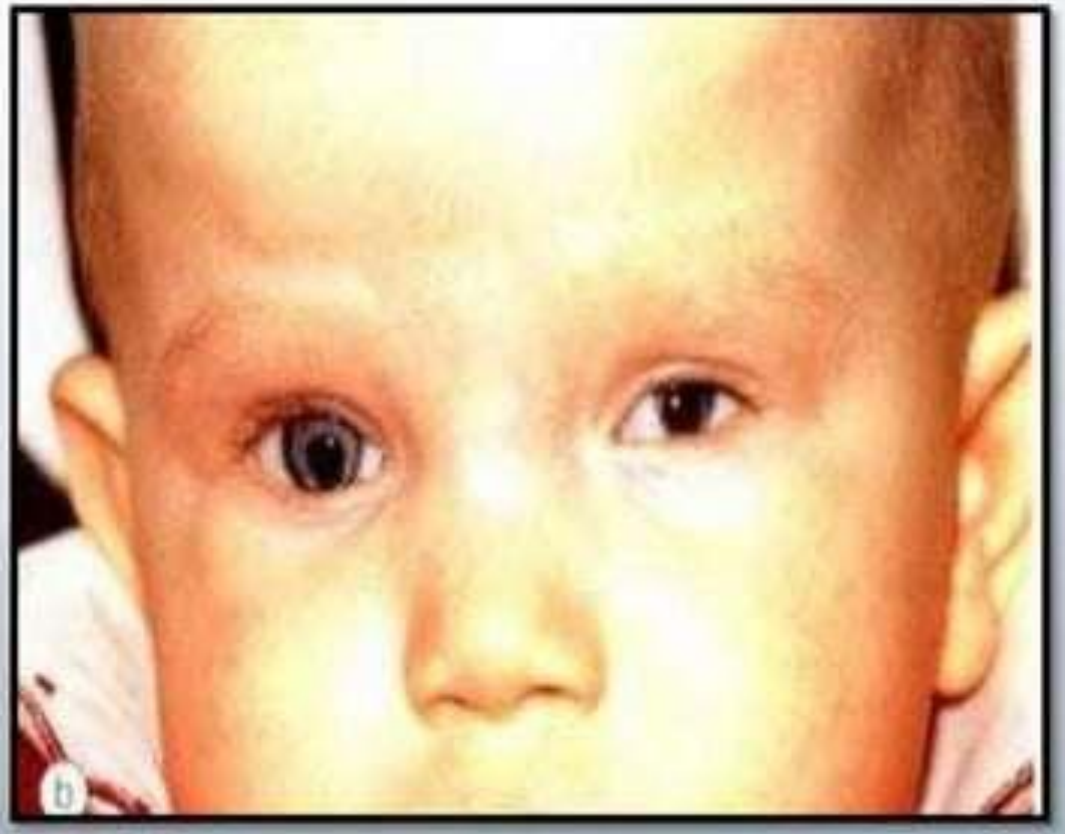
* MICROPHTHALMOS
* ANOPHTHALMOS

MICROPHTHALMOS

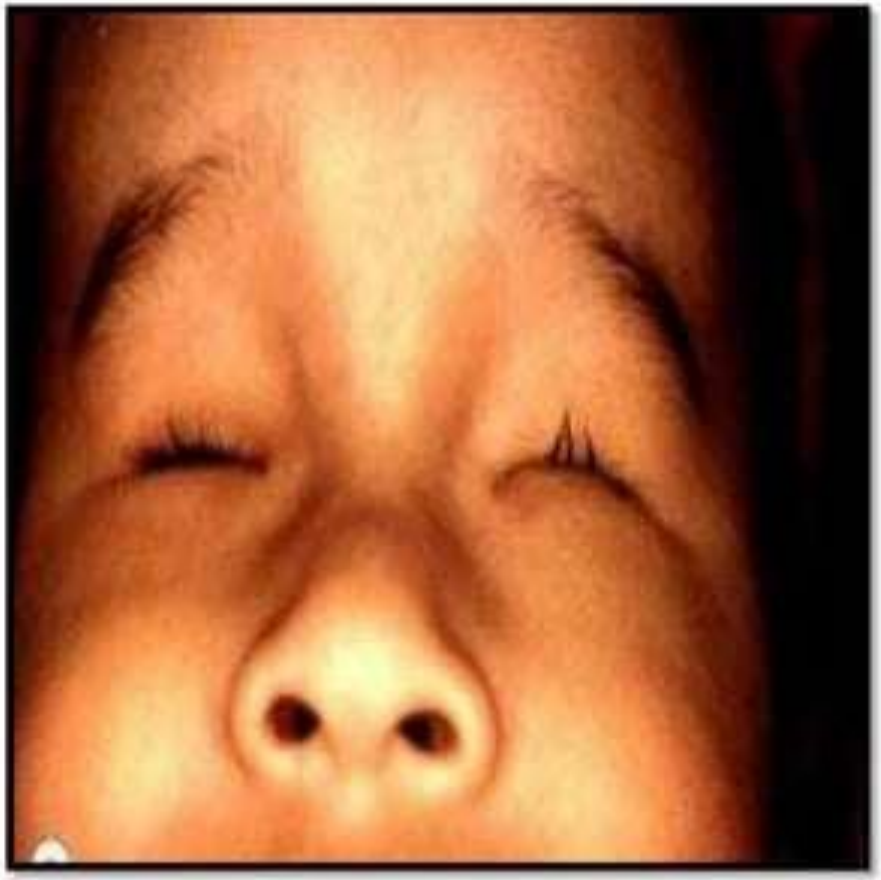
- Microphthalmos is a result of developmental arrest of ocular growth , defined as total axial length atlist 2 deviation below age similar controls.
- It may be unilateral or bilateral.

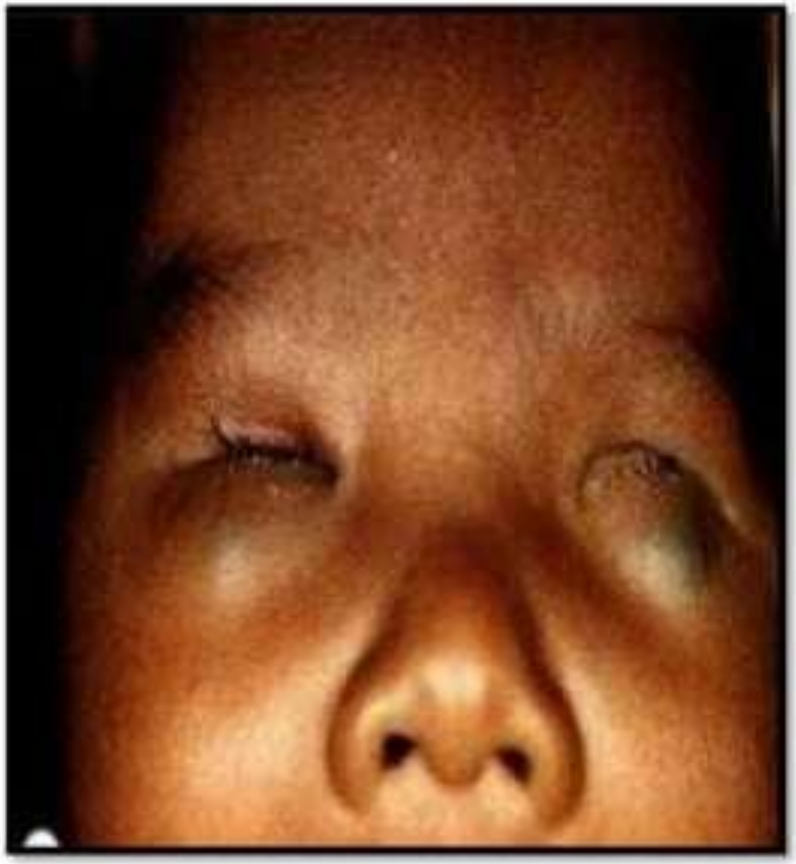
(1) Simple : it is not associated with other major ocular malformation.

(2) Complex : it is associated with coloboma usually of the iris.



ANOPHTHALMOS






Thank
you