
بسم الله الرحمن الرحيم

CONGENITAL UPPER EYELID COLOBOMA

MAJOR REVIEW

OPHTHAL PLAST RECONSTR SURG, VOL. ٣١, NO. ١, ٢٠١٥

استادراهنما:جناب آقای دکترملازاده
ارایه دهنده:فرشته جوانمردی (رزیدنت پزشکی خانواده)

METHODS OF LITERATURE SEARCH

- English and world medical literatures were searched from ١٨٨٠ up through May ٢٠١٤ using Google
- .We used the following keywords: upper eyelid coloboma, colobomata, Fraser syndrome, cryptophthalmos, cryptophthalmia, cryptophthalmos, Goldenhar syndrome, oculoauriculovertebral spectrum, HFM, epibulbar
- .Articles in all languages were considered. For articles in French, we carried out the translation ourselves (with extreme difficulty) and for German articles, a professional medical translator was hired
- Important references in these articles were retrieved and reviewed.

COLOBOMA:

- derived from the Greek word Κολόβωμα ➡ a defect in a tissue ➡ exclusively used in Ophthalmology

could involve any layer of the eye

Eyelid colobomas, however, can be unilateral or or asymmetrical, bilateral,

can involve animals or humans

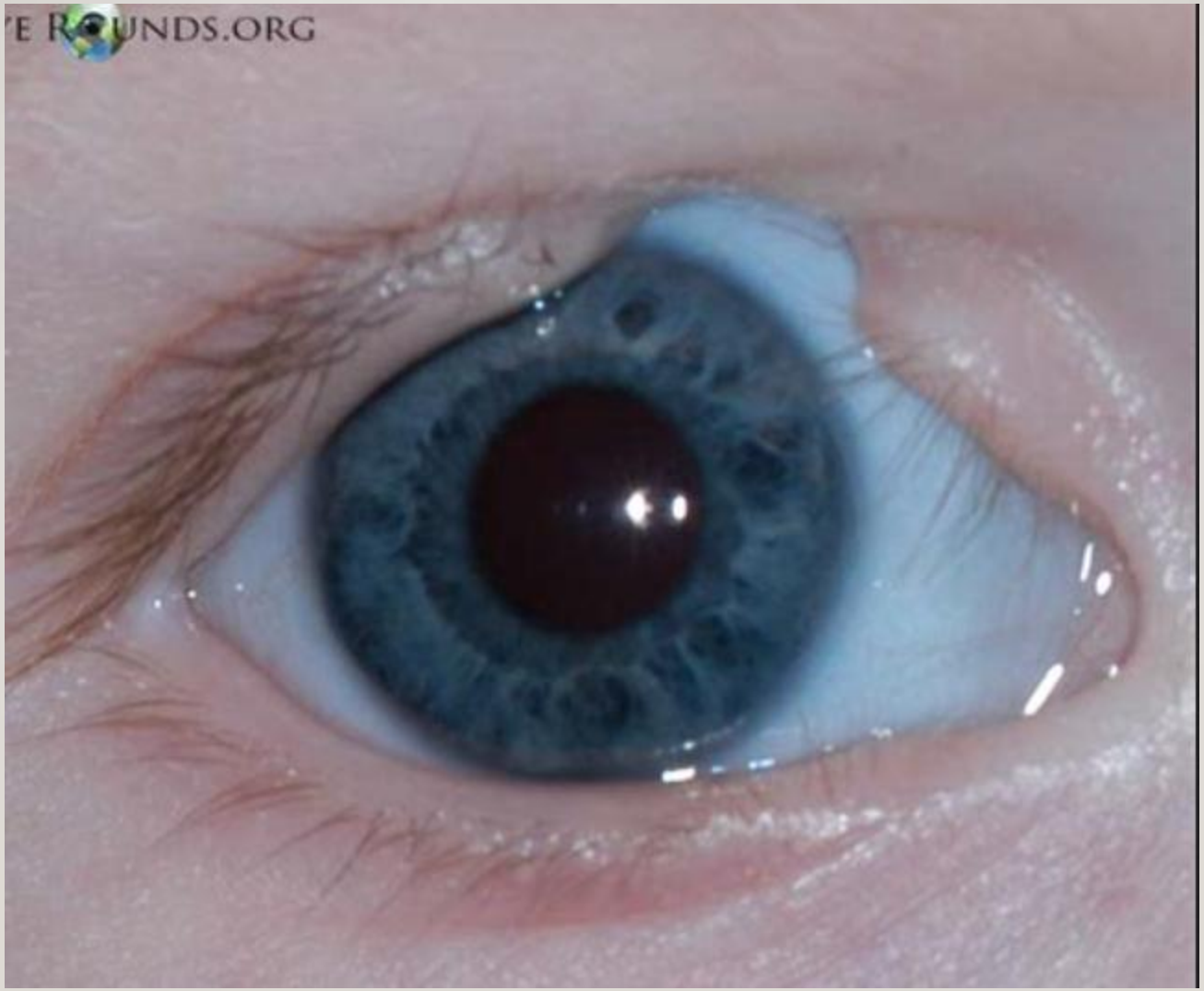
may threaten vision at a very early age

CLINICAL PICTURE

- Isolated Coloboma → Complete CO. (cryptophthalmos)
  incomplete cryptophthalmos
-
- Simple Coloboma

- SYNDROMIC variants**(Fraser** Syndrome , **Goldenhar** Syndrome)(Rare syndromes:**CHARGE** syndrome, . Amniotic band sequence, Neurocutaneous syndromes,
- **Alcoholic syndrome**.)

- The classic congenital upper eyelid defect includes a shortage of conjunctiva, tarsal plate, orbicularis muscle, and skin, which leaves the cornea unprotected, resulting in possible exposure keratopathy.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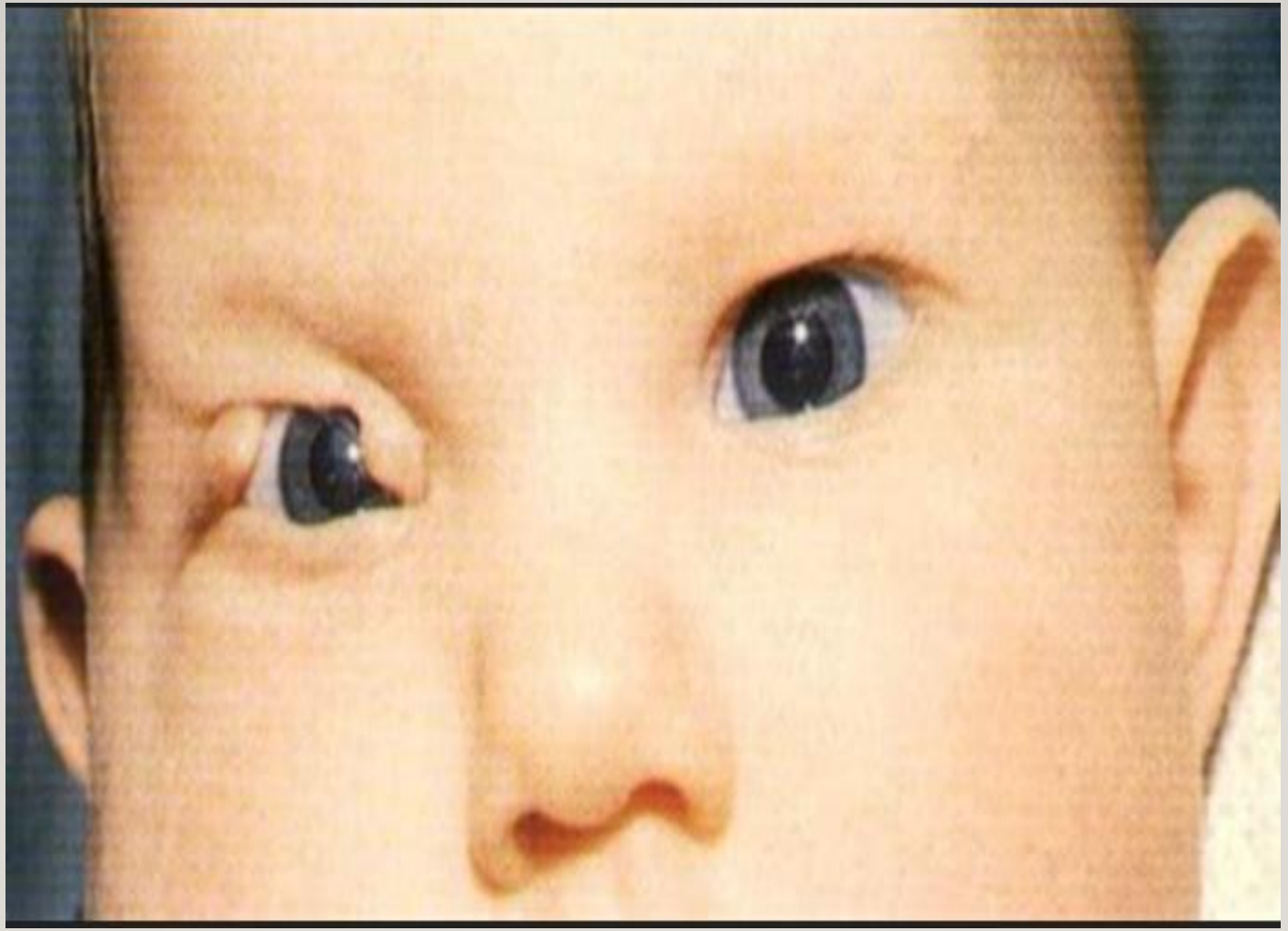
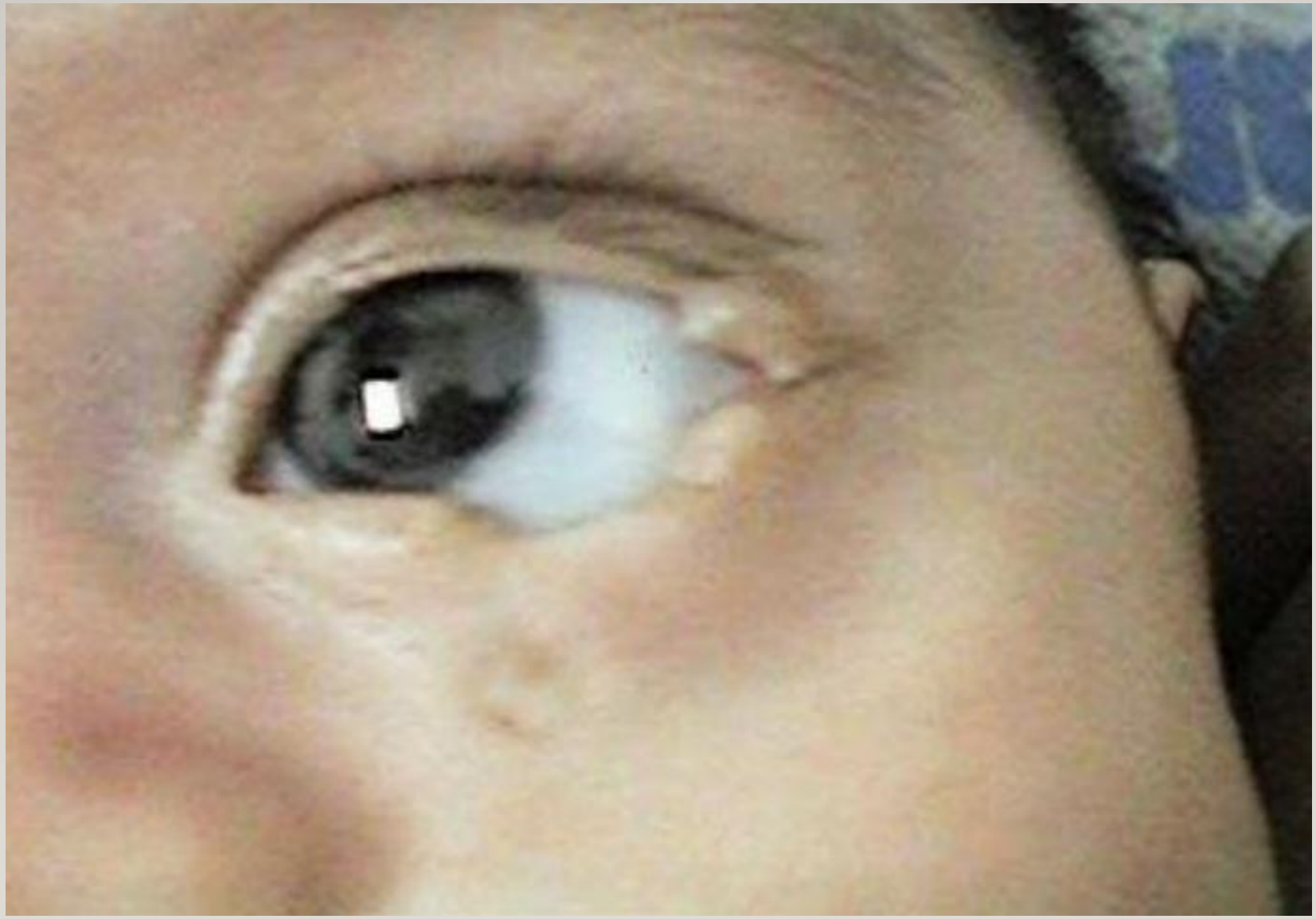
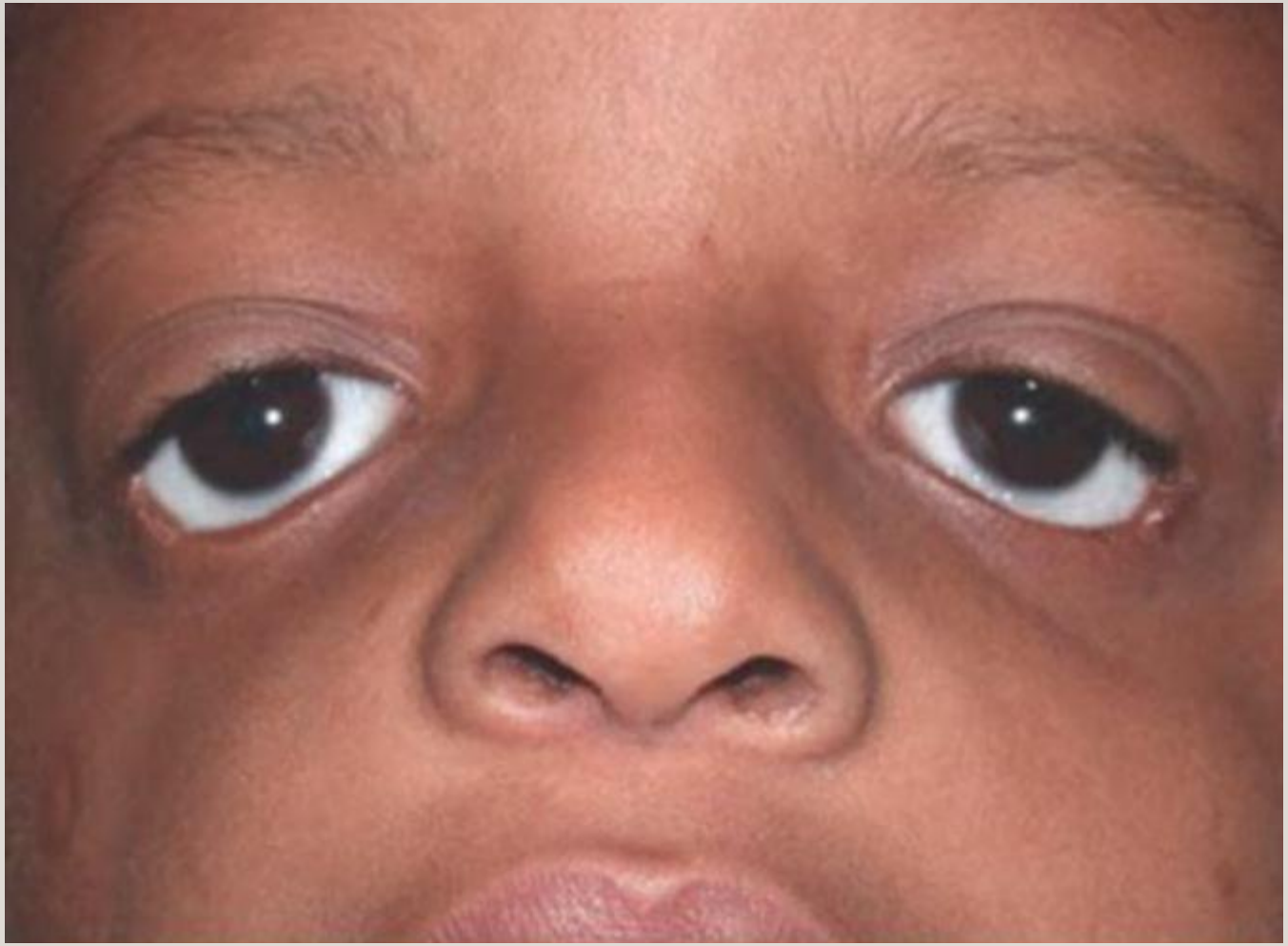
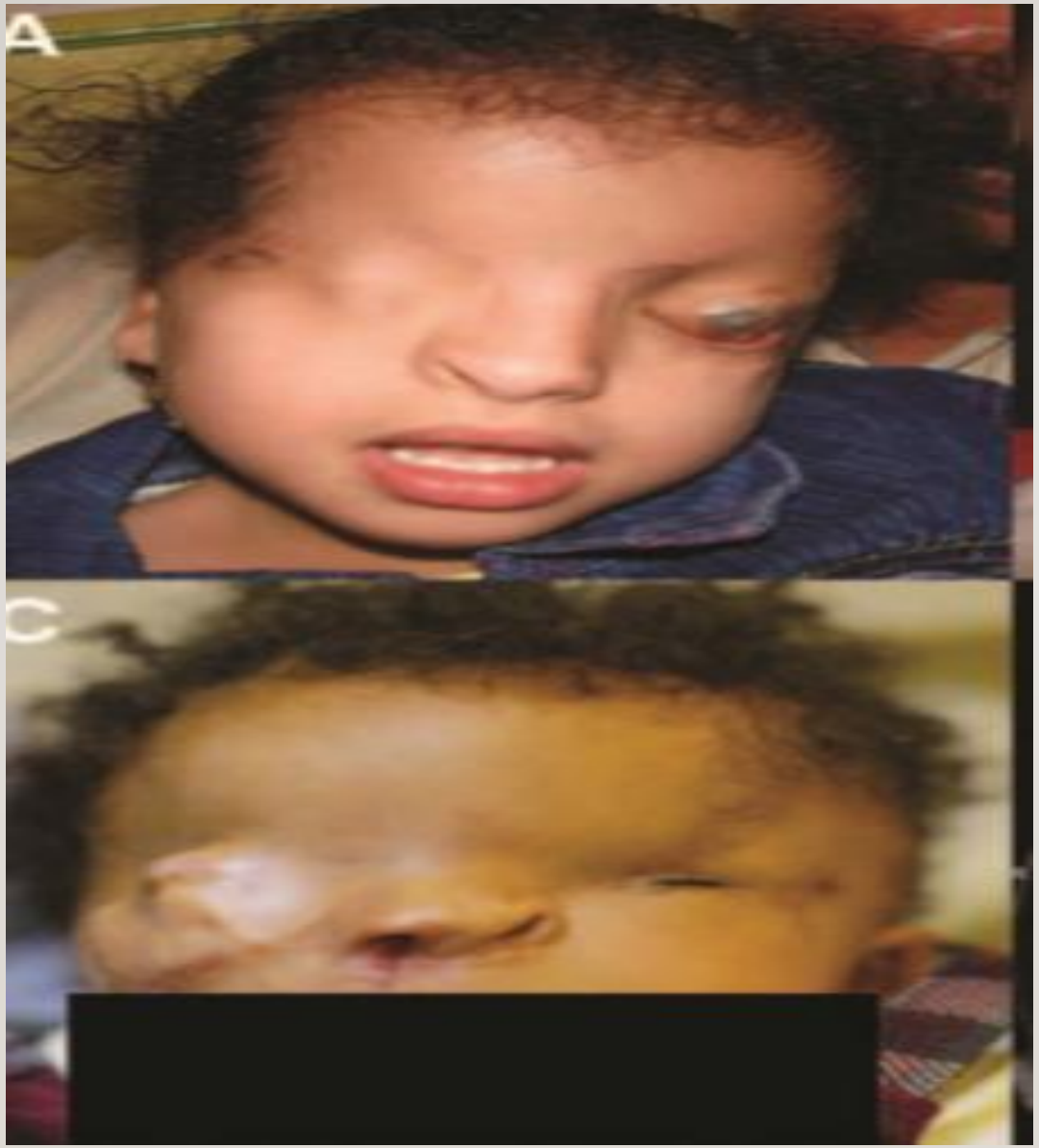
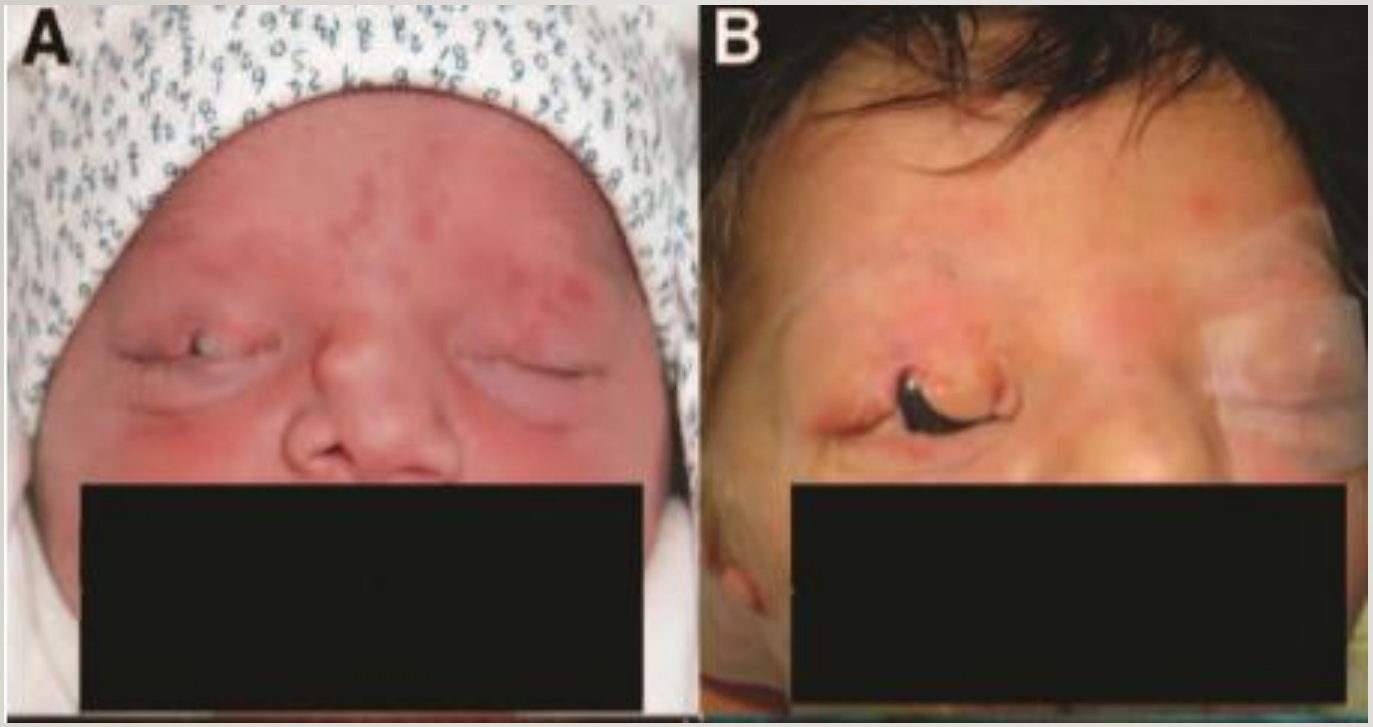




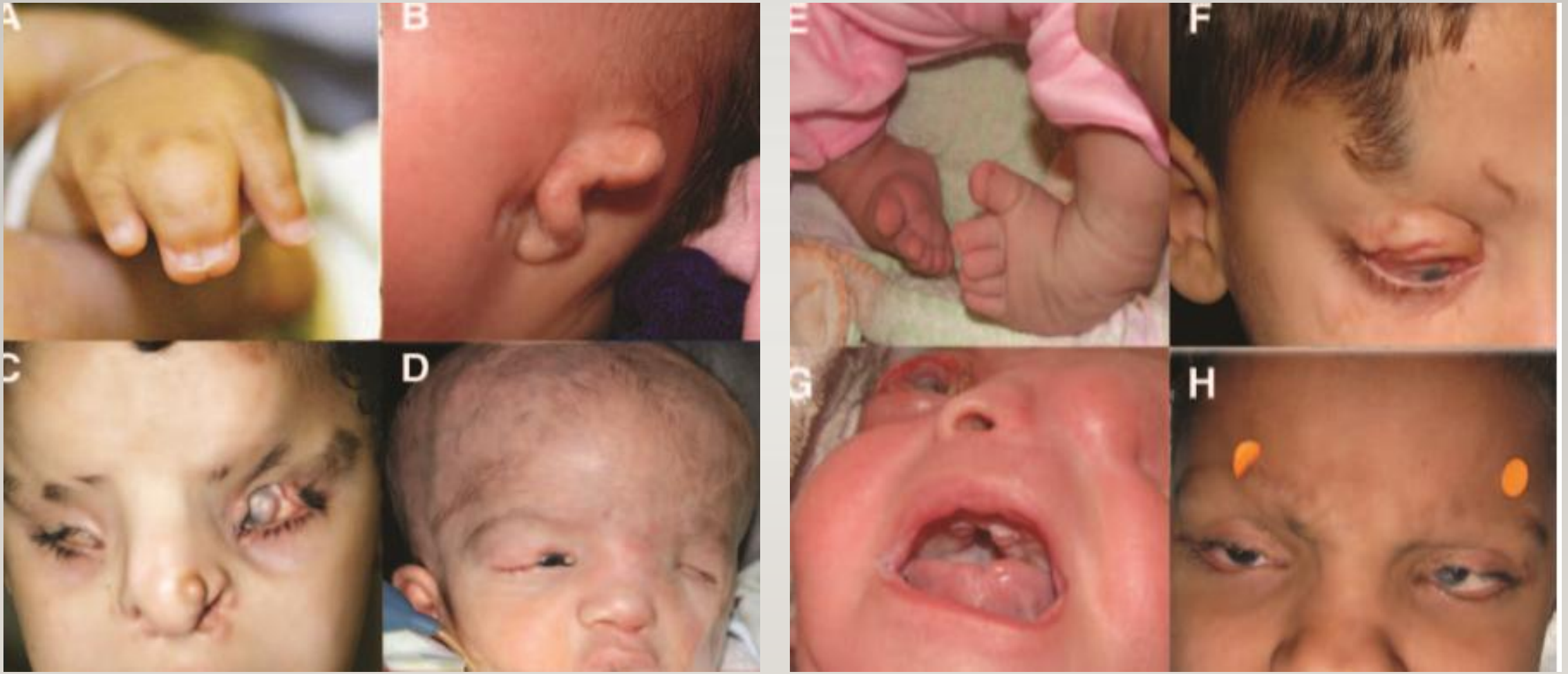




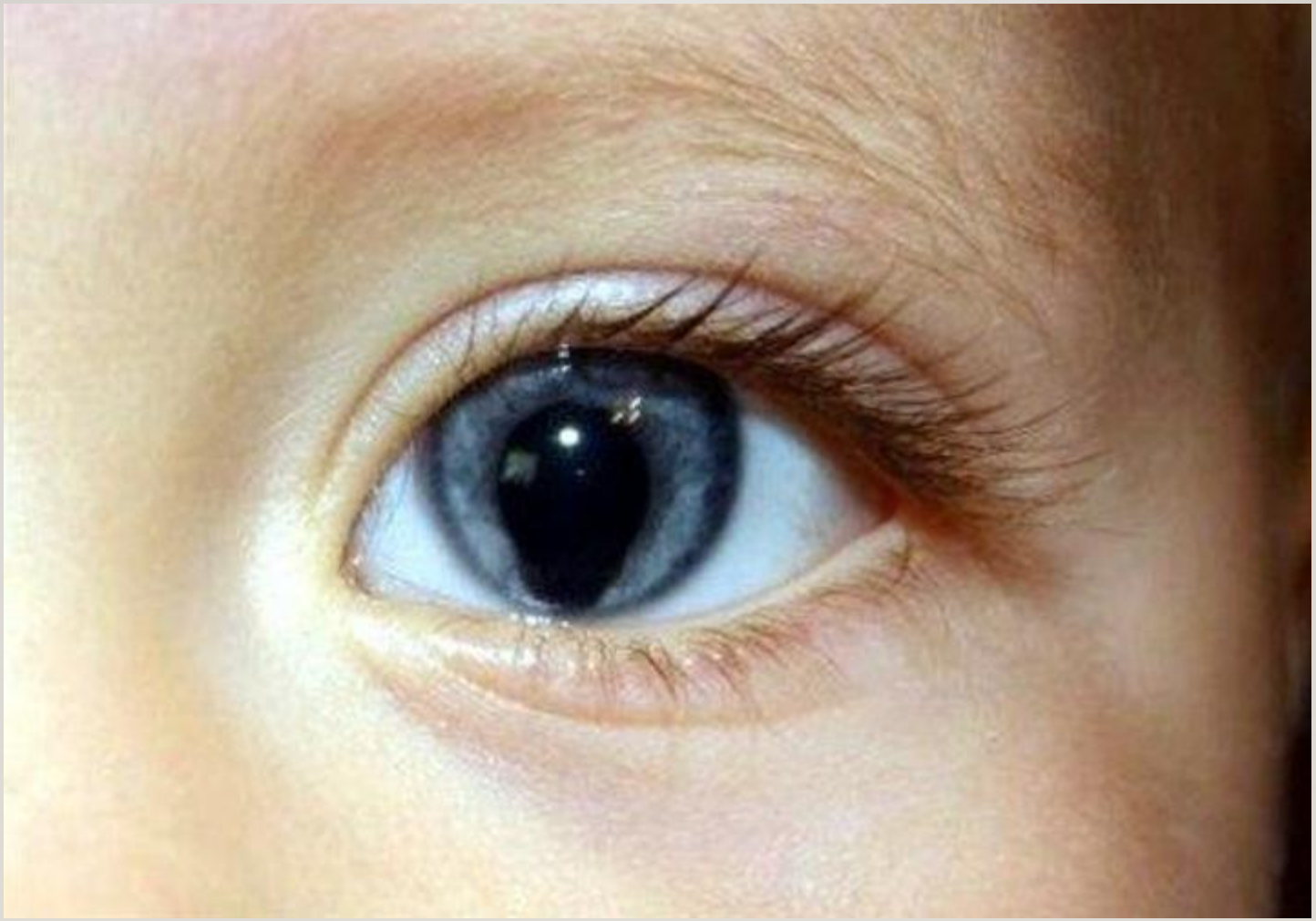
Complete cryptophthalmos

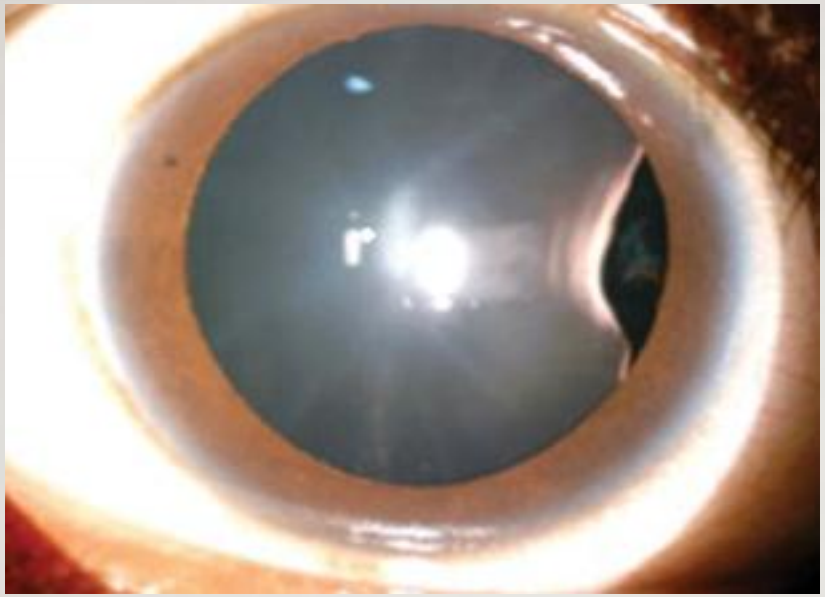


incomplete cryptophthalmos

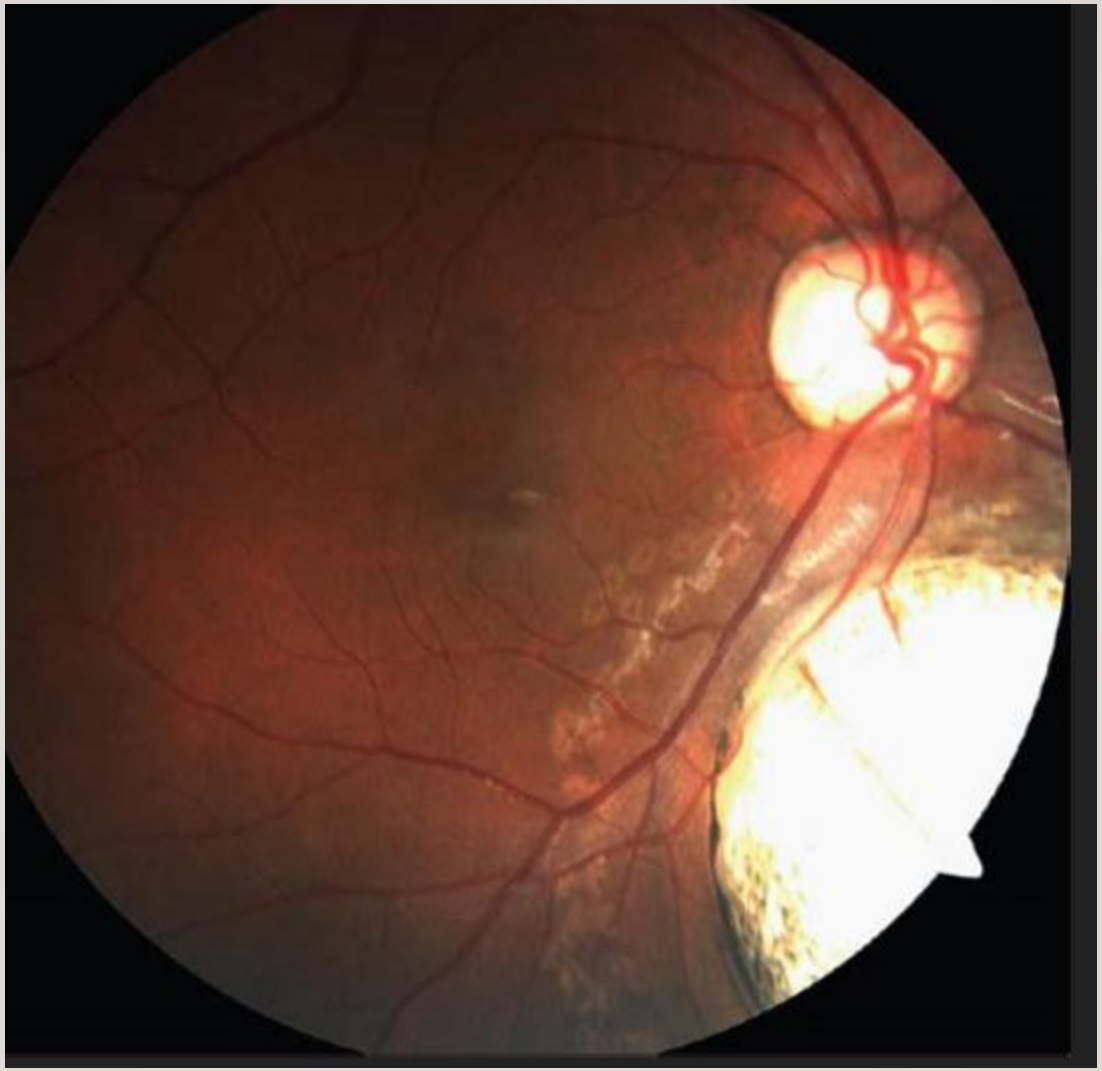


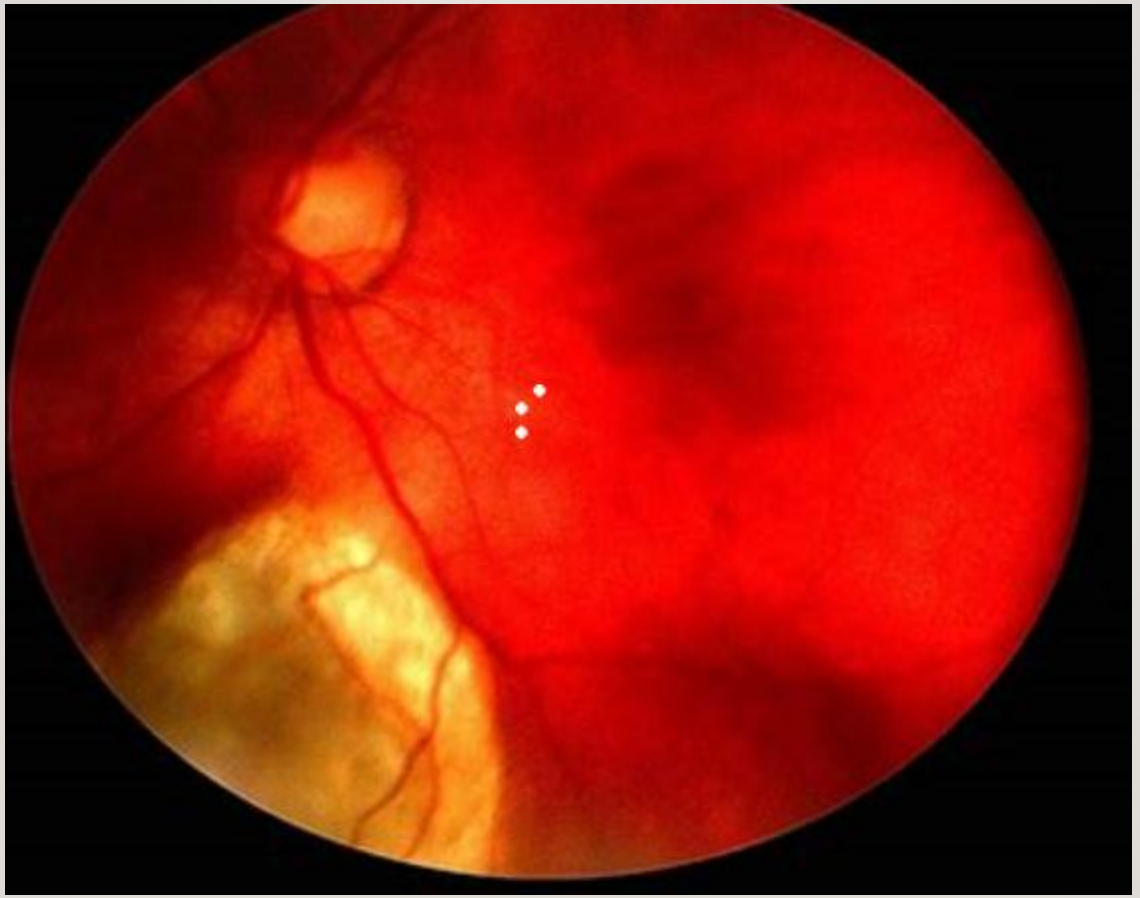
Systemic features of Fraser syndrome






EyeRounds.org





EPIDEMIOLOGY

- The number of cases is around ٠,٥ to ٠,٧ per ١٠,٠٠٠ births, making it a relatively rare condition
- In their comprehensive review in ١٩٨٦, Thomas et al.[٣٧] reported a total of ٢٧ patients with isolated CO with equal sex distribution of whom ١٦ were sporadic and ١١ were familial.
- Fraser Syndrome:fewer than ٣٠٠ cases
- Goldenhar Syndrome:from ١/٣,٥٠٠ to ٣,٨/١٠٠,٠٠٠

CAUSES

- Coloboma can be associated with a mutation in the *PAX ٢* gene
- Eye abnormalities have been shown to occur in over ٩٠% of children with fetal alcohol syndrome

SIGNS AND SYMPTOMS

- The effects a coloboma has on the vision can be mild or more severe depending on the size and location of the gap
- Sometimes, the eye may be reduced in size, a condition called microphthalmia. Glaucoma, nystagmus, scotoma, or strabismus may also occur.

MANAGEMENT

- emergencies
- very early age
- For small defects that do require suturing (up to ٢٥%), direct closure may suffice

For larger defects (٥٠% or more of the eyelid)(flap)

١. EMERGENCIES

٢.COULD INVOLVE ANY LAYER OF THE EYE

٣.SYNDROMICVARIANTS