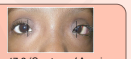
6.17.1. Eyelid Disorders (I): Congenital Eyelid Disorders (I)

Introduction
- ± isolated
- ± associated with orbital malformations
- ± part of a syndrome
  - evaluation of eyelids, ocular adnexa, and interpupillary distance is an essential part of clinical evaluation of a dysmorphic infant
- morphologic measurements of the eyelids and orbit
  - transparent rulers or calipers
  - compared with normal reference measures
    -
    - Figure 17-1 (Courtesy of American Academy of Ophthalmology)
  - Farkas canthal index
    - hypotelorism → <28
    - hypertelorism → >42
    - ratio of inner canthal distance to outer canthal distance × 10

Telecanthus
- greater-than-normal distance between the inner canthi
- types
  - 1 primary → interpupillary distance is normal
  - 2 secondary → interpupillary distance is greater than normal
- common in many syndromes

Dystopia Canthorum
- lateral displacement of both the inner canthi and the lacrimal puncta
  - an imaginary vertical line connecting upper and lower puncta crosses the cornea
- Waardenburg syndrome type 1
- 
  - Figure 17-2 (Courtesy of American Academy of Ophthalmology)

Ablepharon
- absence or severe hypoplasia of the eyelids
- very rare
- at high risk for exposure keratopathy
- ablepharon macrostomia syndrome

Congenital Coloboma of the Eyelid
- from a small notch to a defect encompassing the horizontal length of eyelid
  -
  - Figure 17-4 (Courtesy of American Academy of Ophthalmology)
- usually involves the upper eyelid
- eyelid may be fused to the globe
- unrelated to other ocular colobomas
- medial upper eyelid
- isolated
- ? associations
  - lower eyelid
    - facial clefts (eg, Goldenhar syndrome)
      - Goldenhar syndrome (oculoauriculovertebral dysgenesis)
        - Upper lid coloboma
        - Epibulbar dermoid
        - Preauricular skin tags
        - Vertebral anomalies
        - Lower lid coloboma
        - downward palpebral fissure
        -
        - Figure 3-1 (Courtesy of American Academy of Ophthalmology)
      - mandibulofacial dysostosis (Treacher Collins–Franceschetti syndrome)
    - amniotic band syndrome
    - lacrimal deformities
- should be observed for exposure keratopathy
- surgical closure of the eyelid defect is required in most cases

Cryptophthalmos
- rare
- isolated
- forms
  - Fraser syndrome
    - autosomal recessive
      - mutations in the FRAS1 gene
        - 4q21
        - encodes a putative extracellular matrix protein
    - acrofacial malformations
      - partial syndactyly
    - genitourinary anomalies
- blends in with the cornea, which is usually malformed
- skin passes uninterrupted from the forehead, over the eye, to the cheek
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  - Figure 17-3 (Courtesy of American Academy of Ophthalmology)

Congenital Tarsal Kink
- entropion
  - ulceration
  - cornea may be exposed
- tarsal plate of upper eyelid is folded at birth
- managment
  - minor defects
    - manually unfolding the tarsus
    - taping the eyelid shut with a pressure dressing for 1–2 days
  - more severe defects
    - surgical incision of the tarsal plate
    - excision of a V-shaped wedge from the inner surface

Epiblepharon
- most commonly, the lower eyelid
- horizontal fold of skin adjacent to either the upper or lower eyelid
- may turn the lashes inward
- cornea often tolerates this condition surprisingly well
- management
  - often resolves spontaneously
  - ocular lubricants
  - surgical repair
- cases that do not resolve or that cause chronic corneal irritation

Congenital Entropion
- rare
- eyelid inversion is present at birth
- does not resolve spontaneously
- when corneal integrity is threatened → surgery

Congenital Ectropion
- usually involves the lower eyelid
- eversion of the eyelid margin
- secondary to a vertical deficiency of the skin
- treatment
  - lateral tarsorrhaphy → mild cases
  - skin flap or graft → severe cases

Ankyloblepharon
- fusion of part or all of the eyelid margins
- ± dominantly inherited
- treatment is surgical
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  - Figure 17-5 (Courtesy of American Academy of Ophthalmology)
- eyelid margins are connected by fine strands
- ankyloblepharon filiforme adnatum
- Hay-Wells syndrome
  - ankyloblepharon–ectodermal dysplasia–clefting syndrome
  - includes cleft lip or palate

Ophthalmology Mind Maps/Arman Mashayekhi, MD