7.10.2. Congenital Eyelid Anomalies (II)

Congenital Coloboma

- true coloboma
  - includes defect in the eyelid margin
    - Figure 10-9 (© 2020 American Academy of Ophthalmology)
- associations
  - medial upper eyelid
    - isolated
      - ?
  - lower eyelid
    - facial clefts (eg, Goldenhar syndrome)
      - Goldenhar syndrome (oculoauriculovertebral dysgenesis)
        - Upper lid coloboma
        - Epibulbar dermoid
        - Preauricular skin tags
        - Vertebral anomalies
        - oculoauriculovertebral dysgenesis)
      - mandibulofacial dysostosis (Treacher Collins–Franceschetti syndrome)
        - Lower lid coloboma
        - downward palpebral fissure
        - Figure 3-1 (© 2020 American Academy of Ophthalmology)
    - lacrimal deformities
- management
  - full-thickness defects ≤ 1/3 of eyelid
    - creating raw vertical margins and sliding flaps along the eyelid crease
    - ± lateral cantholysis
  - large defects
    - variation of lateral canthal semicircular flap
  - eyelid-sharing procedures that occlude the visual axis are avoided
    - risk of amblyopia !

Congenital Entropion

- eyelid margin inversion
  - Figure 10-7 (© 2020 American Academy of Ophthalmology)
- pathogenesis
  - lower eyelid retractor dysgenesis
  - structural defects in the tarsal plate
  - relative shortening of the posterior lamella
- management
  - unlikely to improve spontaneously
  - may need surgical correction
    - removal of a small amount of skin and orbicularis along the subciliary portion of the eyelid
      - +
        - advancement of the lower eyelid retractors to the tarsus
- tarsal kink
  - unusual form of congenital entropion
  - tarsal plate of the upper eyelid is folded
  - treatment
    - removal of the kink in combination with a margin rotation
    - ± skin grafting for the anterior lamella

Congenital Distichiasis

- rare
- ± hereditary
- embryonic pilosebaceous units differentiate into hair follicles
- extra row of eyelashes in place of the orifices of the meibomian glands
  - Figure 10-8 (© 2020 American Academy of Ophthalmology)
- treatment
  - indications
    - symptomatic patient
    - keratopathy
  - lubricants
  - soft contact lenses
  - electrolysis
  - cryoepilation
  - eyelid splitting with removal of the follicles

Infantile (Capillary) Hemangioma

- natural course
  - most are not apparent at birth
  - usually appear weeks or months after birth
  - increase in size until age 1 year
  - decrease over the next 3–7 years
- ± involve the orbit
- associated with a high incidence of amblyopia
- indications for treatment
  - occlusion of the visual axis
  - anisometropia
  - strabismus
  - significant disfigurement
- lesions limited to the eyelid
  - topical timolol gel
    - fewest adverse effects
  - intralesional steroids
    - for patients who cannot take β-blockers or do not respond to timolol
    - act by rendering the tumor's vascular bed more sensitive to the body's circulating catecholamines
    - relatively safe, simple, and repeatable
    - rare complications
      - eyelid necrosis
      - embolic retinal vascular occlusion
      - systemic adrenal suppression
- treatment
  - more widespread
    - systemic propranolol
    - oral corticosteroids
      - eliminates the risks attributed to intralesional injection
      - systemic adverse effects
  - other treatment options
    - topical clobetasol propionate
      - does not eliminate the systemic side effects
    - interferon-α
      - for life-threatening or sight-threatening lesions unresponsive to other forms of treatment
      - serious adverse effects
    - surgical excision
      - rare well-circumscribed lesions
    - carbon-dioxide laser
      - for controlling bleeding when lesions must be removed
    - topical skin lasers
      - superficial (1–2 mm) layers of the skin to diminish lesion redness
      - do not penetrate deeply enough to shrink a visually disabling lesion

Cryptophthalmos

- rare
- forms
  - isolated
  - recessive syndromic
    - Fraser syndrome
      - autosomal recessive
      - acrofacial malformations
        - partial syndactyly
      - genitourinary anomalies
      - +- cryptophthalmos
    - mutations in the FRAS1 gene
      - 4q21
      - encodes a putative extracellular matrix protein
- clinical presentation
  - unilateral or bilateral
    - usually bilateral
  - severe ocular defects are present in the underlying eye
  - eyelids and associated structures of the brows and lashes fail to form (ablepharon)
    - Figure 9-1 (© 2020 American Academy of Ophthalmology)
    - Figure 10-10 (© 2020 American Academy of Ophthalmology)
    - partial or complete absence of
      - eyebrow
      - palpebral fissure
      - eyelashes
      - conjunctiva
      - levator
      - orbicularis oculi muscle
      - tarsus
      - meibomian glands
  - cornea is merged with the epidermis
  - conjunctiva is typically absent
  - anterior chamber, iris, and lens are variably formed or are absent
  - associated ocular findings
    - absence of the lacrimal glands and canaliculi
    - corneal and conjunctival dermoid
- pseudocryptophthalmos
  - eyelids and associated structures form but fail to separate (ankyloblepharon)
- treatment
  - surgical intervention
    - cosmesis
    - relief of pain from absolute glaucoma
  - pseudocryptophthalmos
    - eyelid reconstruction
    - fornix reconstruction
  - attempts at reconstruction are difficult

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