Blepharophimosis–ptosis–epicanthus inversus syndrome

Case Description
young child brought for bilateral abnormal eyelid appearance and upper eyelid droopiness

Image Description

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external photograph shows bilateral ptosis, epicanthus inversus, and telecanthus

Differential Diagnosis
Blepharophimosis–ptosis–epicanthus inversus syndrome
- AD
  - chromosome 3
  - FOXL2 mutation
  - can transmit to children
- blepharophimosis
  - palpebral fissures are shortened horizontally and vertically
- severe ptosis
- telecanthus
- epicanthus inversus
  - fold of skin extending from the lower to upper eyelid
- poorly developed nasal bridge
- lateral lower lid ectropion
- hypoplasia of the superior orbital rims
- hypertelorism
- ear deformities
- systemic associations
  - premature ovarian failure
    - type 1
  - cardiac defects

isolated blepharophimosis

congenital ptosis

facial dysmorphisms
- Crouzon syndrome
  - craniosynostosis

Data acquisition
History
- family history of similar condition
- ★ growth & developmental delay
- ★ cardiac disease

Physical Exam
- VA
  - high rate of amblyopia
- EOM
  - high rate of strabismus
- head positioning
- head & neck exam
  - cleft palate, high-arched palate
  - ear abnormalities
    - cup-shaped ears
- lacrimal system
- corneal epitheliopathy
- ★ optic disc coloboma
- refraction
  - high rate of refractive error & anisometropia

Assessment
Blepharophimosis–ptosis–epicanthus inversus syndrome

Management
Medical
- glasses for refractive error
- amblyopia treatment
- lubrication for corneal epitheliopathy

Surgical
multi-stage plan
- epicanthal folds ①
  - may exacerbate the ptosis
  - double Z plasty, Y-Z plasty + transnasal wiring of medial canthal tendon
- ptosis repair ②
  - brow suspension or frontalis suspension
  - can be performed simultaneously with repair of telecanthus
  - visually obstructive ptosis should be corrected promptly

Referrals
- refer to geneticist
- refer to cardiologist
- refer female patients to endocrinologist

Patient Education/ Counselling
Diagnosis/Pathogenesis
- genetic disease
  - AD
  - can transmit to children

Natural course
- epicanthus and telecanthus may improve with age

Follow-up
- for refractive error and amblyopia

Ophthalmology Mind Maps/Arman Mashayekhi, MD