7.3.1. Congenital Orbital Anomalies

Craniofacial Clefting and Syndromic Congenital Craniofacial Anomalies

- **clefting syndromes affecting orbit and eyelids**
  - oculoauriculovertebral spectrum
    - hemifacial microsomia
    - oculoauriculovertebral dysplasia (Goldenhar syndrome)
    - mandibulofacial dysostosis (Treacher Collins–Franceschetti syndrome)
      - Figure 3-1 (© 2020 American Academy of Ophthalmology)
    - some forms of midline clefts with hypertelorism
- **craniosynostosis**
  - types
    - isolated
      - premature closure of 1 or more sutures in the bones of the skull
    - syndromic
      - genetically heterogeneous disorder
        - fibroblast growth factor receptor 2 (FGFR2) genes
          - autosomal dominant
          - chromosome 10
      - frequently associated with
        - hypertelorism
        - proptosis
      - Crouzon syndrome (craniofacial dysostosis)
        - most common AD craniosynostosis syndrome
        - Figure 3-2 (© 2020 American Academy of Ophthalmology)
        - no syndactyly/polydactyly
      - Apert syndrome (acrocephalosyndactyly)
        - syndactyly > polydactyly
        - Figure 3-3 (© 2020 American Academy of Ophthalmology)
      - Pfeiffer syndrome
        - milder manifestations
      - Saethre-Chotzen syndrome
        - TWIST gene
          - chromosome 7
  - complications
    - various skeletal deformities
      - including orbital defects
    - ophthalmic problems
      - strabismus
      - astigmatism
      - ptosis
      - exophthalmos
      - nasolacrimal duct obstruction
      - amblyopia
    - secondary intracranial hypertension
- **intracranial contents can herniate through congenital clefts of skull or orbit**
  - types
    - meningocele
    - encephalocele
    - meningoencephalocele
  - most commonly present anteriorly with subcutaneous protrusion near the medial canthus or over the bridge of the nose
    - straining or crying may increase the size of the mass
    - globe may be displaced temporally and downward (inferolaterally)
  - less commonly move into the posterior orbit
    - anterior displacement and pulsation of the globe
  - meningoceles/encephaloceles adjacent to the orbit are frequently associated with anomalies of the optic disc
    - morning glory disc
  - treatment
    - collaboration with a neurosurgeon
- treatment
  - bony and soft tissue reconstruction
  - operations are staged and require a team approach
- developmental arrest or mechanical disruption of development
  - failure of neural crest cell migration
  - failure of fusion or movement of facial processes
- skeletal facial clefts are distributed around the orbit and maxilla
- soft tissue clefts are most apparent around the eyelids and lips

Introduction

- most significant congenital anomalies of the eye and orbit are apparent on ultrasonography before birth
- the more profound the abnormality, the earlier in development it occurred
- types
  - slowing/cessation of a normal stage of development
    - pure arrest
    - e.g. microphthalmia
  - superimposed aberrant growth following the original arrest
    - resulting deformity does not represent any previous normal stage of development
    - e.g. orbital cyst following incomplete closure of the fetal fissure

Anophthalmia

- total absence of tissues of the eye
- primary anophthalmia
  - rare
  - usually bilateral
  - primary optic vesicle fails to grow out from the cerebral vesicle
- secondary anophthalmia
  - rare
  - lethal
  - gross abnormality in the anterior neural tube
- consecutive anophthalmia
  - secondary degeneration of the optic vesicle
- orbital development is partially dependent on the size and growth of the globe
  - orbital bones, eyelids, and adnexal structures also fail to develop

Microphthalmia

- much more common than anophthalmia
  - most infants with a unilateral small orbit and no visible eye actually have a microphthalmic globe
- small eye with axial length ≥2 standard deviations below the mean axial length for age
  - vary in size depending on the severity
- types
  - isolated
  - part of a well-defined syndrome
    - multiple genetic mutations have been reported
      - developmental phenotype !

Treatment of Anophthalmia/Microphthalmia

- most microphthalmic eyes have no potential for vision
- all children with microphthalmia have hypoplastic orbits
- treatment focuses on achieving a cosmetically acceptable appearance that is reasonably symmetrical
- treatment begins shortly after birth
- techniques
  - socket expansion with progressively larger conformers
    - until the patient can be fitted with a prosthesis
    - enucleation is usually not necessary for the fitting of a conformer or an ocular prosthesis
      - enucleation should be avoided because it may worsen the bony hypoplasia
  - orbital volume augmentation with autogenous materials
    - dermis-fat grafts
      - may grow with the child, resulting in progressive socket expansion
    - synthetic implants
  - intraorbital tissue expanders
    - progressively inflated to enlarge the hypoplastic orbit
  - craniofacial techniques
    - severe bony asymmetry
    - severe microphthalmia or anophthalmia
    - older children with previously untreated microphthalmia
- microphthalmia with orbital cyst
  - failure of the choroidal fissure to close in the embryo
  - usually unilateral
    - may be bilateral
  - orbital cyst may stimulate normal growth of involved orbital bone and eyelids
  - in some cases, orbital cyst may be removed for fitting of an ocular prosthesis

Ophthalmology Mind Maps/Arman Mashayekhi, MD