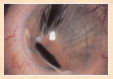
# 8.9.3. Congenital Anomalies (III): Cornea (II): Anterior Segment Dysgenesis

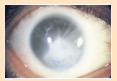
## Posterior embryotoxon

- Schwalbe line is visible in 8%–30% of normal eyes
  - opaque ridge 0.5–2.0 mm central to the limbus
- thickened and anteriorly displaced schwalbe line
  - visible by external examination!
  - bilateral
-
  Figure 9-4 (© 2020 American Academy of Ophthalmology)
- autosomal dominant
- eye is usually normal
- ocular or systemic syndromes
  - arteriohepatic dysplasia (Alagille syndrome)
  - X-linked ichthyosis
  - familial aniridia

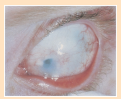
## Axenfeld-Rieger syndrome

- transmission
  - autosomal dominant (75%)
    - chromosome region 6p25 (forkhead genes)
  - sporadic
- clinical presentation
  - anteriorly displaced schwalbe line (posterior embryotoxon)
  - attached iris strands
  - 
    Figure 9-5 (© 2020 American Academy of Ophthalmology)
  - glaucoma
    - 50%
    - late childhood or in adulthood
  - associated skeletal, cranial, facial, and dental abnormalities

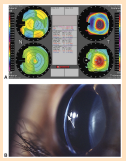
## Peters anomaly

- genetics
  - most cases are sporadic
  - autosomal recessive and dominant
    - mutations in
      - PAX6 gene (11p13)
      - PITX2 gene (4q25-26)
      - CYP1B1 gene (2p22-21)
      - FOXC1 gene (6p25)
    - See under Axenfeld-Rieger
- clinical presentation
  - central corneal opacity
  - 
    Figure 9-6 (© 2020 American Academy of Ophthalmology)
  - localized absence of the corneal endothelium and Descemet membrane
  - types
    - type I
      - iridocorneal adhesions
      - better prognosis for vision rehabilitation with corneal transplantation
    - type II
      - cataractous lens or corneolenticular adhesions
  - associated ocular abnormalities
    - congenital glaucoma
    - microcornea
    - aniridia
    - PFV
  - bilateral>unilateral
    - bilateral cases associated with systemic malformations
  - systemic involvement
    - Peters plus syndrome
      - cleft lip/palate
      - short stature
      - external ear abnormalities
      - intellectual disability
      - other systemic malformations
        - heart defects
        - hearing loss
        - central nervous system deficits
        - spinal defects
        - gastrointestinal and genitourinary defects
        - skeletal anomalies
      - systemic malformations may be associated with genetically transmitted syndromes
        - trisomy 13–15
          - not trisomy 21 !
        - Peters plus syndrome
        - Kivlin syndrome
        - Pfeiffer syndrome
        - exception rather than the rule

## Sclerocornea

- 
  Figure 9-8 (© 2020 American Academy of Ophthalmology)
- sporadic
- transmission
- autosomal dominant and recessive
- no sex predilection
- 90%
- bilateral
- corneal periphery or the entire cornea
- scleralization of the cornea
- limbus is usually ill-defined
- superficial vessels cross the cornea
- 80%
- cornea plana
- angle structures are also commonly malformed
- systemic anomalies

## Circumscribed posterior keratoconus

- without any protrusion of the anterior surface!
- 
  Figure 9-7 (© 2020 American Academy of Ophthalmology)
- localized central or paracentral indentation of the posterior cornea
- in contrast to Peter anomaly
- corneal thinning
- overlying stromal haze
- Descemet membrane and endothelium are usually present
- at the margins of the opacity
- focal deposits of pigmentation and guttae
- irregular astigmatism
- amblyopia
- unilateral
- sporadic
- nonprogressive
- similar to Peters plus
- short stature
- intellectual disability
- cleft lip and palate
- vertebral anomalies
- bilateral
- autosomal recessive form

## Keratectasia & congenital anterior staphyloma

- rare
- unilateral
- sporadic
- cornea
  - opaque
  - thin
  - protruded
    - congenital anterior staphyloma
      - lined by uvea
    - keratectasia
      - not lined by uvea
  - absent Descemet membrane and endothelium
- resembles unilateral Peters anomaly
- disorganized anterior segment
- lens occasionally adheres to cornea
- treatment
  - PK
  - sclerokeratoplasty
- poor visual prognosis

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