Developmental Abnormalities of Vitreous II

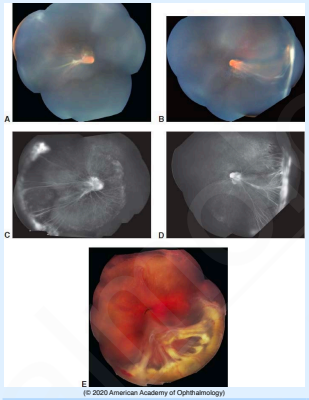
Familial Exudative Vitreoretinopathy

Genetics

- autosomal dominant
  - EVR1 (FDZ4)
  - EVR3
  - EVR4 (LRP5)
  - EVR5 (TSPAN12)
- X-linked
  - EVR2 (NDP)

Clinical Features

- failure of temporal retina to vascularize
  - ± asymmetric
  - bilateral
  - brush border
  - end abruptly a variable distance from the ora
  - dragging & straightening of peripheral retinal vessels
  - peripheral fibrovascular proliferation
    - retinal folds
    - tractional RD
    - pseudoexotropia
    - temporal dragging of the macula
  - exudative RD
  - late-onset rhegmatogenous RD
  - the earlier disease presents, the more severe the manifestations



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Differential Diagnosis

- Differentiation of FEVR from ROP
  - ROP
    - full term
    - normal respiratory status
    - examine all family members
      - family history
      - parents & siblings of affected children may be mildly affected and asymptomatic
      - wide-field angiography
        - straightening of vessels
        - peripheral retinal nonperfusion
          - laser photocoagulation of avascular retina
- dyskeratosis congenita
- Coats plus disease
- facioscapulohumeral muscular dystrophy
- progressive hemifacial atrophy (Parry-Romberg syndrome)

Hereditary Hyaloideoretinopathies With Optically Empty Vitreous

General

- *optically empty vitreous cavity*
- vitreous liquefaction (synchysis)
  - thin layer of cortical vitreous behind lens
  - threadlike, avascular membranes that run circumferentially and adhere to retina
- equatorial and perivascular (radial) lattice degeneration
- ERG response may be subnormal

Wagner disease

- autosomal dominant
- optically empty vitreous cavity
- myopia, strabismus, cataract
- no associated systemic findings
- not associated with retinal detachment

Stickler syndrome

- autosomal dominant
- COL2A1 gene
  - encodes type II procollagen
  - various mutations produce Stickler syndrome phenotypes of differing severity
- most common variety of HHR with associated systemic findings
- other ocular abnormalities
  - myopia, open-angle glaucoma, cataract
- orofacial findings
  - midfacial flattening
  - Pierre Robin malformation complex
    - cleft palate
    - micrognathia
    - glossoptosis
- generalized skeletal abnormalities
  - joint hyperextensibility and enlargement
  - arthritis
  - mild spondyloepiphyseal dysplasia
- high incidence of retinal detachment
  - COL2A1 mutations
    - 91% retinal tear
    - 53% RD
  - difficult to repair
    - multiple, posterior, or large breaks
    - tendency toward proliferative vitreoretinopathy
    - cortical vitreous condensations that are firmly adherent to retina
  - consider prophylactic treatment of retinal breaks

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Weill-Marchesani syndrome

Dwarfism

- some varieties

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