5.4.7. Patient With Decreased Vision: Classification & Management (VII): Congenital Optic Disc Anomalies

Excavated optic disc anomalies

optic pit
- depression of the optic disc surface
- gray or white
- inferotemporal
- paracentral or arcuate
- mild visual field defect
- 25%–75%
- serous detachment of the macula
- liquefied vitreous through communication between the optic pit and the macula

coloboma
- incomplete closure of the embryonic fissure
- inferior
- ± extension to adjacent choroid and retina
- ± visual field defect
- ± RAPD
- iris
- choroid
- ± colobomas of other structures

dysplastic nerve of papillorenal syndrome (renal coloboma syndrome)
- PAX2 gene mutations
- autosomal dominant
- renal failure secondary to renal hypoplasia
- visual acuity is often normal
- emanating and exiting from the disc edge
- multiple cilioretinal vessels
- excavated with absence or attenuation of the central retinal vessels
- superonasal visual field defects

morning glory disc anomaly
- F>M
- most often unilateral
- funnel-shaped staphylomatous excavation of optic nerve and peripapillary retina
- optic disc
  - enlarged
  - pink or orange
  - elevated or recessed within the staphyloma
  - chorioretinal pigmentation surrounds the excavation
  - white glial tissue is present on the central disc surface
  - emanation of retinal vessels from the periphery of the disc
- In contrast to dysplastic nerve
  - can be normal
  - visual acuity ≤20/200
  - RAPD
  - 26%–38%
  - serous retinal detachment
- moyamoya disease
  - basal encephalocele
  - CNS vascular anomalies
  - neuroimaging is warranted

Optic nerve hypoplasia
- teratogens associated with optic nerve hypoplasia
  - quinine
  - ethanol
  - anticonvulsants
    - phenytoin
- clinical presentation
  - 56%–92% bilateral
  - variable visual acuity
  - nearly all eyes have visual field loss
  - optic disc
    - small
      - 1/2 - 1/3 normal diameter
      - subtle cases may require a comparison of the 2 eyes
      - comparison of horizontal disc diameter to disc–macula distance
    - pale, gray, or (less commonly) hyperemic
    - may be surrounded by a yellow peripapillary halo
      - bordered by a ring of increased or decreased pigmentation
        - double-ring sign

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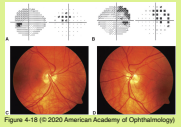
    - retinal vessel diameter may seem large
      - vessels may appear tortuous
  - Unilateral or bilateral optic nerve hypoplasia may be associated with
    - midline or hemispheric brain defects
      - thin or absent corpus callosum
    - endocrinologic abnormalities
      - deficiency of growth hormone and other pituitary hormones
        - growth retardation
        - hypoglycemic seizures
    - congenital suprasellar tumors
    - skull-base defects
      - basal encephaloceles
  - septo-optic dysplasia
    - optic nerve hypoplasia
    - absent septum pellucidum
    - pituitary dwarfism
  - superior segment hypoplasia
    - children of diabetic mothers
    - inferior visual field defect
    - bilateral, symmetric
- diagnostic workup
  - MRI is recommended in all cases
  - endocrinologic consultation

Congenital tilted disc syndrome
- 80% bilateral
- clinical presentation
  - superotemporal portion of the disc remains relatively intact
    - can seem elevated, simulating mild edema
  - inferonasal colobomatous excavation of optic nerve
  - superotemporal visual field defects
    - fail to respect the vertical midline
      - in contrast to visual field loss of chiasmal compression
    - partial improvement with myopic refractive correction
  - associated with thinning of adjacent RPE and choroid
  - retinal vessels are often nasalized



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- differential diagnosis
  - myopic tilted optic discs with temporal crescent

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