5.4.1. Patient With Decreased Vision: Classification & Management (I): Retinopathy (I)

Vitamin A Deficiency

- predisposing conditions
  - malnutrition
  - malabsorption
  - liver disease
  - zinc deficiency
- cofactor in the conversion of retinol to 11-cis-retinal
- clinical presentation
  - nyctalopia
  - peripheral visual field loss
  - ± central vision loss
  - xerosis
  - conjunctival Bitôt spots
  - keratomalacia
- dramatic recovery with treatment
  - vitamin A supplementation
  - treatment of any underlying systemic disorder

Ocular Media Abnormality

- irregularities/opacities of the ocular media
  - reduce visual acuity
  - ± cause generalized reduction of sensitivity on automated perimetry testing
  - do not affect pupils, color vision, or the appearance of the posterior pole

Retinopathy

- clinical presentation
  - decreased visual acuity
  - central visual field loss
    - focal and central
    - optic nerve disease: visual field defects are larger, often ceccocentral, and part of a generalized field depression
  - variable color vision loss
    - color vision loss = visual acuity loss
    - optic nerve disease: color vision loss > visual acuity loss
  - metamorphopsia
    - almost never in optic neuropathy
  - absent relative afferent pupillary defect (RAPD)
- diagnostic workup
  - OCT
  - fluorescein angiography
  - multifocal electroretinography (mfERG)
- maculopathies/retinopathies mistaken for optic nerve disease
  - acute idiopathic blind-spot enlargement
    - overlaps with multiple evanescent white dot syndrome
  - cone dystrophy
  - cancer-associated retinopathy (CAR) and melanoma-associated retinopathy (MAR)
  - central serous retinopathy
  - cystoid macular edema
  - acute zonal occult outer retinopathy (AZOOR)
- See Table 4-1

Paraneoplastic Syndromes

Cancer-associated retinopathy (CAR)

- symptoms
  - photopsia
  - nyctalopia
    - impaired dark adaptation
  - dimming
    - ring scotoma
    - peripheral and/or central visual field loss
  - often (50%) before the underlying malignancy is identified
  - progressively worsen over weeks to months
- etiology
  - small cell lung carcinoma
  - other lung tumors
  - breast cancer
  - uterine cancer
  - cervical cancer
- fundoscopy
  - initially normal
  - attenuated retinal arterioles
  - late
    - thinned and mottled RPE
    - optic disc atrophy
- ERG
  - markedly reduced amplitudes
    - both a- and b-waves
    - both rods and cones affected
- pathogenesis
  - tumor expresses an antigen homologous to 23-kDa retinal photoreceptor protein
    - calcium-binding protein
    - recoverin
  - circulating autoantibodies against the tumor-associated antigen
    - immune-mediated degeneration of rods and cones
    - cross-react with retinal recoverin
- treatment
  - treatment of inciting tumor has an unclear effect on retinal function
  - corticosteroids
  - plasmapheresis
  - intravenous immunoglobulin
- prognosis for vision is poor
  - vision loss is typically severe
- CaR = Cone + Rod ; ReCoverin
- maR = Rod

Melanoma-associated retinopathy (MAR)

- extremely rare
- etiology
  - previously diagnosed melanoma
  - investigation often reveals recurrence or metastasis
- symptoms
  - involves rod bipolar cells
  - photopsia
  - nyctalopia
  - prominent peripheral visual field loss
  - visual acuity, color vision, and the central visual field are often initially normal
  - develop rapidly over weeks to months
  - may have a sudden onset
- fundus
  - bilateral
  - may appear normal
  - RPE irregularity
  - may show
    - retinal arteriolar attenuation
    - optic disc pallor
- diagnostic workup
  - full-field ERG
    - negative ERG waveform
    - rod dysfunction
  - mfERG
    - relatively preserved
- prognosis
  - unlike CAR!
  - visual function may remain stable and nonprogressive
- treatment
  - no treatment has proven effective
  - intravenous immunoglobulin
- CaR = Cone + Rod ; ReCoverin
- maR = Rod
- CAR with auto-antibodies against alpha-enolase affects cones
- CAR affects rods and cones
- MAR affects rod function !
- MAR
- similar to Duchenne muscular dystrophy
- similar to CSNB
- mfERG measures photopic (cone) responses

Acute Idiopathic Blind-Spot Enlargement (AIBSE)

- differential diagnosis of enlarged blind spot
  - optic disc edema
  - optic disc tilting
  - AIBSE
- clinical presentation
  - AIBSE
    - symptoms
      - monocular scotoma
        - temporal location
      - photopsias
        - reflects disease of the outer retina
    - funduscopy
      - ± normal
      - may show
        - disc edema
        - peripapillary retinal lesions
        - choroiditis
        - retinal pigment epithelium (RPE) changes
        - uveitis
  - multiple evanescent white dot syndrome (MEWDS)
    - photopsia
    - retinal white spots
      - small
      - deep
      - posterior retina
      - resolve spontaneously
        - last weeks
  - ±RAPD
- diagnostic workup
  - perimetry
    - enlargement of the blind spot
  - mfERG
    - peripapillary depression
  - spectral domain OCT
    - peripapillary attenuation of outer retina
  - fluorescein angiography
    - early wreathlike hyperfluorescence and late staining
  - ICG angiography
    - peripapillary hypofluorescent lesions
      - correspond to the enlarged blind spot
  - full-field ERG
    - depressed a-waves
    - substantial intereye asymmetry
- prognosis
  - good visual prognosis

Figure 4-1 (© 2020 American Academy of Ophthalmology)

Cone Dystrophy

- symptoms
  - any age
  - bilateral
    - gradually progressive decline in visual acuity and color vision
  - photophobia
  - hemeralopia ("day blindness")
- clinical presentation
  - early stages
  - late stages
    - funduscopy
      - macular RPE becomes atrophic in a central oval region
      - "bull's-eye" pattern
    - similar to that observed in late chloroquine-induced maculopathy
- diagnostic workup
  - fluorescein angiography
    - highlight these abnormalities before they become clinically apparent
    - fundus autofluorescence
      - may be normal initially
  - full-field ERG
    - eventually shows markedly depressed photopic (cone) response and less prominently affected scotopic (rod) response
  - multifocal ERG
    - central depression
  - OCT
    - thinning of outer macular layers

Hydroxychloroquine and Chloroquine Retinopathy

- risk factors
  - duration of use > 5 years
  - cumulative dose
    - 1000 g (hydroxychloroquine)
    - 460 g (chloroquine)
  - daily dose
    - 400 mg (hydroxychloroquine) or
      - <6.5 mg/kg/day
      - <5 mg/kg/day is safer
    - 250 mg (chloroquine)
      - <3 mg/kg/day
  - older age (>60 years)
  - kidney or liver dysfunction
  - preexisting macular disease
- revised screening recommendations (2011)
  - baseline evaluation within 1 year of beginning treatment
  - annual examinations after 5 years of treatment
    - dilated fundus examination
    - automated, white 10-2 threshold perimetry
    - 1 or more of the following 3 objective tests
      - spectral domain OCT (to detect parafoveal outer retinal attenuation)
      - multifocal ERG (to detect parafoveal depressed amplitudes)
      - fundus autofluorescence (to detect parafoveal increased autofluorescence)
      - detect toxicity before fundus abnormalities become visible
  - fundus photography, Amsler grid testing, and color vision testing no longer recommended

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