OPTIC NERVE GRAND ROUNDS
"YOU'VE GOT SOME NERVE!"

Joseph W. Sowka, OD, FAAO, Diplomate
Professor of Optometry
Nova Southeastern University, College of Optometry
3200 South University Drive ◆ Fort Lauderdale, Florida 33328
Phone: (954) 262-1472
Email: jsowka@nova.edu

Tilted Disc Syndrome

- Appears rotated about axis- long axis may be horizontal rather than vertical
- No actual rotation occurs. Inferior tissue is missing while superior tissue is crowded together. Overall appearance is of rotation.
- Abnormal morphology of chorioscleral canal gives this appearance
- Optic nerve and fundus typical coloboma due to incomplete closure of fetal fissure at 6 weeks gestation.
- Findings include:
  - Inferior conus –inferior and inferior nasal
    - Can extend to involve the optic nerve as well and mimic glaucomatous notching
    - Corresponding field defect
  - Ectasia
  - Staphyloma
  - Hypopigmentation of inferior fundus (retina, choroid, and RPE)
    - Colobomatous disruption of RPE can lead to choroidal neovascularization
  - Situs inversus
  - Myopic astigmatism of oblique axis
    - Due to inferior staphyloma
- Unchanging with possible stationary field defect- temporal defect
- Field defect is superior temporal corresponding to inferior nasal conus

Buried Drusen of the Optic Disc

- Pseudo disc edema and disc elevation
  - Often confused with papilledema
- Retained hyaline bodies in anterior optic nerve head
- 1% incidence; 70% bilaterality
- Primarily in Caucasians
- Autosomal dominant with incomplete penetrance
- Asymptomatic
  - Mild acuity decrease and field loss may be present
- Small or non-existent cup
- Anomalous branching pattern arising from central vessel core

- SVP present
- May have associated sub-retinal hemorrhage
- Field defect: nasal step, arcuate scotomas
- Hyaline bodies in anterior optic nerve head can compress fibers and vascular supply
  - Leads to optic atrophy and visual loss in rare cases
- High reflectivity on b scan ultrasonography (except in children)
  - B scan is definitive diagnosis in most cases

Optic Nerve Head Hypoplasia
- Congenitally small nerves with pronounced scleral crescent (double ring sign)
- Dysplasia of retinal ganglion cells with loss of NFL
- ONH underdevelopment and sclera "fills in"
- Associated brain disorders and gestational diseases
- Maldeveloped growth
- Associated disorders include:
  - Gestational diabetes
  - Maternal infection (CMV, syphilis, rubella)
  - Fetal alcohol syndrome
  - Maternal drug abuse
- Septo-optic dysplasia
  - Short stature
  - Congenital nystagmus
  - Hypoplastic disc
- Unchanging, but may have drastically reduced acuity and field
- Normal acuity to NLP
- Strabismus, typically esotropia, frequently present (50%)
  - Must distinguish from amblyopia
  - Amblyopia, refractive error, binocular anomalies are secondary to hypoplasia
- Upon discovery, patient (if child) should be referred for full neurologic and endocrinologic evaluation, esp. if associated neurological signs are present or child is abnormally small for age

Optic Pits
- A type of optic nerve coloboma (atypical) resulting from incomplete closure of the fetal fissure
- Confused with notching of neuroretinal rim in glaucoma
- Will have field/vision loss corresponding to axon absence
- Serous detachment of the posterior pole
- Atypical because not always found in inferior disc

Morning Glory Syndrome
- Specific type of coloboma
  - Associated retinal vascular anomalies, glial proliferation, and perivascular pigmentation

- Funnel-like ectasia of disc and posterior fundus
  - Optic disc is displaced posteriorly into a funnel shaped staphyloma
- Vision highly variable
  - Typically less than 20/100 (to hand motion) in complete form
  - Vision may be good in forme fruste
  - Complete form is typically unilateral (though with other fellow eye colobomas) and forme fruste is typically bilateral
- Typically unilateral, but fellow eye often has other congenital defects
- Associated with non-rhegmatogenous retinal detachment of the posterior pole

Oblique Insertion
- Hyperopia is typically present
- Typically bilateral
- Unchanging with no field defect
- Nasal aspect heaped up while temporal aspect depressed or buried
  - Frequently misdiagnosed as glaucoma and disc edema
- Especially prominent in Asian patients

Physiologically Large Nerves and Cups: Megalopapillae
- Occur more commonly in patients with normally large chorioscleral canals and large discs
- Patients of color
- Common misdiagnosis of NTG and POAG

True Disc Edema
- **Papilledema**
  - Defines disc edema secondary to intracranial hypertension
    - Increased intracranial pressure must be present in order to diagnose papilledema
  - Bilateral
  - May present with transient visual obscuration, intermittent diplopia, headache, nausea, vomiting, tinnitus
  - Optic nerve is hyperemic; juxtapapillary retina is edematous; vessels are engorged, distended, and tortuous; peripapillary hemorrhages are common
  - Visual fields show enlarged blind spot (early) and arcuate defects with constriction (late)
  - Associated with intracranial abnormalities:
    - Increased brain volume (intracranial mass lesion)
    - Increased intracranial blood volume
    - Increased cerebrospinal fluid volume
  - Management involves stat neuro-imaging, lumbar puncture, and neuro consult

Anterior Ischemic Optic Neuropathy (AION)
- Results from local infarction at the level of the optic nerve

- Unilateral presentation but high incidence of subsequent contralateral involvement
- May be arteritic (AAION) or non-arteritic (NAAION)
    - AAION results from giant cell arteritis (GCA) and constitutes a medical emergency
    - NAAION secondary to other systemic disorders, most notably arteriolosclerosis, hypertension, and diabetes
- Presents with sudden devastating vision loss; associated scalp tenderness, weight loss, headache and jaw claudication when associated with GCA
- Optic nerve is pale (more so with AAION) with extensive nerve fiber layer edema; arteriolar constriction, peripapillary hemorrhages evident
- Visual fields variable. Patient is often blind
- Management involves stat erythrocyte sedimentation rate (ESR) and temporal artery biopsy if GCA suspected

Optic Atrophy

- Primary optic atrophy
    - Uniform nerve fiber degeneration, resulting in glial replacement but no architectural alteration of the optic nerve head.
    - Disc appears chalky white but the margins remain distinct and retinal vessels appear normal.
        - Trauma and compression (e.g. tumor) causes
- Secondary optic atrophy
    - Results from pathological chronic disc edema
        - malignant hypertension, papilledema, or infiltrative diseases like leukemia or sarcoidosis.
- Consecutive optic atrophy
    - Degenerative retinal conditions
        - Retinitis pigmentosa, pathological myopia and central retinal artery occlusion.
        - Pale, waxy disc, normal margins and marked attenuation of the arterioles.
- Temporal disc pallor
    - Toxic/ nutritional or demyelinating optic neuropathy (optic neuritis)
- Numerous potential etiologies
    - Infarction, infection, infiltration, inflammation, trauma, toxicity, metabolic dysfunction or direct compression of the nerve or chiasm
- Evaluation:
    - MRI studies should be obtained of the orbits, the optic chiasm and the brain with and without contrast, fat suppression for orbits, in a high field scanning unit.
        - Contrast dye (gadolinium) is beneficial in discerning malignant lesions, demyelinating plaques indicative of multiple sclerosis.
- Systemic causes of optic atrophy
    - sarcoidosis, tuberculosis, Behçet's disease, lymphoma, leukemia, systemic lupus erythematosus, nutritional or metabolic disorder (e.g. pernicious anemia,

folate deficiency), syphilis, Lyme disease, and antiphospholipid antibody syndrome.

- Complete blood count (CBC) with white cell differential, erythrocyte sedimentation rate (ESR), angiotensin-converting enzyme (ACE), antinuclear antibody (ANA), serum cardiolipin, serum homocysteine, serum B12 and folate levels, and rapid plasma regain (RPR) for syphilis. Additionally, chest x-rays could prove helpful in suspected cases of TB or sarcoidosis.

Compressive Optic Neuropathy

- Results from compression of the optic nerve at the orbital apex, secondary to:
  - Space occupying orbital lesions, including tumor masses
  - Infiltrated extraocular muscles (Graves' ophthalmopathy) in thyroid disease (most common)
- Unilateral with orbital masses, bilateral in Graves' disease
- Presents with slowly progressive, variable vision loss; variable proptosis and motility restriction
- Optic nerve is typically hyperemic with retinal edema, tortuous vessels, and associated hemorrhages; with prolonged compression, may see pallor and optic disc collateral vessels
- Visual fields consistent with papilledema in early stages, ischemic optic neuropathy in later stages
- Management involves orbital imaging and serum thyroid profile if Graves' suspected

Infectious/infiltrative optic neuropathy

- **Infectious**
  - Syphilis
    - Retrobulbar, papillopathy, neuroretinitis, perineuritis
    - Retrobular, bulbar: severe vision reduction
    - Perineuritis has normal vision, normal CSF pressure, normal MRI
  - Lyme
    - Mimic syphilitic optic neuropathy
  - Toxoplasmosis, HIV/AIDS, CMV
    - Destructive to vision
  - Neuroretinitis
    - Good visual function
    - Typically benign lymphoreticulosis (cat scratch disease)
    - Gram-negative bacillus

- **Infiltrative**
  - Sarcoidosis
  - Systemic lupus erythematosus
  - Leukemia
  - Lymphoma
  - Carcinoma