

THE **SWOLLEN** OPTIC DISC: EMERGENCY OR ANOMALY?

Alan G. Kabat, OD, FAAO
Memphis, Tennessee

(901) 252-3691
alan.kabat@alankabat.com

Course description: The “swollen disc” presents a diagnostic dilemma. While some presentations are benign, true disc edema may constitute a medical emergency. This course examines the differential diagnosis, ancillary testing, and management strategies for patients with swollen discs.

LEARNING OBJECTIVES: At the conclusion of this lecture, the attendee will be able to:

1. Identify the most common pathological etiologies of the swollen optic disc, i.e. optic disc edema.
2. Identify the most common benign etiologies of the crowded optic disc, i.e. pseudopapilledema.
3. Describe the key characteristics of the optic nerve and retina, as well as ancillary diagnostic findings that help to distinguish these conditions.
4. Understand the basic pathophysiology behind optic disc edema and its various etiologies.
5. Recognize the key risk factors (age, sex, systemic conditions and medications, etc.) associated with the various etiologies of optic disc edema.
6. Recognize those conditions that require immediate intervention, and identify the specific therapy and/or medical professional to whom the patient must be referred.

CASE PRESENTATIONS: The following cases will be used to demonstrate and enhance understanding of the presentation, pathophysiology, diagnosis and management of a number of “swollen discs”:

- A 28-year-old black female with intermittent blurred vision & visual “blackouts”, intermittent diplopia, and chronic headache steadily worsening over the past two weeks.
- A 25-year-old white male pharmacy student complaining of headaches that awaken him with nausea and vomiting, persisting for several months and getting worse.
- A 34-year-old white female with sudden onset of blurred vision and dull pain in the left eye for the past two days.
- A 24-year-old black female presenting for comprehensive examination with no visual or ocular complaints; her current medications consist of oral contraceptives only.
- A 54-year-old Hispanic female with intense pain in her right eye and associated vision loss; the pain is exacerbated by eye movements and has been worsening over several days.
- A 42-year-old white female with unilateral painless reduction of vision for approximately one week.
- A 72-year-old white female with sudden vision loss in her left eye, a headache for the past 6 weeks, and amaurosis fugax OS three times during the past 4 weeks.

Please follow along with the presentation and “test your knowledge” by participating in the discussion. If you would like more detailed notes, please email me.

Differential Diagnosis: Conditions Presenting with “Swollen Discs”

- ***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 arteriosclerosis, 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
- ***Buried Optic Disc Drusen***
 - Results from associated hyaline deposition within the structure of the nerve
 - Bilateral but variably asymmetric presentation
 - Typically asymptomatic; may have mildly reduced vision and/or afferent pupillary defect when asymmetric
 - Optic nerve is elevated but not hyperemic; borders may be “scalloped” or “bumpy”; vessels demonstrate unusual branching patterns; typically no retinal edema unless associated with subretinal neovascular membranes (SRNVM); refractile drusen may be evident within nerve head and/or juxtapapillary tissue
 - Visual fields highly variable, consistent with focal nerve compression
 - Management involves proper differential using ultrasonography and/or CT; rule out SRNVM with fluorescein angiography if suspected; photodocumentation and monitoring constitute long-term plan
- ***Congenitally “Full” or Hypermetropic Disc***
 - Results from congenitally small eye and posterior scleral foramen; often associated with hyperopic refractive error
 - Bilateral
 - Patients are asymptomatic without specific acuity or field deficits
 - Optic nerve is elevated but not hyperemic; vessels demonstrate unusual branching patterns; no disc edema or hemorrhage
 - Management involves proper differential using ultrasonography to measure axial length; photodocumentation and monitoring constitute long-term plan

- **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 optociliary shunt 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
- **Demyelinating Optic Neuropathy**
 - Referred to clinically as "optic neuritis" or "papillitis"
 - Demyelinating optic neuropathy is a unique subset of this category, since it is not truly an inflammatory condition
 - Unilateral
 - Presents with sudden onset vision loss, pain on palpation and eye movements
 - Optic nerve is hyperemic; juxtapapillary retina is mildly edematous and may show exudate; vessels are engorged and distended; posterior vitritis likely
 - Visual fields may demonstrate arcuate, altitudinal, or cecocentral scotomas
 - Numerous systemic etiologies (e.g., multiple sclerosis)
 - Management involves targeted systemic workup (hematology/serology and radiology) with MRI (crucial) to r/o multiple sclerosis
- **Embryonic vestiges**
 - Represent arrest in normal embryonic development, resulting in redundant tissue
 - Typically unilateral, but no definitive pattern
 - Patients are asymptomatic without specific acuity or field deficits
 - **Bergmeister's papilla**
 - **Prepapillary glial veils**
 - **Medullated or Myelinated Nerve Fibers**

- **Infectious/infiltrative optic neuropathy**
 - **Infectious**
 - Syphilis
 - Retrobulbar, papillopathy, neuroretinitis, perineuritis
 - Retrobulbar, 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
- **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

- ***Papillophlebitis***
 - Big blind spot syndrome, presumed phlebitis of the ONH, benign retinal vasculitis, etc.
 - Presumably inflammatory in nature
 - CRVO in young adult
 - May present with only unilateral disc swelling with minimal visual dysfunction (blind spot enlargement only)

- ***Tilted disc syndrome***
 - Disc coloboma
 - Inferior/ inferior nasal conus
 - Abnormal chorioscleral canal morphology
 - Heaped up superior aspect of nerve
 - Normal acuity
 - Unchanging superior temporal field defect not respecting vertical hemianopic line