



## Jeopardy! Optic Nerve

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## Anomalous Optic Nerves

### CONGENITAL

- Minimal cupping
- Usually no hemorrhages
- Vessels emerge centrally from disc
- Disc may be small or large, with relative normal vessel caliber

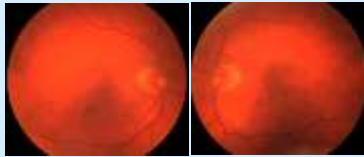
### ACQUIRED

- Hyperemia
- Disc / retinal hemorrhages
- Abnormal blood vessels
- Cotton wool spots
- Loss of spontaneous venous pulsation

### Jeopardy! Optic Nerve

## Congenital Optic Nerve Anomalies

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- ❖ Usually no hemorrhages
- ❖ Vessels emerge centrally from disc
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## Astrocytoma

- Glial tumors of the retinal nerve fiber layer that arise from retinal astrocytes
- May be found as an isolated finding or in patients who have tuberous sclerosis (TS), or rarely, in those with neurofibromatosis
- Clinical features:
  - Can be seen as optic nerve or retinal glial tumors
  - Initially appear as grayish, flat and translucent lesions
  - May enlarge and become chalky white, or glistening, yellow, mulberry-like depending on the amount of calcification
- Management:
  - Monitor optic nerve/retinal lesions for complications
  - Referral for systemic work up, possible genetic counseling if TS diagnosed

## Buried Disc Drusen

- Results from the accumulation of axoplasmic debris due to abnormal axonal transport and metabolism
- Drusen advance anteriorly with age
- Optic nerve drusen may be associated with:
  - Visual field defects, in particular, altitudinal defects
  - Disc hemorrhages
  - Disruption of papillary tissue resulting in choroid neovascular membrane formation
- Ophthalmic testing includes:
  - B-scan ultrasonography (most sensitive)
  - Optical coherence tomography
  - CT imaging
  - Auto-fluorescence
  - Fluorescein Angiography
- Management:
  - Typically, observation

## Congenitally Full Disc

- Descriptive term used to describe atypical presentations of crowded and elevated optic nerve heads which are otherwise normally functioning
- Appear as small to normal-sized optic nerve heads that appear elevated, swollen and crowded, yet have relatively distinct margins and no evidence of nerve fiber layer edema
  - Retinal vessels may appear unusually tortuous and demonstrate anomalous branching patterns
  - Presence of reverse loops in vessels
- Usually no physiologic cup
  - Neural rim tissue pink; although rim may appear slightly hyperemic
- High association of hyperopia
- VA, color vision, visual fields normal

## Morning Glory Syndrome

- Rare, unilateral, associated with very poor vision
- Right eye more common; more common in females
- No known hereditary pattern
- Due to the defective embryologic development of the sclera and lamina cribrosa:
  - Results in an enlarged scleral foramen and posterior displacement of the lamina cribrosa
  - The optic nerve is larger than normal with an unusual vascular pattern radiating from the disc
- 3 cardinal signs:
  - The cup is enlarged, with a deep conical depression and white tissue at the base
  - A raised annulus surrounds the disc with variable amount of pigment
  - The retinal blood vessels emerge from the disc in a radial fashion, like the spokes of a wheel

## Optic Disc Melanocytoma

- Benign, highly pigmented tumor arising from melanocytes
  - A variant of melanocytic nevus
  - Malignant transformation has been reported and confirmed histopathologically
  - Occurs on the optic disc and often extends into the peripapillary retina and choroid
- Both sexes are equally affected; increased incidence in blacks
- Typically asymptomatic and diagnosed during routine fundus examination.
- Most lesions do not affect visual acuity
  - 25% have mildly reduced vision, due to subretinal fluid or exudation
  - Severe vision loss rare: CRVO, tumor necrosis, malignant transformation
- Most have visual field defects
  - Enlarged blind spot (75%); typically asymptomatic
  - Can also have nerve fiber layer defects due to optic disc compression
- Should have regular eye exams, can be monitored yearly if no effect on vision or visual field

## Optic Nerve Hypoplasia

- A result of failure of the development of the ganglion cell layer in the neural retina
  - Results in a reduction in the number of nerve fibers in the optic nerve
  - The posterior scleral foramen "fills in" with connective and scleral tissue
- The optic nerve is reduced in size, the nerve fibers are concentrated centrally giving the disc a contracted appearance encircled by an outer ring of aberrant tissue ("double ring sign")
- Vision may range from normal to subnormal levels
- Sporadic, but may be associated with iatrogenic conditions (gestational diabetes, maternal CMV infection, syphilis, rubella, maternal drug use)
  - The association with fetal alcohol syndrome is very significant

## Optic Disc Pit

- Thought to represent an abnormality arising early in the development of the presumptive optic disc (epithelial papilla)
- Grayish round depression within the disc margin tissue (typically infra-temporal)
- Variable size of depression; typically ranging from  $\frac{1}{4}$  to  $\frac{1}{2}$  disc diameter
  - Disc with pit will be larger than contralateral disc
- Approximately one-half to two-thirds of optic disc pits are associated with maculopathy; classically, serous retinal detachment

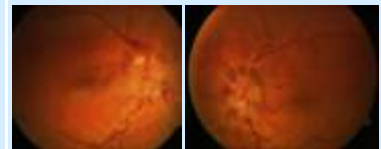
## Tilted Disc Syndrome

- Results from incomplete closure of the fetal / embryonic fissure
- Degree of tilting varies along spectrum up to  $90^\circ$
- Anatomical associations include:
  - Fuchs coloboma
  - Fundus ectasia (commonly infero-temporal)
  - Situs inversus
- Tilted disc may manifest with:
  - Temporal (commonly superior-temporal) visual field defect
  - Myopic astigmatism

### Jeopardy! *Optic Nerve*

- Hyperemia
- Disc / retinal hemorrhages
- Abnormal blood vessels
- Cotton wool spots
- Loss of spontaneous venous pulsation

### Acquired *Optic Nerve Anomalies*



## Anterior Ischemic Optic Neuropathy

- Two forms of anterior ischemic optic neuropathies (AION):
  - Non-arteritic → N-AION
  - Arteritic → A-AION
- The commonality between both forms of optic neuropathy is compromise to the vasculature supply to the anterior aspect of the optic nerve

## Anterior Ischemic Optic Neuropathy

### NAION

- Pathogenesis:
  - Ischemia to posterior ciliary arteries results in lost of ONH perfusion
- Symptoms:
  - Sudden painless vision / visual field
  - Prodrome symptoms
- Findings:
  - Afferent pupillary defect
  - Optic nerve swelling, with possible disc hemorrhages, progressing to pallor
  - Optic nerve pallor
  - Disc at risk in contra-lateral eye
  - Color vision & perimetric defects
- Treatment and management:
  - Systemic cardio-vascular workup
  - Treat glaucoma / ocular hypertension

### A-AION

- Pathogenesis:
  - Inflammation of the elastic tissue in the media and adventitia of the arterial walls resulting in vessel occlusion
- Association with giant cell arteritis
- Symptoms:
  - Sudden vision loss
- Associated symptoms include prodrome, headaches, jaw claudication, and scalp tenderness
- Findings:
  - Afferent pupillary defect
  - Optic nerve hemorrhages, pallor & atrophy
- Treatment and management:
  - Laboratory testing
  - Temporal artery biopsy
  - Rheumatology & neuro-ophthalmology evaluation

## Bilateral Disc Edema

- Strictly defined as (bilateral) optic disc elevation due to increased intra-cranial pressure (ICP)
  - Disc elevation primarily due to axonal swelling and degeneration
- Space occupying lesion must always be ruled out
  - MRI with and without contrast absolutely indicated
- Other possible etiologies:
  - Infection
  - Anatomic abnormality
  - Malignant HTN
- Pseudotumor cerebri is a diagnosis of exclusion

## Pseudotumor Cerebri

- AKA idiopathic intracranial hypertension, benign intracranial hypertension
- Diagnosis of EXCLUSION:
  - Signs of increased ICP, typically manifested by papilledema
  - Absence of neurologic signs, 6th nerve palsy is the exception
  - No imaging evidence of ventriculomegaly or intracranial mass lesion
  - ICP > 200 mm H<sub>2</sub>O and normal cytology
- 0.9 per 100,000 of the general population
  - Incidence rises to 14 in 100,000 in females between 20-40 over ideal weight
- Obesity = most commonly associated condition, found in 50% of pts with PTC
- Female to male ratio = 8:1
- PTC in kids: males = females, and obesity less common

## Diabetic Papillopathy

- Patients present with asymptomatic swollen, hyperemic optic disc, peripapillary capillary dilatation, blurred disc margins, retinal hemorrhages
- "Typically" found in young type 1 insulin dependent diabetics, but also occurs in type 2 diabetes
- More often bilateral, but can also be unilateral
- Diagnosis of exclusion
  - Must rule out other causes of disc edema
- Strong correlation with CSDME – need to monitor carefully

## Hypertensive Disc Edema

- Associated with malignant hypertension
  - Systolic: BP > 220 mm Hg
  - Diastolic: BP > 120 mm Hg
  - Malignant hypertension represents medical emergency
- Findings
  - Unilateral or bilateral disc swelling
  - Blurring of disc margins
  - Hemorrhages
  - Cotton wool spots
- Treatment and management
  - Systemic management
  - Observation with dilated fundus examination

## Normal Tension Glaucoma

- Glaucomatous optic neuropathy with IOP less than "21 mm Hg" and associated with typical glaucomatous visual field defects
- Diagnosis of exclusion, made after ruling out other mimickers
  - Need to rule out differentials:
    - Non-low tension glaucoma (secondary)
    - Other optic nerve conditions
      - Compressive
      - Neuropathy
      - Congenital / hereditary
    - Other systemic or ocular conditions

## Optic Atrophy

- Physical sign, not a diagnosis, used when there is excessive pallor of the disc
  - Manifests as changes in the color and the structure of the optic disc associated with variable degrees of visual dysfunction
  - Usually, associated visual field defect, reduced acuity and optic nerve pallor are observed
- Can occur following numerous conditions
  - Optic nerve disease, Inflammatory, Compressive, Vascular, Deficiencies, Toxic, Traumatic, Congenital, Metabolic, Associated with retinal lesions, Associated with CNS disease

## Optic Disc Collaterals

- Pre-existing blood vessels that become patent as a result of an upstream obstruction or impedance to blood flow
  - Do not represent new vessel formation
- Occur in approximately 3% of patients with glaucoma or ocular hypertension

## Optic Neuritis

- Primary demyelination of the optic nerve which results in acute reduction in vision
  - The myelin sheath is under direct attack with relative sparing of axons
- Epidemiology:
  - 2:1 female:male ratio
  - White > black
  - 20-50 years of age (mean age = 3rd decade)
- Clinical appearance can be typical (isolated or associated with MS) versus atypical
- Classic triad of symptoms:
  - Reduced visual acuity
  - Reduced color vision
  - Pain on eye movement
- Atypical optic neuritis may occur with diseases other than MS, e.g. connective tissue disease, sarcoid, syphilis, CMV, toxoplasmosis, HZV, meningitis, Lyme