

# Optic Nerve Anomalies

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No disclosures

## Goals for today

- Review some of the optic nerve anomalies that can be seen in practice
- Review signs and symptoms of varied conditions
- Understand management and treatment options
- Review clinical tips and cases

## Optic Disc Pits

### Optic Disc Pits: Background

- Spectrum of congenital cavitory anomalies of the optic disc, which also includes optic disc coloboma, morning glory syndrome
  - Due to incomplete closure of the superior edge of the embryonic fissure
- Optic disc pits are usually unilateral and sporadic in occurrence
- Rare and occur equally in men and women with an estimated incidence of 1 in 11,000 people

## Signs and Symptoms

- **Signs**
  - Single, oval-shaped depressions at the optic disc
  - Commonly found at the infero-temporal aspect
    - Can be found elsewhere, including centrally
  - Occasionally, an optic disc can have more than one pit
  - ODPs are usually grayish, but may also be yellow or black
  - Maculopathy
- **Symptoms:**
  - Asymptomatic
  - **Decreased VA**
  - VF: enlarged blind spot, paracentral scotoma



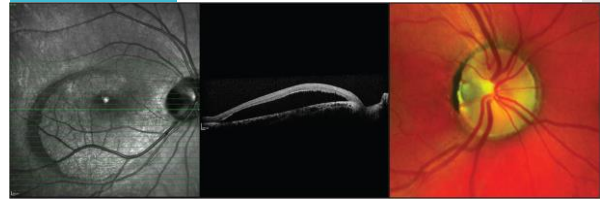
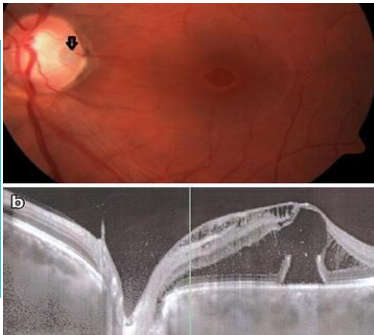
### Optic Disc Maculopathy

- Optic disc pit maculopathy: macular changes that occur which include intraretinal and subretinal fluid accumulation, and RPE changes
- Maculopathy occurs in 25–75% of patients with an ODP
- Fluid source: vitreous, CSF, blood vessels, choroid
  - ODPs are congenital, but the development of maculopathy has no known triggers

### Optic Disc Maculopathy

- Can occur at any age, from early childhood to the ninth decade of age
  - Usually occurs in the third and fourth decades of life
  - PVD process?
- Poor prognosis, with a natural history of gradual worsening and a final VA of 20/200 or worse

### Optic Nerve Pits



### Treatment

- No established guidelines for the treatment of ODP-M, and no consensus on the mechanism of pathogenesis or the optimal surgical technique
- Argon laser photocoagulation
  - laser scars at temporal disc margin will create a chorioretinal adhesion which will act as a barrier between the ODP and the subretinal space
- Vitrectomy with gas tamponade
  - Traction relief
  - Gas-barrier that blocks passage of fluid through the pit

### Idiopathic Intracranial Hypertension

(Pseudo tumor Cereberi)

## Idiopathic Intracranial Hypertension

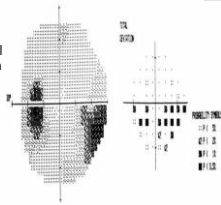
- Disease of increased intracranial pressure with no known cause
- Can lead to progressive visual loss (acuity and field)
- The annual incidence of IIH is 0.9/100,000,
  - 3.5/100,000 in females 15 to 44 years of age.
- More than 90% of IIH patients are obese and over 90% are women of childbearing age.

## Disease Background: Etiology/Pathogenesis

- Poorly understood
- Most popular: reduced CSF absorption due to dysfunction of arachnoid granulations
- Absorption dysfunction possibly through lymphatics
- Increased intra-abdominal pressure with cerebral venous hypertension
- **A good hypothesis should account for why it occurs in obese women in childbearing years**

## Mechanism of VA Loss

1. Disruption of axonal transport
  - Elevated CSF disturbs normal pressure gradient resulting in axoplasmic stasis
2. intraneuronal optic nerve ischemia
  - Hayreh: delays in arterial filling with FA
  - Visual field defects similar to other optic neuropathies



## Symptoms

- VA loss
- Headache
  - **usual presenting sign**
  - Severe, constant, pulsatile
- Transient Visual Obscurations
  - Usually less than 30 seconds
  - Possibly due to transient ischemia of the ONH
- Tinnitus
  - Thought to be due to transverse sinus collapse (venous stenosis) from high CSF pressure

## Signs:

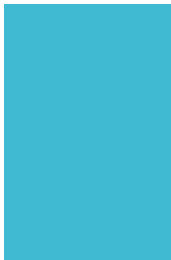
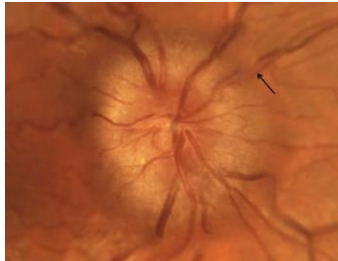
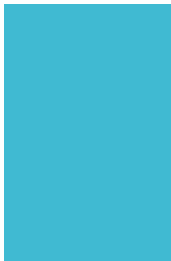
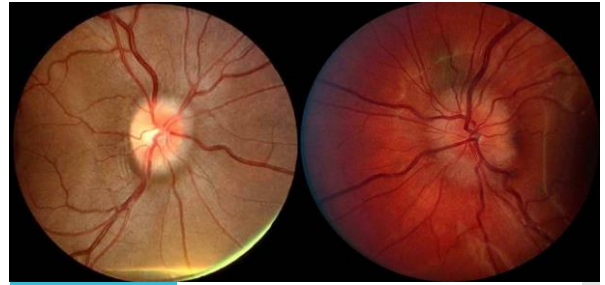
- Papilledema: cause of visual loss
  - Grading system from 0-5
- Ocular Motility Disturbance:
  - Sixth nerve palsy, horizontal diplopia
- Perimetry
  - Enlargement of blind spot
  - Visual field loss other than enlarged blind spot in 92% of patients
  - Inferior nasal step, peripheral nasal loss, arcuate
  - Good for following pt for improvement

## Diagnostic Testing

- Intracranial imaging – CT adequate for mass lesions
- MRI & MRV are preferred due to superior ability to diagnose venous sinus thrombosis.
- **Lumbar Puncture –**
  - Measure opening pressure and send CSF for analysis (routine, infection, cytology, etc.)

Modified Dandy Criteria

- signs and symptoms of increased intracranial pressure, such as papilledema and headache
- no localizing findings on neurological examination except for cranial nerve six palsy
- normal MRI/CT scan
- high intracranial pressure of 250mm/H<sub>2</sub>O or above on a spinal tap, with no abnormalities of cerebrospinal fluid
- is awake and alert
- has no other cause of increased intracranial pressure found



Treatment/Out comes

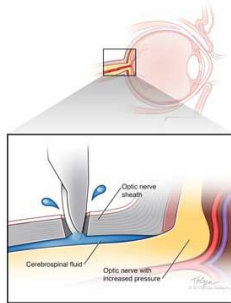
- Most important factor is amount and progression of visual loss
- Severity of patient symptoms
- **Eliminate presumed causal factors**
  - Oral contraceptives
  - Tetracyclines
  - Nalidixic acid (anti-biotic)
  - Vitamin A

### Treatment: Medical Therapy

- **Weight Loss**
  - Papilledema can resolve with modest weight loss(5-10% of total body mass)
- Lumbar Puncture
- Corticosteroids-side effects, rebound pressure increase
- **Diamox**
  - Reduces CSF production at the choroid plexus
- Topomax
  - CAI, weight loss common, comparable to diamox

### Surgical Therapy

- Optic Nerve Sheath Fenestration
  - Preferred treatment for patients w/progressive **VA loss** and mild headaches
  - CSF drains into orbital fat where it is absorbed into the venous circulation
- CSF Shunting Procedures
  - **Indicated** if failed medical therapy or intractable **headache**
  - Initial success but re-operations in almost 50% of patients



### Idiopathic Intracranial Hypertension Treatment Trial

- multicenter, double-blind, randomized, placebo-controlled study
  - All patients: lifestyle modification program of weight reduction with a low-sodium diet.
  - Then randomized to receive either acetazolamide or matching placebo
- Outcomes:
  - better visual outcomes than those taking placebo along with the diet.
  - significantly improved papilledema
  - quality of life measures
  - lower cerebrospinal fluid pressure.

### Optic Nerve Head Drusen

### Background

- consist of acellular, intracellular, and extracellular protein deposits(hyaline) that often become calcified over time
  - Due to disturbance in axonal metabolism with slowed axoplasmic flow
- typically buried early in life and generally become superficial, and therefore visible, later in childhood
  - average age of 12 years

## Background

- **1% of general population** affected
- Higher risk in **Caucasian** population
- Can be associated with other conditions (pseudoxanthoma elasticum, retinitis pigmentosa, angioid streaks)

## Signs and Symptoms

- Signs:
  - 75-85% bilateral
  - Yellowish depositions on and around optic nerve
  - Edges of optic disc or cup may be distorted
    - scalloped
  - Loss of optic cup and disc borders (may resemble papilledema)
  - Marked bifurcations and trifurcations
- Symptoms:
  - Usually asymptomatic
  - Up to 8.6% reported to have transient visual obscurations
  - Visual field defects
  - central visual acuity loss may occur

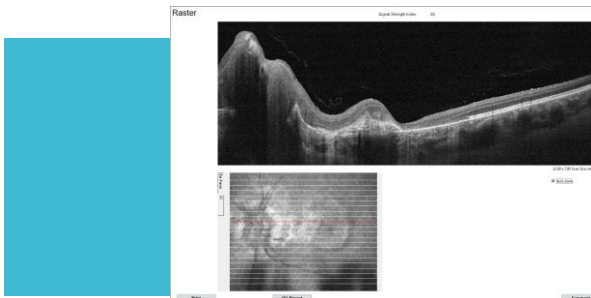
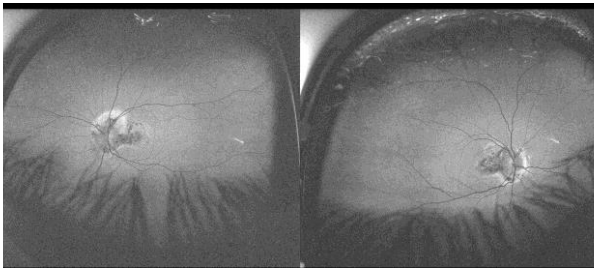
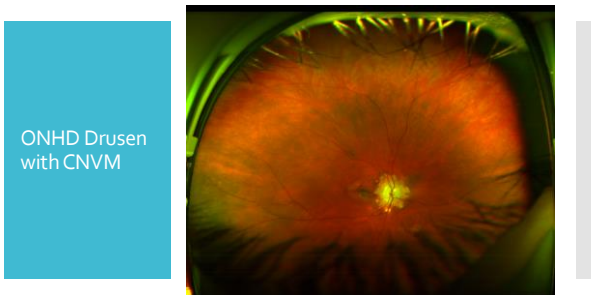
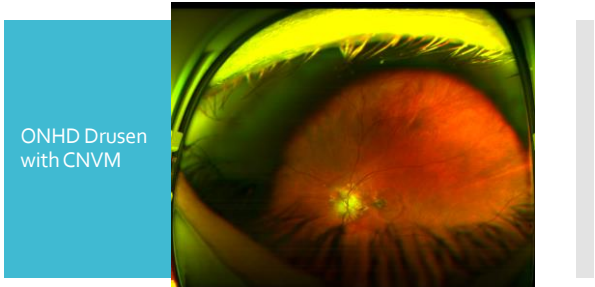
## Complications:

- visual field defects
- Hemorrhages
- choroidal neovascular membrane
- nonarteritic anterior ischemic optic neuropathy
- retinal vascular occlusions



## Clinical Pearls

- How can I confirm the ONH drusen?
  - Gold standard...
  - OCT
  - FAF
    - Autofluorescence of drusen depends on it's depth
    - deep buried drusen may be difficult to assess using FAF.
- It looks like IIH?
  - Symptoms...(headaches, VA loss, tinnitus)
  - Presence of a spontaneous venous pulse (SVP)
    - Rules out true papilledema?
    - Absence of SVP occurs in 20% of the normal population



**Treatment**

- No definitive therapy available
- Clinical pearl:**
- **Should I prescribe glaucoma drops for ONHD?**
- Depends on the IOP...
  - VFL occurs more frequently in eyes with ONHD that also have OHT
- Patients with elevated IOP and ONHD should remain under close surveillance for disease progression and be treated appropriately to prevent additional visual field loss\*
- In the absence of elevated IOP, there is no evidence that IOP reduction will have any effect on preventing visual morbidity\*
- **Problems with both thoughts of research...all retrospective!**
  - no controlled clinical studies that show a benefit to lowering IOP in any eyes with ONHD.

- Problems with long term iop treatment??
- Anti-vegf for CNVM
- Manage concurrent conditions

## Nutritional and Toxic Optic Neuropathies

### Nutritional and Toxic Optic Neuropathies

- Toxic and nutritional optic neuropathies often **both present in the same patient** and have similar clinical presentation
- History of exposure to foreign substance
  - Would need to ask appropriate history questions..
- Onset usually after months of exposure
  - Acute in some cases...
  - May be asymmetric presentation, but usually equal

### Pathogenesis

- Pathogenesis remains unknown in most cases
- For vitamin B<sub>12</sub> deficiency and at least some toxic agents, including methanol, ethambutol, and linezolid, the final common pathway probably involves **damage to mitochondrial oxidative phosphorylation**
  - Decreased ATP and generation of ROS
- Leads to damage to the papillomacular bundle

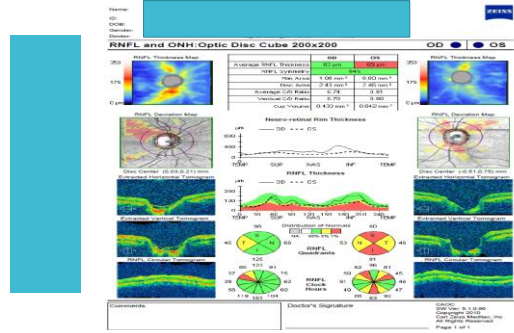
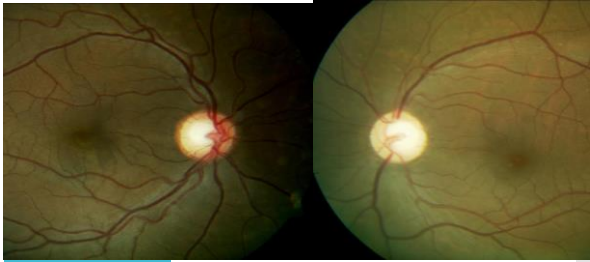
### Clinical Picture

- Signs:
  - Optic nerve pallor (temporal) (papillomacular)
  - Visual field defects (ceco-central)
  - Color vision defects
  - OCT: RNFL thinning, especially in the papillomacular bundle
- Symptoms:
  - Painless, bilateral, gradual vision loss

### Causes:

- Toxic:
  - methanol, ethylene glycol(antifreeze), **ethambutol**, **isoniazid**, digitalis, cimetidine, vincristine, cyclosporine, toluene(feedstock, paint thinner), and amiodarone
- Nutritional:
  - associated with malnutrition or poor dietary habits, incorrectly applied vegetarian diet, or chronic alcohol abuse.
    - folic acid
    - vitamin B complex deficiency: B<sub>1</sub>, B<sub>2</sub>, B<sub>12</sub>
  - Patients after bariatric surgery
    - copper





## Treatment

- Nutritional:
  - Blood testing
  - Administer appropriate nutrient
  - Prognosis depends on length of exposure and individual factors
- Toxic:
  - Immediate withdrawal from toxic agent
  - Prognosis dependent on toxicity of agent, length of exposure, individual factors

## Optic Nerve Head Hypoplasia

## Background

- Optic Nerve Hypoplasia is the under-development of the optic nerve combined with **possible brain and endocrine abnormalities**
- Possible systemic associations:
  - endocrine abnormalities
  - developmental delay
  - cerebral palsy
  - seizures
- There is a greater incidence of clinical neurologic abnormalities in patients with bilateral ONH (65%) than patients with unilateral ONH

## Pathophysiology

- ONH occurs due to **diminished number of axons** in the involved nerve with normal development of supporting tissues and the retinal vascular system
  - ~6-17 weeks, there are approximately 3 million optic nerve axons which ultimately are reduced to approximately one million at the time of birth
  - **Hypoplasia may therefore be an overdone, but normal, process of involution**
- The timing of coexistent CNS injuries suggest that some cases of optic nerve hypoplasia may result from intrauterine destruction of a normally developed structure whereas others represent a primary failure of axons to develop

Signs and Symptoms

- Signs:
  - Small optic disc with large vasculature
  - Visual field defects
  - May be associated with nystagmus
  - (OCT) shows a mild degree of foveal hypoplasia with associated thinning of the retinal ganglion cell and nerve fiber layer
- Symptoms:
  - 20/20 to NLP
    - Acuity variance??
    - Does size play a factor?
    - VA remains mainly unaffected and the finding of visual field defects later in life may result in a late diagnosis
  - localized visual field defects, often combined with a generalized constriction of the visual fields

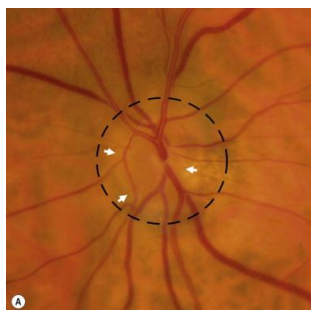


Figure 1. Optic Disc Size Measurement

Measurement of optic disc size:

Biomicroscopy:

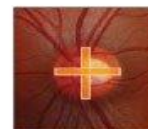
- Volk lens
- Measure length of slit beam

Correction factors:

Volk 60 D - x 1.0

Volk 78 D - x 1.1

Volk 90 D - x 1.3



Average vertical diameter: 1.8 mm

Average horizontal diameter: 1.7 mm

Vertical and horizontal disc diameter can be obtained during slit-lamp examination with a fundoscopic lens, applying correction factors according to the lens magnification.

Source: Felipe A. Medeiros, MD, PhD

### Clinical Pearls:

- In more mild cases, disc to macula distance/disc diameter ratio will be increased. A ratio of 2.94 is seen in the normal population and greater than three indicates milder forms of ONH Hypoplasia.
- The "double ring sign" can be seen in some patients and is characterized by a pigmented ring surrounding the disc.
- Retinal vascular tortuosity is also an important but inconsistent sign.
- **None of the above is considered pathognomonic**

### Treatment

- No treatment for optic nerve hypoplasia.
  - monocular precautions
  - low vision rehab
- Appropriate referrals:
  - Pituitary abnormalities: pediatric endocrinologist for hormonal supplementation is necessary.
  - Neuro
  - Occupational, physical and speech therapists