Optic Disc Variations, Anomalies, and Pathologies

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Disclosures

Dr. Yudcovitch does not hold proprietary financial interest in any of the products or companies mentioned in this presentation.

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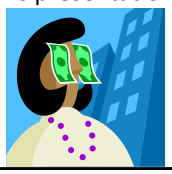
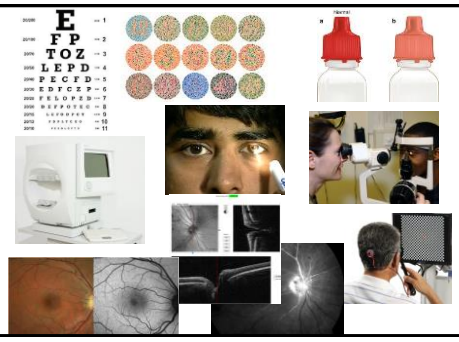
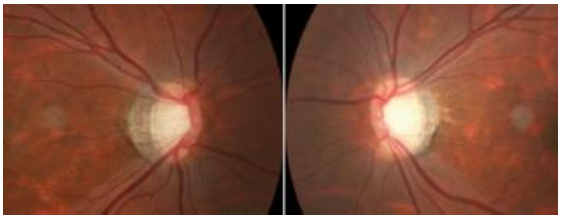




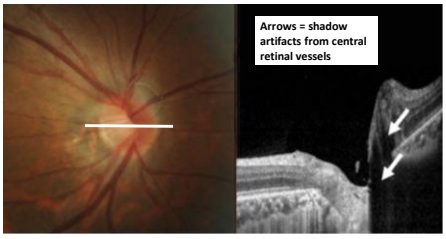
Malinserted Optic Nerve Head

- Oblique insertion of optic nerve into scleral canal
- Almost always bilateral
- Usually elevated nasal rim, tilting downwards temporally with temporal scleral and/or choroidal crescent
- No rotation of optic nerve, nasal staphyloma or situs inversus
- Common; more frequent in myopia
  - Especially pathological (>6D) myopia
  - Temporal, PPA (peripapillary atrophy)
- Generally benign

Malinserted Optic Nerve Head



Malinserted Optic Nerve Head

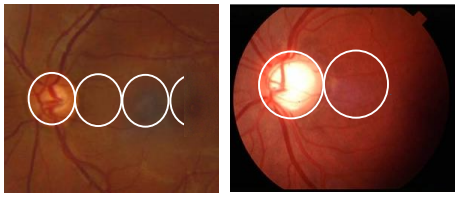


https://journal.opted.org/article/the-use-of-oct-in-differential-diagnosis-of-elevated-optic-discs/

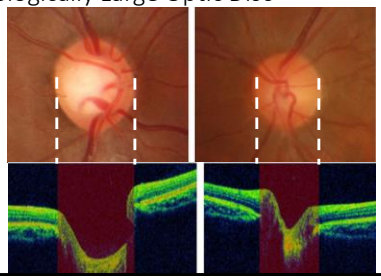
Physiologically Large Optic Disc

- Average size disc diameter range: 1.3-1.7mm
- Physiologically-large optic discs: >1.8mm diameter
- Primary macrodiscs: Not related to axial length
  - Asymptomatic: no morphologic or functional defects
  - Symptomatic: Morning Glory Syndrome, optic pits, etc.
- Secondary macrodiscs: high axial length (>26.5mm)
  - Due to primary high myopia
  - Due to secondary high myopia from congenital glaucoma

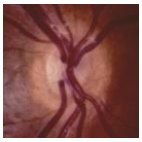
Physiologically Large Optic Disc



Physiologically Large Optic Disc

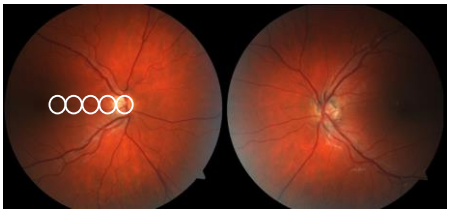


Hypoplastic Disc (Optic Nerve Hypoplasia)



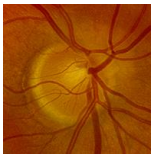
- Small pale grey optic nerves
- Decreased nerve fibers from birth
- Small nerve in normal scleral canal
- "Halo sign"("Double ring sign")
  - Yellow-white ring around the disc
  - Sclera to lamina junction and retina to lamina junction
- Visual acuity varies from normal to light perception
- Associated with absence of septum pellucidum
  - Septo-Optic Dysplasia
- Hypothyroidism, pituitary abnormalities common

Hypoplastic Disc (Optic Nerve Hypoplasia)



https://casereports.bmj.com/content/casereports/2017/bcr-2017-220343.full.pdf

Superior Segmental Optic Nerve Hypoplasia (Topless Disc Syndrome)

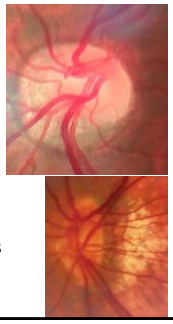


- Optic nerve hypoplasia sub-category
- Superior disc pallor
- Superior peripapillary scleral halo
- Inferior visual field defect
- Superior retinal nerve fiber layer thinning
- Superior entrance of central retinal artery
- Strong association with maternal diabetes

https://webeye.ophth.uiowa.edu/eyeforum/atlas/pages/SSONH/index.htm

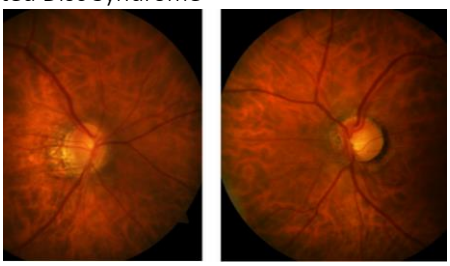
Superior Segmental Optic Nerve Hypoplasia (Topless Disc Syndrome)

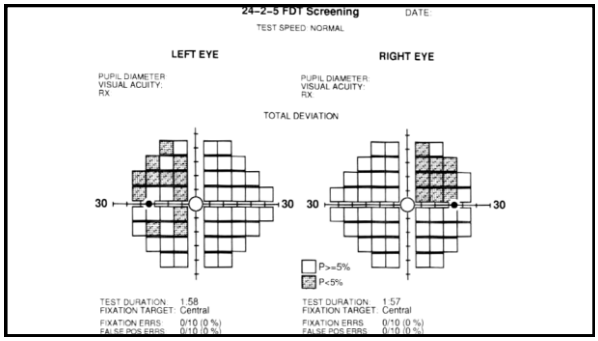
Tilted Disc Syndrome

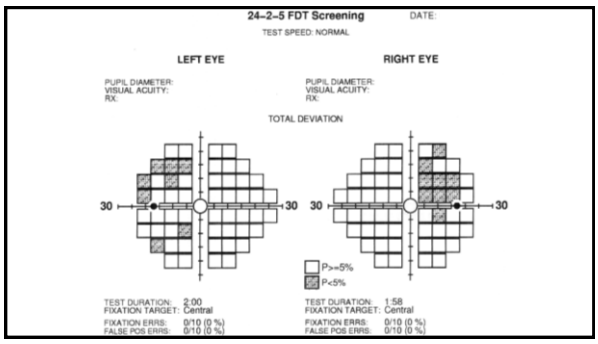


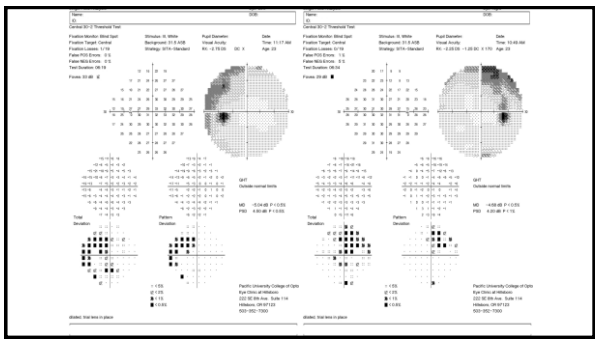
- 1-2% population
- Almost always bilateral
- Inferior-nasal tilt of optic disc
- Myopia, astigmatism association
- Bitemporal superior visual field defects
  - May be mistaken for chiasmal field defect
- Inferior crescent, vascular situs-inversus
- Lower fundus ectasia/staphyloma

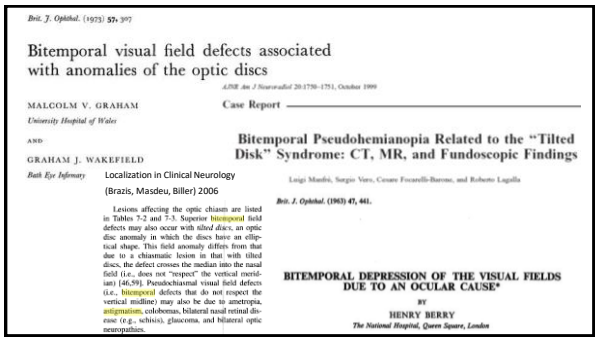
Tilted Disc Syndrome











Brit. J. Ophthal. (1973) **57**, 397

Bitemporal visual field defects associated with anomalies of the optic discs

MALCOLM V. GRAHAM
University Hospital of Wales
AND
GRAHAM J. WAKEFIELD
Bath Eye Infirmary

AJNR Am J Neuroradiol 20:1750–1751, October 1999

Case Report

Bitemporal Pseudohemianopia Related to the "Tilted Disk" Syndrome: CT, MR, and Fundoscopic Findings

Luigi Mandelli, Sergio Vaes, Cesare Focarelli-Barone, and Roberto Lagalla

Localization in Clinical Neurology (Brazis, Masdeu, Biller) 2006

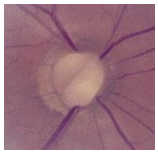
Lesions affecting the optic chiasm are listed in Tables 7-2 and 7-3. Superior bitemporal field defects may also occur with *tilted discs*, an optic disc anomaly in which the discs have an elliptical shape. This field anomaly differs from that due to a chiasmatic lesion in that with tilted discs, the defect crosses the median into the nasal field (i.e., does not "respect" the vertical meridian) [46,59]. Pseudochiasmal visual field defects (i.e., bitemporal defects that do not respect the vertical midline) may also be due to ametropia, astigmatism, colobomas, bilateral nasal retinal disease (e.g., schisis), glaucoma, and bilateral optic neuropathies.

Brit. J. Ophthal. (1963) **47**, 461.

BITEMPORAL DEPRESSION OF THE VISUAL FIELDS DUE TO AN OCULAR CAUSE*

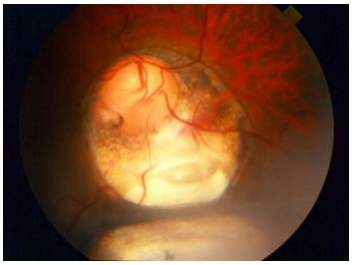
BY
HENRY BERRY
The National Hospital, Queen Square, London

Optic Disc Coloboma

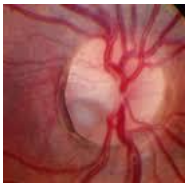


- Incomplete optic fissure closure
- Inferior-nasal quadrant most common
- 0.5-2.2 cases per 10,000
- Associated with many craniofacial syndromes
  - Goldenhar syndrome, Treacher-Collins syndrome, others
- Associated with numerous ocular conditions
  - Cataract, glaucoma, retinal detachment, field defects, etc.
  - Colobomas of eyelid, iris, lens, retina, choroid
- Monitor for retinal detachment, cataract, glaucoma
- Iris prosthetic lens, eyelid reconstruction

Optic Disc Coloboma



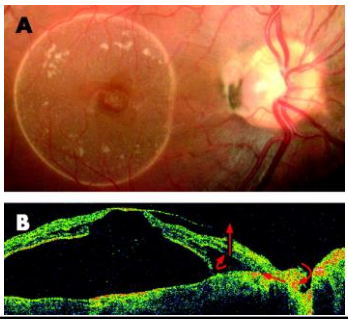
Optic Pit



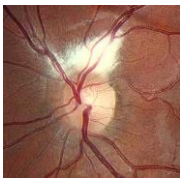
- 1 case per 10,000
- Usually unilateral
- Round or oval depression in optic cup
- Grey, yellow, or black in colour
- Inferio-temporal region of disc most common
- Caused by incomplete closure of optic fissure
- Serous macular detachment, edema, schisis possible
  - Debate as to origin of fluid (CSF, vitreous, sub-retinal)
- Visual field loss associated based on pit location

Optic Pit

Schaal KB, Wrede J, Dithmar S Internal drainage in optic pit maculopathy British Journal of Ophthalmology 2007;91:1093.



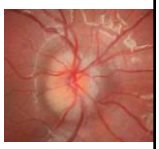
Myelinated Optic Nerve Fibers



- <1% general population
- Almost always unilateral
- Present at birth, static lesion
- Usually no symptoms
- Myelinated retinal nerve fibers anterior to lamina
- Large lesions can cause visual field defects
- Occasional association with anisometropic myopia, amblyopia, strabismus
- Association with Turner's syndrome, Trisomy 21, other syndromes

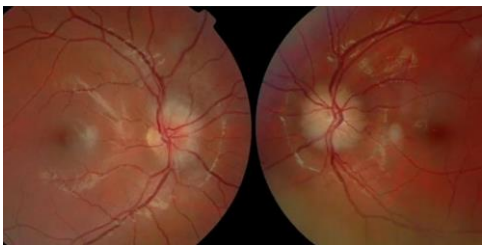
Myelinated Optic Nerve Fibers

Pseudopapilledema



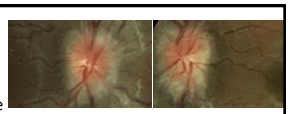
- "Pseudo" = false
- "Papilledema" = swollen optic disc from increased intracranial pressure (ICP)
- Elevated disc appearance without RNFL edema
- Most common cause: small hyperopic discs
- Other causes: disc drusen, myelinated disc, prominent Bergmeister's papilla, infiltration of the disc by mass or inflammatory debris, vitreopapillary traction, other causes
- Determine etiology; VF, Fundus exam, B-Scan, OCT, FAF, FANG, CT, MRI as indicated

Pseudopapilledema



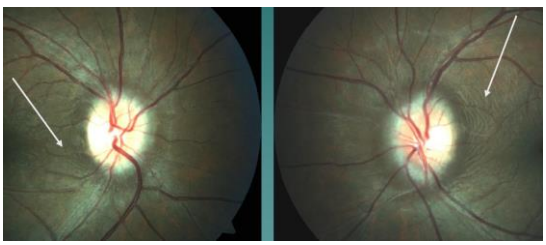
https://www.aao.org/annual-meeting-video/point-counterpoint-is-oct-useful-diagnosis-of

Papilledema



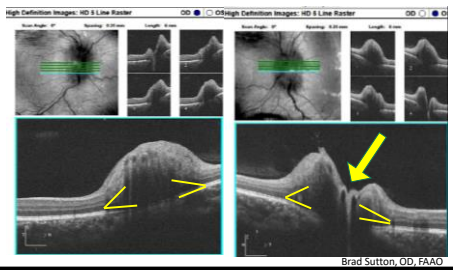
- BILATERAL optic disc swelling from increased intracranial pressure (IIH = idiopathic intracranial hypertension; formerly pseudotumor cerebri)
- Blurred margins, splinter hemes, exudates, RNFL edema, BV tortuosity, Paton's folds (retinal folds from disc) Transient blur, headaches, tinnitus, diplopia
- Normal acuities, color vision, pupils, and fields
  - Above may become abnormal if chronic (>21 days)
- Young slightly overweight females 8x > males
- Sleep apnea, medication (i.e. tetracyclines) link
- VF, OCT, MRI, MRV (to r/o cerebral venous sinus thrombosis), lumbar puncture (>250mm abnormal)

Papilledema



Brad Sutton, OD, FAAO

Papilledema



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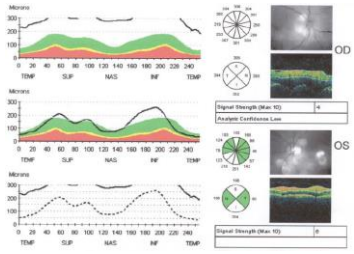


Brad Sutton, OD, FAAO

Side-note: Foster-Kennedy Syndrome

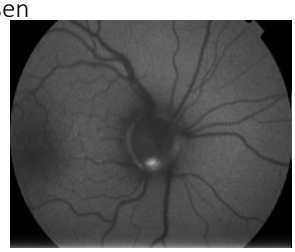
- One eye has swollen optic disc while other eye has advanced optic atrophy
  - Advanced optic atrophy prevents swelling
  - Makes bilateral condition appear to be unilateral
- Chiasmal tumors can cause; compresses and incr. ICP

Pastora-Salvador N, Peralta-Calvo J. Foster Kennedy syndrome: papilledema in one eye with optic atrophy in the other eye CMAJ December 13, 2011 183 (18) 2135

Disc Drusen

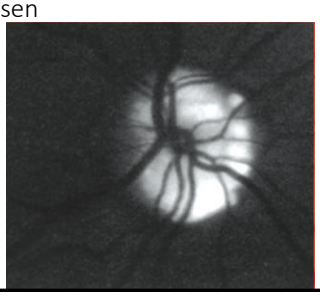
- Calcified hyaline protenaceous crystals in the ONH
- From axoplasmic flow disruption and excretion
- Builds up *in utero*; can push forward over years
- 3-24/1000; AD with variable penetrance, M = F
- Bilateral but may be asymmetric
- Optic nerves have a raised, "lumpy bumpy" look
- Vascular trifurcation off the central retinal stalk
- Severe RNFL and VF loss, but patients asymptomatic
- Fundus autofluorescence, B-Scan, OCT, VF diagnose

Disc Drusen

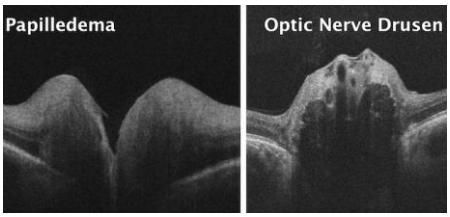


Pacific University Longitudinal Lipofuscin Study
John R Hayes, David Glabe, Len Hua, James Sheedy, Denise Goodwin, Dina Erickson, Lorne Yudcovitch, Nada Lingel, Tracy Doll, James Kundart, Matthew Lampa, Beth Kinoshita, Scott Pike

Disc Drusen

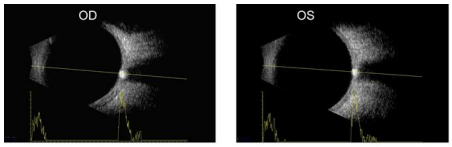


Disc Drusen



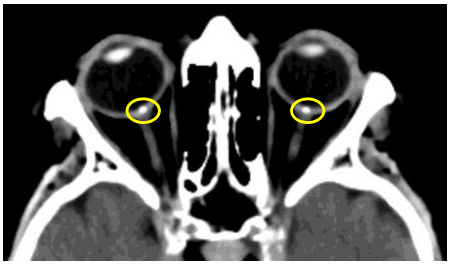
https://www.restorevisionclinic.com/optic-nerve-structure-function-damage-signs-treatment

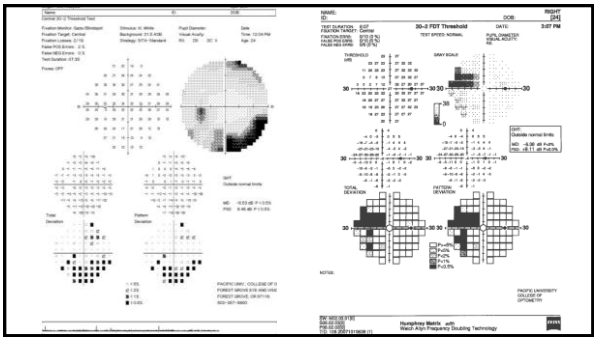
Disc Drusen

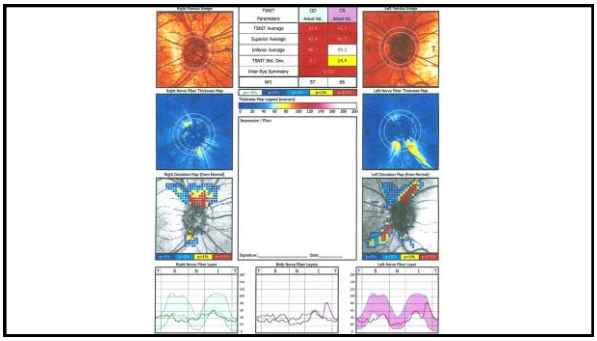


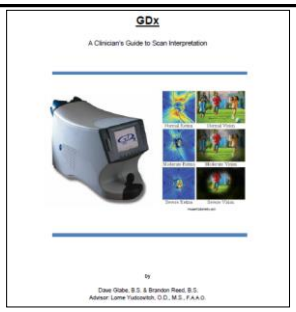
https://eyewiki.aao.org/Ultrasound_in_neuro-ophthalmology

Disc Drusen







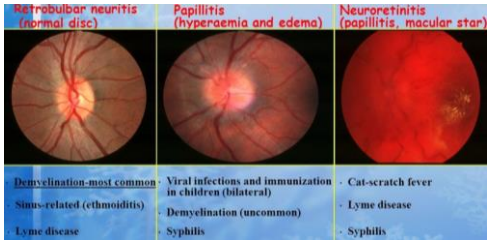


https://commons.pacificu.edu/work/ns/91027535-c925-45f4-824a-957ceee0abcb

Papillitis (Optic Neuritis)

- Autoimmune inflammation of optic nerve due to MS
- Demyelinating disease as cause, mechanism unknown
- Age 20-44, 3:2 female:male, flu may precede attack
- Relatively sudden decreased central and/or peripheral vision, reduced color and contrast/brightness sense, relative afferent papillary defect (RAPD), periorbital pain that worsens with eye movement
- May be first sign of multiple sclerosis; can recur
- Optic disc may look normal (retrobulbar optic neuritis)
- MRI of orbits, brain, and brainstem; IV steroid Tx

Papillitis (Optic Neuritis)



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Clinical Study

Hyperacute Corticosteroid Treatment of Optic Neuritis at the Onset of Pain May Prevent Visual Loss: A Case Series

G. T. Plant,[1,2,3] N. A. Sibtain,[4] and D. Thomas[2]

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[2] *Department of Neuro-Ophthalmology, Moorfields Eye Hospital, London EC1V 2PD, UK*
[3] *The Medical Eye Unit, St Thomas' Hospital, London SE1 7EH, UK*
[4] *Department of Neuro-Radiology, King's College Hospital, London SE5 9RS, UK*

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Academic Editor: Axel Petzold

Aim. To show that high-dose corticosteroids may prevent visual loss in patients with optic neuritis (ON) treated at the prodromal, hyperacute, phase of retrobulbar pain. *Method.* Prospective case series: patients were recruited with a history of ON associated with pain. The patients were advised to report immediately to the investigators should the pain recur in either eye. Where possible, orbital magnetic resonance imaging (MRI) was performed to confirm a recurrence of ON and treatment with high-dose corticosteroids was commenced. Visual function and the patient's subjective account were monitored. *Results.* Eight patients (including cases of MS, CRION and NMO) presented in the hyperacute phase. MRI confirmed optic nerve inflammation in 5/5. Treatment was commenced immediately, and, in all cases, no visual loss ensued. *Conclusion.* MRI can be used to confirm acute optic neuritis prior to visual loss in the hyperacute phase. We suggest that treatment with high-dose corticosteroids may abort the attack and prevent loss of vision in patients with ON who are treated at the onset of pain. This has potential implications for the management of acute ON and also for our understanding of the pathogenesis and potential therapeutic targets in the neuroinflammatory conditions associated with ON.

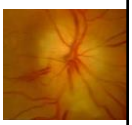
Multiple Sclerosis International Vol 2011 p 1-8.

Neuromyelitis Optica (NMO=Devic's Disease) Myelin Oligodendrocyte Glycoprotein (MOG)

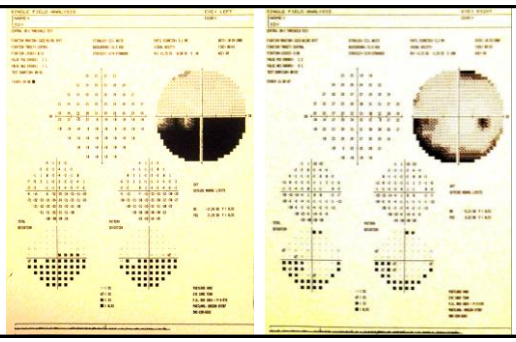
- NMO – any age, Asian or African descent, females more
  - Severe spinal cord and bilateral optic neuritis
  - Mild optic disc swelling but severe optic nerve damage
  - Poor prognosis for physical and visual recovery
  - Lupus, Sjogren syndrome, myasthenia gravis may accompany
- MOG – any age, any ethnicity, any gender
  - Moderate to severe optic disc edema, often bilateral, recur
  - Seropositivity of MOG-Ab in cell assays diagnoses it
  - >50% of optic nerve often seen inflammatory in MRI

For all these demyelinating diseases, IV steroids and immunosuppressant therapy often first-line treatment plan

Anterior Ischemic Optic Neuropathy (AION)



- "A stroke on the nerve"
- Most common optic neuropathy of older patients
- Non-arteritic (NAION) and arteritic (AAION) forms
  - Non-arteritic form less severe, ages 45-70 usually
  - Arteritic form more severe, older (>70) associated with giant cell arteritis (GCA); ocular emergency
- Sudden visual loss in one eye, RAPD, altitudinal field defect, swollen disc with possible splinter heme
- No current treatment for NAION; monitor
- Emergency IV steroid treatment for AION; taper > 1yr
  - 50% chance of bilateral blindness if not treated



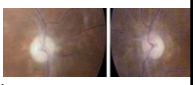
Nonarteritic versus Arteritic Anterior Ischemic Optic Neuropathy

FINDINGS	NON-ARTERITIC	ARTERITIC (GIANT CELL, TEMPORAL)
Patient age	Under age 55	Older than age 55
Visual acuities	20/20 to 20/60	20/70 to NLP
Visual field loss	Altitudinal (spares macula)	Altitudinal (involving macula)
ESR	May be normal	Elevated
Associated finding	None	Ipsilateral temple pain; headache
Associated finding	None	Jaw claudication

Differentiation of Elevated, Blurred Disc

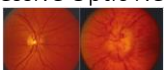
FINDINGS	Papilledema	Papillitis	Pseudo-papilledema	Disc Drusen
Visual Acuity	May be normal	Significant reduction	Normal	Normal to reduced
Pupils	Usually normal	Possible APD	Normal	Normal
Motilities	Normal	Pain	Normal	Normal
Color Vision	Normal	May be reduced	Normal	Usually normal
Visual field loss	Usually none	Significant	Normal (small blind spot)	None to significant
Associated finding	Disc hemes; usu. binocular	Disk hemes; monocular	Axial hyperopia	Disc hyaloid bodies

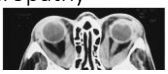
Toxic Optic Neuropathy (Nutritional Amblyopia)



- Optic nerve damage from a toxin and/or lack of a nutritional supplement
  - Long-term use of: Alcohols, Antibiotics, Antimalarials, Antitubercular, Antiarrhythmics, Anticancer drugs, Heavy metals, Carbon Monoxide, Tobacco, others
  - Lack of: thiamine (B1), riboflavin (B2), niacin (B3), pyridoxine (B6), cobalamin (B12), folic acid, others
- Cause: optic nerve mitochondrial damage, ischemia
- Diabetes, liver disease, kidney disease may worsen
- Bilateral, progressive optic atrophy, reduced vision
- Prognosis poor as often too late and irreversible

Compressive Optic Neuropathy



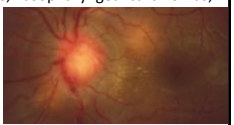


- Crushing of the optic nerve by a mass effect
  - infectious (e.g. orbital mycoses), inflammatory (e.g. orbital pseudotumor), vascular (e.g. aneurysm), traumatic (e.g. fracture, hematoma), neoplastic (e.g. meningioma, glioma), autoimmune (e.g. hyperthyroid)
- Slowly progressive acuity and/or visual field loss, dyschromatopsia/ color defect, RAPD, RNFL loss, initial normal or swollen disc and later optic atrophy
- Treatment depends on cause, with ultimate goal of reducing pressure on optic nerve to preserve vision

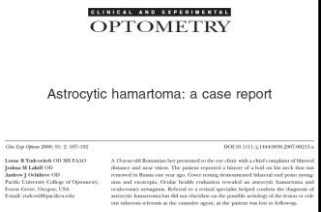
Compressive Optic Neuropathy

Infiltrative Optic Neuropathy

- Optic nerve becomes filled with pathological tissue:
- Tumors
  - Primary tumors - optic gliomas, capillary hemangiomas, cavernous hemangiomas, astrocytomas
  - Secondary tumors - metastatic carcinomas, nasopharyngeal carcinomas, lymphomas, leukemias
- Inflammation
  - Most common: sarcoidosis
- Infection
  - Opportunistic fungi, viruses, and bacteria
- Optic disc may be elevated if infiltration occurs in proximal portion

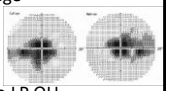


Infiltrative Optic Neuropathy

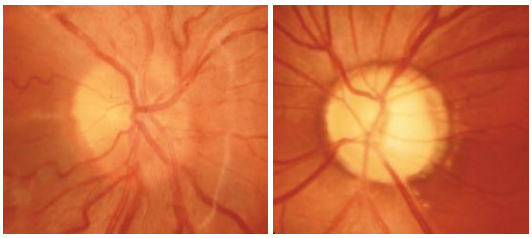


Leber Hereditary Optic Neuropathy (LHON)

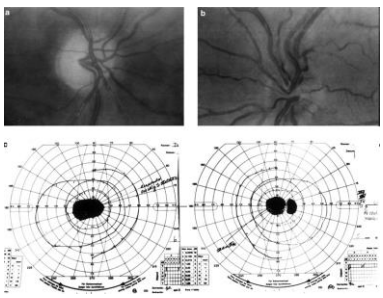
- The most common inherited mitochondrial disease
- Usually affects males between 10-30 years of age
- Unilateral progressive optic neuropathy with fellow eye involved months to years later
  - Disc telangiectic vessels and edema → later pallor
- Cecocentral visual field loss, acuity may drop to LP OU
- Heart conduction abnormalities, dystonia, and MS-like symptoms may accompany ("LHON Plus" disease)
- Antioxidant supplements, gene therapy trials



Leber Hereditary Optic Neuropathy (LHON)



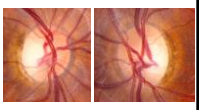
https://www.semanticscholar.org/paper/Clinical-features-of-Leber's-hereditary-optic-with-Hung-Kao/527949443cd5d28735ec79a52831a188ecce3adf/figure/1



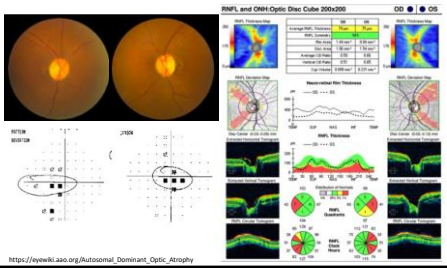
From: Newman NJ, Biousse V. Hereditary optic neuropathies. *Eye* (2004) 18, 1144–1160. doi:10.1038/sj.eye.670159

Dominant Optic Atrophy

- The most common hereditary optic neuropathy
  - 1:12,000 to 1:50,000 prevalence
- Slow bilateral vision loss in 1st to 2nd decade of life
- Focal, wedged-shaped temporal disc atrophy or diffuse disc atrophy
- Papillomacular bundle primarily affected:
  - Central, centrocental paracentral scotomas
  - Color vision defects (including tritan defects)
  - VEP: amplitude reduced, latency prolonged
  - PERG: reduced
- Nuclear OPA1 gene mutation

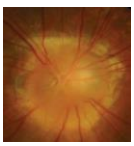


Dominant Optic Atrophy



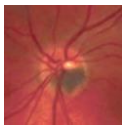
https://eyewiki.aao.org/Autosomal_Dominant_Optic_Atrophy

Morning Glory Anomaly



- Rare congenital optic nerve malformation
- Optic disc looks like a morning glory flower
- Likely defect of fetal optic fissure closing
- Possible craniofacial, cerebrovascular anomalies
  - Morning glory syndrome = optic nerve + systemic anomalies
- RAPD and/or retinal detachment can rarely occur
- Management via best-correction, amblyopia treatment, dilated retinal exams, cranial imaging
  - Basal encephalocele: malformed meninges outpouching which protrudes through sphenoid bone defect

Melanocytoma



- Usually benign pigmented neoplasm of optic disc
  - Raised dark brown to black lesion on the optic disc
- Usually unilateral; slight predilection for females
- Usually no visual impairment or other symptoms
- Very slow to no growth, with rare malignant conversion
- Rare disc edema, hemorrhages, vein occlusion
- Monitor with photos, B-Scan, OCT, FA

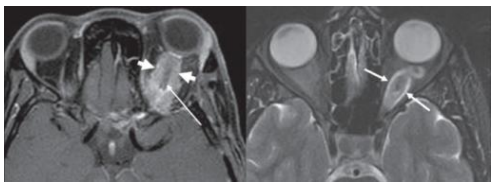
Optic Nerve Meningioma/Glioma

- Tumor derived from cells of the nerve meninges
- More frequent in adult women age 30-40s
- Classic triad:
1) Slow vision loss
2) optic atrophy
3) optociliary shunt vessels

https://commons.wikimedia.org/wiki/File:Optic_nerve_glioma.png

- Optic nerve may look normal or edematous at first
- CT and MRI of orbits and brain indicated

Optic Nerve Meningioma/Glioma

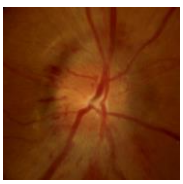


Optic nerve sheath meningioma (thick arrows) and optic nerve (thin arrow)

Glioma of optic nerve

https://commons.wikimedia.org/wiki/File:Optic_nerve_sheath_meningioma.png

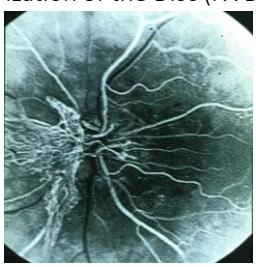
Diabetic Papillopathy



- Rare unilateral or bilateral disc edema
- Different than neovascularization of the disc (NVD)
- Can present in both Type 1 and Type 2 DM
- Possible NAION variant; microangiopathy cause
- Good prognosis, none-minimal symptoms, usually self-resolves
- *Very important to rule-out other causes of bilateral (i.e. papilledema) or unilateral (i.e. AION) disc edema, though!*

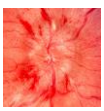
https://www.columbiaeye.org/education/digital-reference-of-ophthalmology/neuro-opthamology/diabetic-papillopathy

Neovascularization of the Disc (NVD)

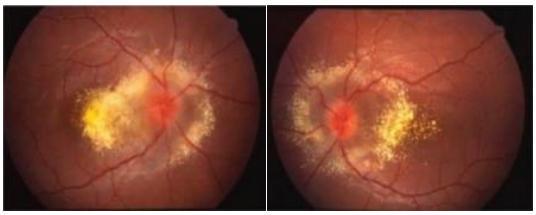


Hypertensive Papillopathy

- Bilateral disc edema caused by very high blood pressure
  - JNC 7 emergency level >180/120 with end organ dysfunction
- Often present with transient or constant visual blur
- Notable disc elevation, with possible splinter hemes
- Exudates surrounding the optic disc may be present
- Macular star (radial exudates in the macula)
- Neuroimaging indicated to rule-out intracranial mass
- Elevated intracranial pressure from increased cerebrospinal fluid may require lumbar puncture
- Aggressive anti-hypertensive treatment indicated



Hypertensive Papillopathy



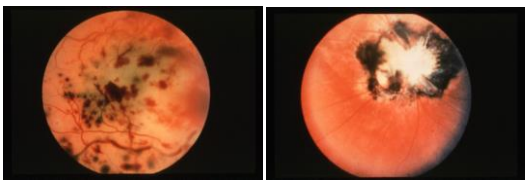
7yo patient, BP 240/160

Shah, Veeral et al. "Bilateral papillopathy as a presenting sign of pheochromocytoma associated with von Hippel-Lindau disease." *Clinical ophthalmology (Auckland, N.Z.)* vol. 8 623-8. 26 Mar. 2014, doi:10.2147/OPTH.S60725

Traumatic Optic Neuropathy

- Various types of physical trauma can cause
  - Penetrating, blunt, crushing, etc.
- Most devastating complication
- Usually results in total blindness (NLP)
- Total afferent pupillary defect
- Avulsion of the optic nerve
  - Optic nerve forcibly disinserted from retina, choroid, and vitreous
  - Lamina cribrosa is retracted from the scleral rim
- B-Scan, orbital CT or MRI confirms

Traumatic Optic Neuropathy



Retinal infarction, retinal edema, cherry red macula, and preretinal hemorrhages obscuring the optic nerve

Damage to circulation and insertion of optic nerve
No vision (NLP)

THANK YOU!

yudcovil@pacificu.edu