Optic Nerve Abnormalities in Children

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Evaluation of the Pediatric Optic Nerve

- Is it Swollen?
- Is it Pale?
- Is it Small?
- Is it Cupped?
- Is it Funky?

Optic Nerve abnormalities in Children

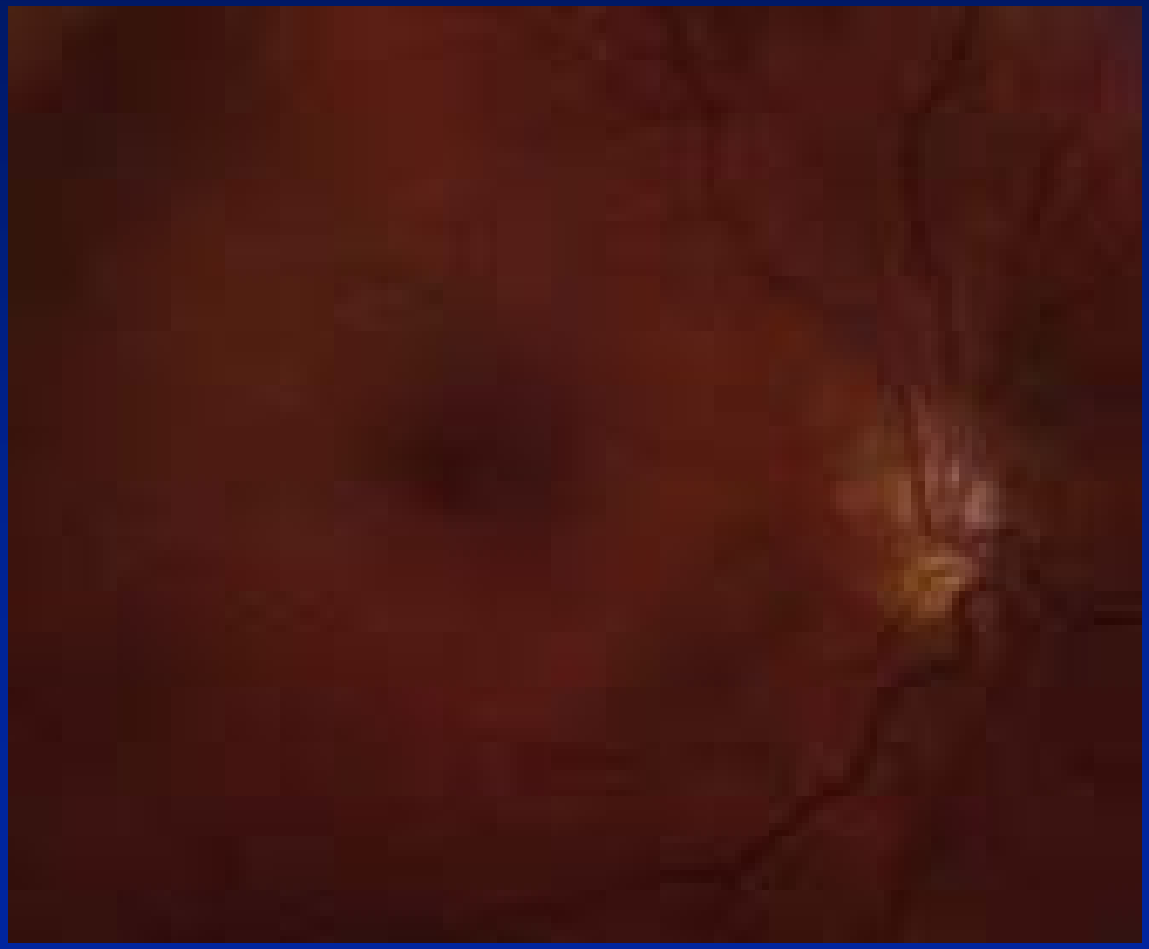
- Not only a cause of poor vision, but often a clue to important systemic abnormalites

- Optic disc may be abnormal in
  - size
    - eg. hypoplastic discs
  - elevation
    - eg. papilledema, inflammation, pseudopapilledema
  - excavation
    - eg. glaucomatous cupping, nonglaucomatous cupping, coloboma
  - color
    - optic atrophy, hyperemia

Optic Nerve hypoplasia

- Decreased number of optic nerve axons from damage before full optic nerve development
  - » 1st or second trimester
    - – later damage would likely manifest as a cupped disc.
- Endocrine abnormalites reported in 6-71%

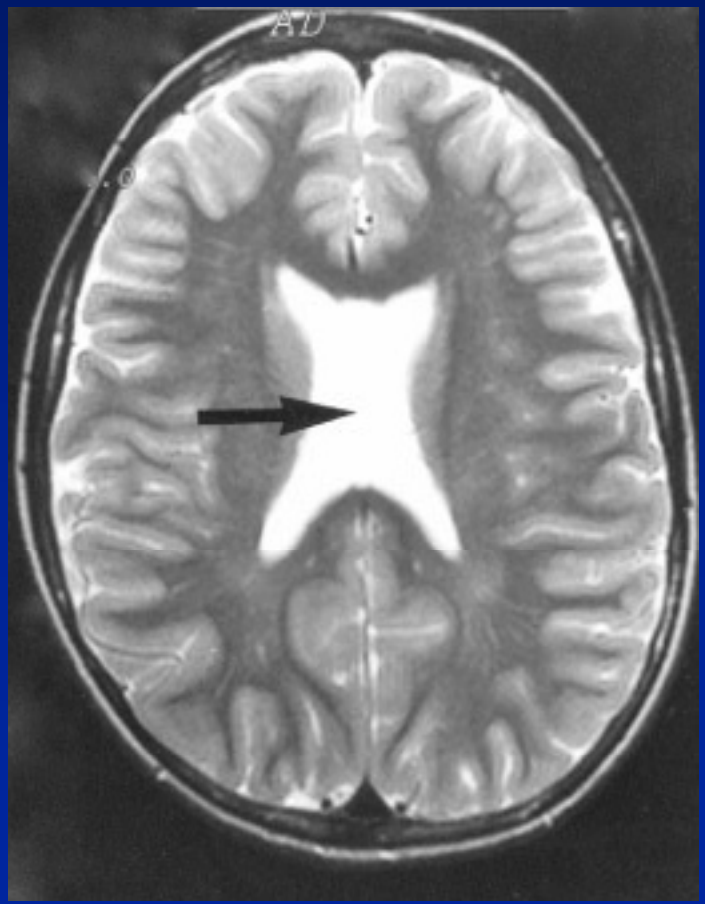
Hypoplastic Optic Nerve



Optic Nerve hypoplasia

- Most commonly bilateral
  - may be unilateral in up to 25%
- Brain abnormalities in 60%
  - absence of septum pellucidum
  - agenesis or thinning of corpus collosum
  - cerebral hemisphere abnormalities

Absence of Septum Pellucidum



Optic Nerve hypoplasia

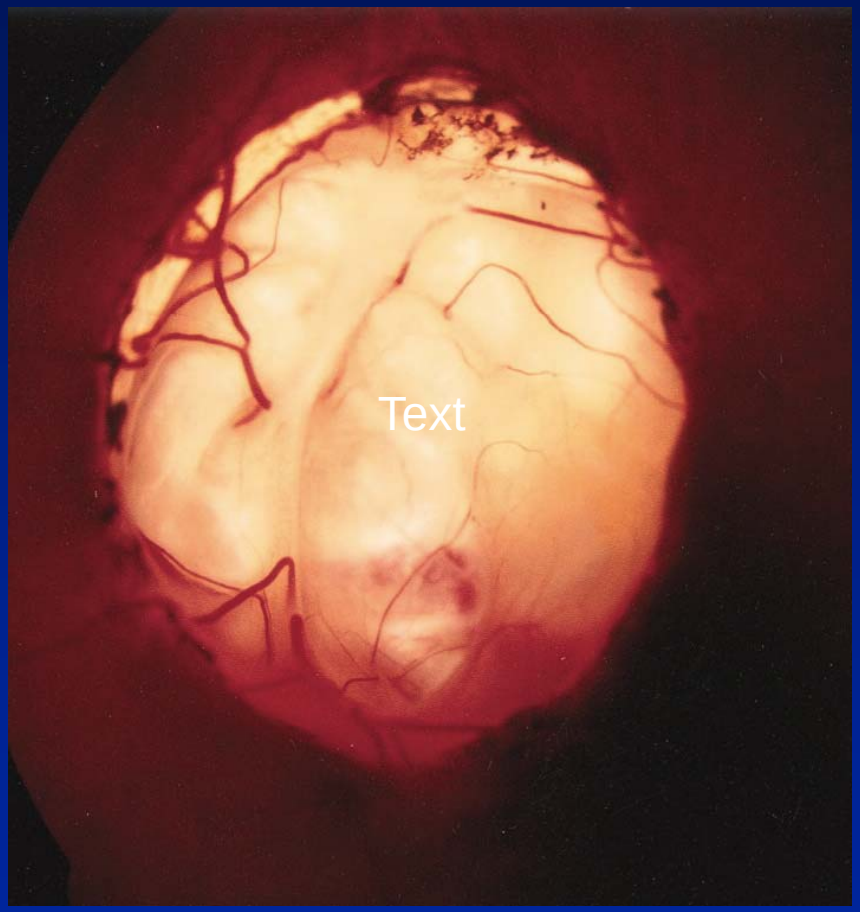
- endocrine abnormalities in majority of patients
  - reported range 6-71%
  - prospective studies with endocrine testing by endocrinologist tend to show higher prevalence than retrospective studies.
  - may not correlate with MRI findings of pituitary abnormalities, absence of septum pellucidum or unilateral versus bilateral hypoplasia
  - hypopituitarism can predispose to development delays

- MRI with contrast and if possible thin cuts through sella turcica recommended for all
- endocrine evaluation - definite if MRI abnormal or clinical evidence of endocrinopathy. Personally recommend for all

Optic nerve hypoplasia

- Developmental delay common
  - up to 71% in some studies
  - High rate of CP
  - higher association with:
    - structural brain abnormalities
    - bilateral hypoplasia
    - hypothyroidism

Excavated Disk



Coloboma

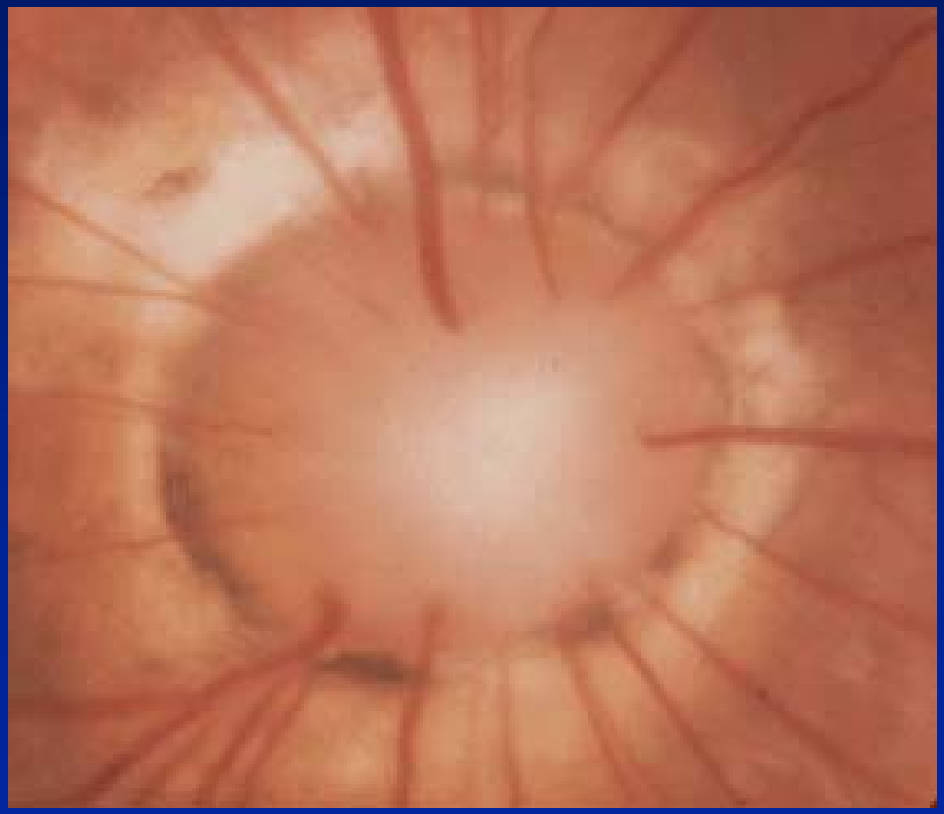
Optic nerve excavation

- Colobomas
  - proximal end of embryonic fissure fails to close
  - 80% sporadic, 20% inherited (AD)
  - risk of serous detachment of macula
- Associated systemic abnormalities
  - CHARGE
    - chorioretinal/optic nerve colobomas, heart defects, choanal atresia, growth retardation, genital anomalies, ear anomalies or deafness
    - CHD7 gene defect

Colobomas

- Systemic abnormalities
  - Brachio-Oculo-Facial Syndrome
    - (TFAP2A mutation)
    - multiple systemic anomalies, including dental, auricular pits etc.
  - Renal Coloboma Syndrome
    - AD
    - PAX2 gene mutation
    - bilateral disk anomalies, renal hypoplasia, sensorineural hearing loss

Morning Glory Disk



Morning Glory Disc

- Sporadic
- Usually unilateral
- Central glial tuft
- Characteristic retinal vascular pattern
- Likely due to poor development of the lamina cribosa and posterior sclera with herniation of neural elements
- Serous RD in 1/3 of patients over 10 years

Morning Glory Disc

- Associated findings
  - » cleft lip and palate
  - » basal encephalocele
  - » agenesis of the corpus collosum
  - » Moya Moya Disease
    - – constriction of vessels (ICA), with collateralization, aneurysms, thromboses
- MRI, MRA or CTA indicated

Nonglaucomatous Cupping

- Most young children have small cups
  - only .3% of newborns >0.3 CD
- Optic disk diameter increases during childhood
  - .93mm in infants, 1.59mm in 10 year olds
  - some increase in CD during this time.

Nonglaucomatous Cupping

- Increased CD associated with low birth weight
  - » reported associations with:
    - – periventricular leukomalacia/intraventricular hemorrhage
- hydrocephalus
- dominant optic atrophy (rim pallor)

Glaucoma in children

- Usually other visible signs
  - » Infantile glaucoma
    - – large globes, cloudy corneas, photophobia
  - » history of cataract with aphakia, pseudophakia
  - » signs with other syndromes
    - – eg. Aniridia
  - » normal tension glaucoma is disease of adults
    - – extremely rare to have childhood glaucoma with normal IOP

Optic Atrophy

- Prematurity
- Compressive Optic Atrophy
- Dominant Optic Atrophy
- Lebers Hereditary Optic neuropathy

Optic Atrophy

- Prematuriy
  - » in study by Mudgil and Repka at tertiary care facility, prematurity was the most frequent cause of optic atrophy in Children
    - – most of these children had history of intraventricular hemorrhage
- Tumor
  - » second most common cause
- Hydrocephalus
  - » third most common cause

Compressive Optic Neuropathy

- Get MRI in all cases of optic atrophy
  - » optic atrophy is not acceptable diagnosis
- Tumors may affect visual pathways by
  - » Direct compression
  - » Infiltration
  - » increased intracranial pressure

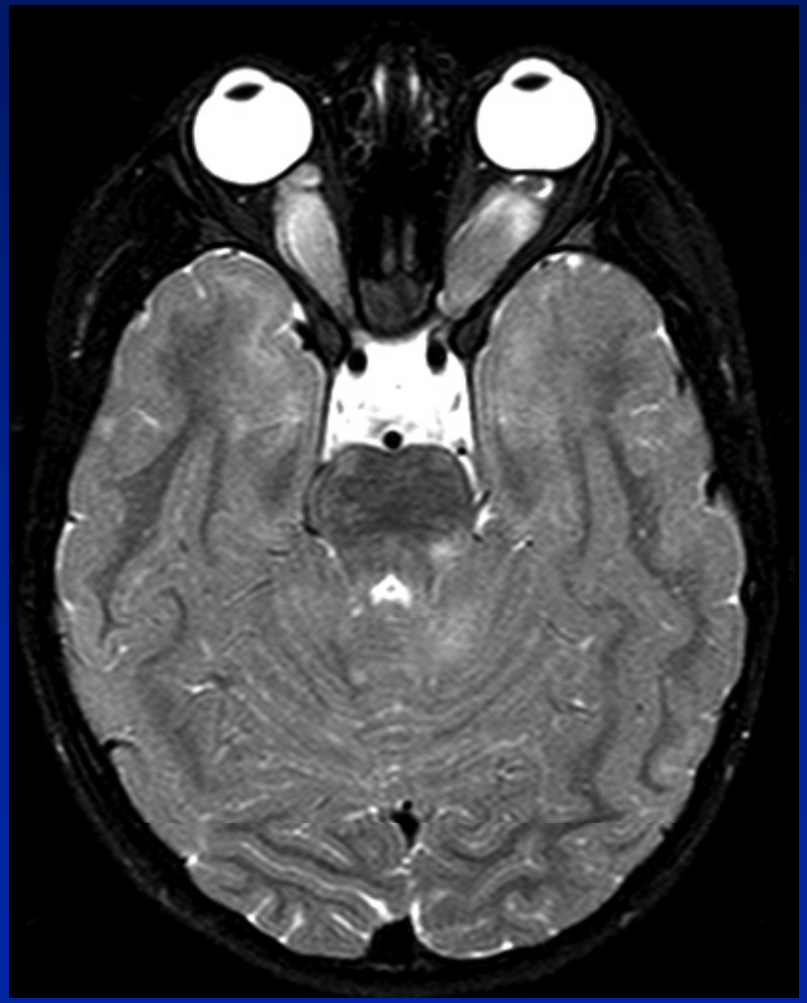
Compressive Optic Neuropathy

- Most common tumors affecting vision in children
  - » optic pathway gliomas
  - » craniopharyngiomas
  - » suprasellar gliomas (aka Juvenile pilocytic astrocytoma
  - » pineal tumors
  - » pituitary adenoma

Optic Pathway Glioma

- Commonly associated with Neurofibromatosis type 1
  - » 15-20%
  - » usually occur in the first 6 years of life
  - » unusual, but possible to develop or progress beyond age 7
  - » 88% remain stable
  - » vision loss in 20-70%
    - – usually at time of dx
    - – may progress
  - » important to monitor into adolescents
    - – vision, color vision, fields, pupils and ?OCT

Bilateral Optic Nerve Glioma



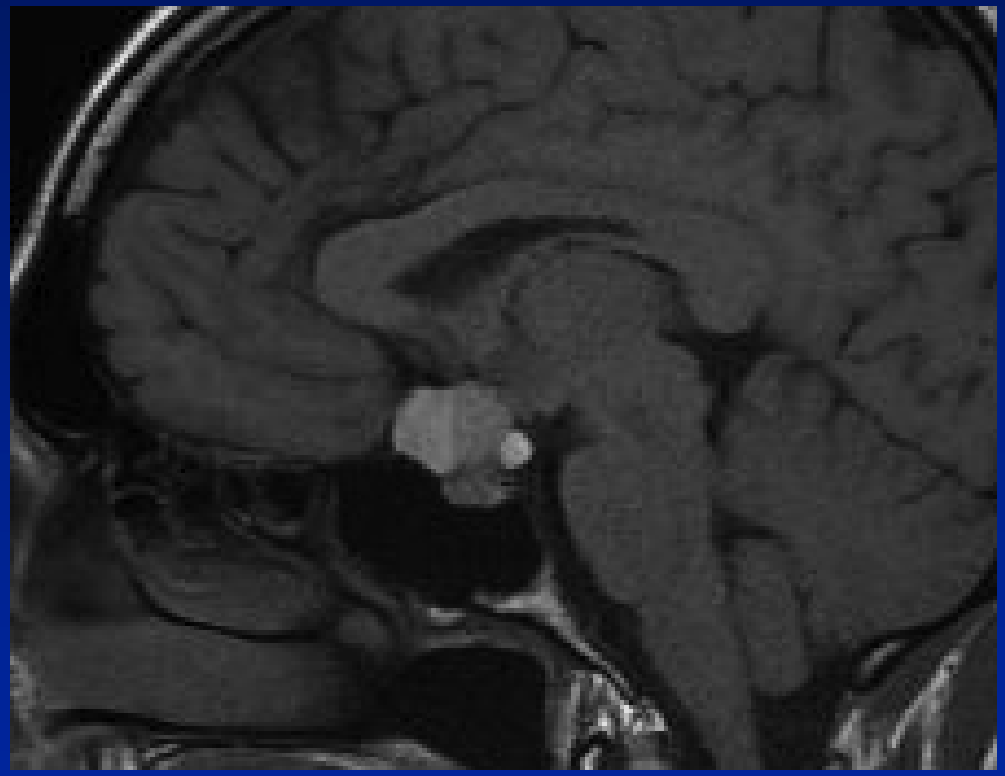
Optic Pathway Glioma

- Asymptomatic gliomas in NF-1 do not require treatment
  - » imaging of all children controversial
- 1st line treatment for symptomatic glioma is chemotherapy
  - » goal is tumor shrinkage and stability
  - » radiation increases risk of additional tumors and vascular malformations such as Moya Moya
  - » surgery may be considered for proptosis in a blind eye

Optic Pathway Glioma

- Sporadic (not associated with NF-1)
  - tend to be much more aggressive
  - also tend to occur in young age group
  - higher incidence of increased ICP, decreased acuity, optic atrophy, proptosis
  - unilateral
  - monitor closely
  - less risk of second tumors related to radiation

Craniopharyngioma



Craniopharyngioma

- typically age 5-14 and in older adults
- benign suprasellar mass
- causes local destruction from slow growth
- solid and cystic components
- form from remnants of Rathke's pouch
- may compress optic chiasm, nerves or tract

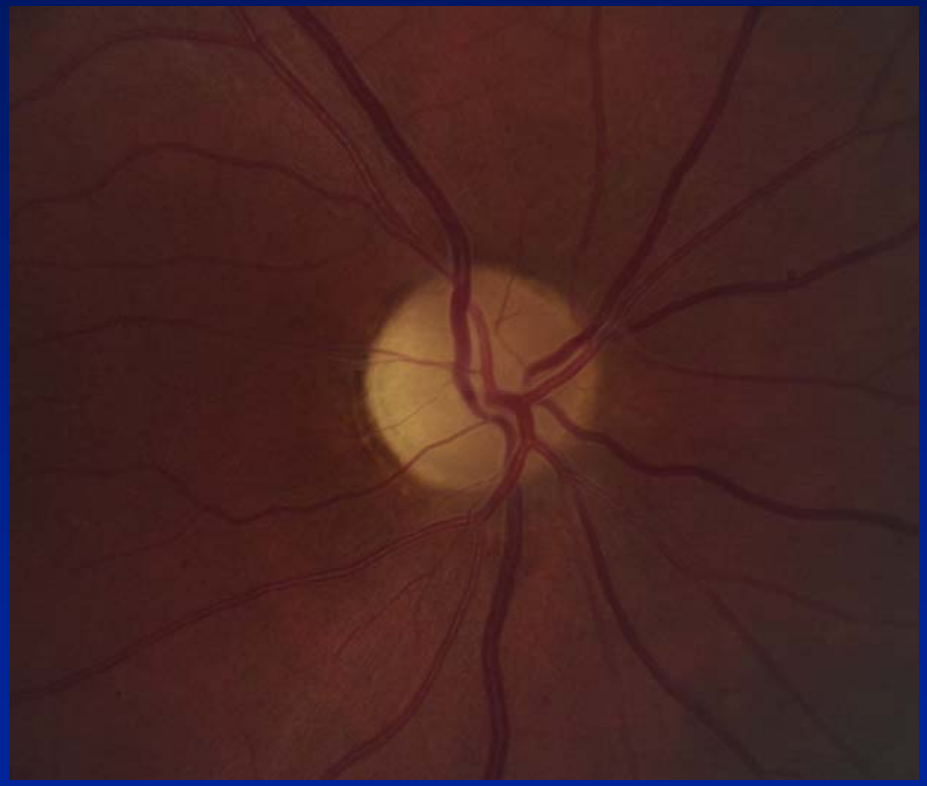
Craniopharyngioma

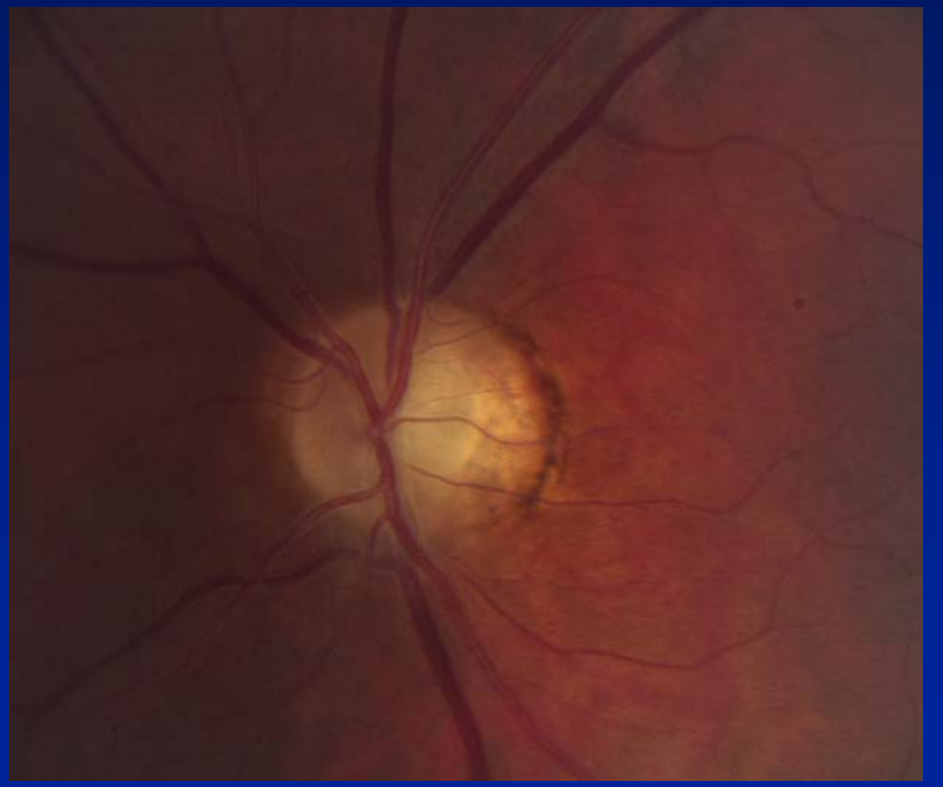
- most commonly present with headache or vision loss
- about half have major visual field defects
- Treatment is typically surgery to debulk tumor and treat hydrocephalus.

Dominant Optic Atrophy

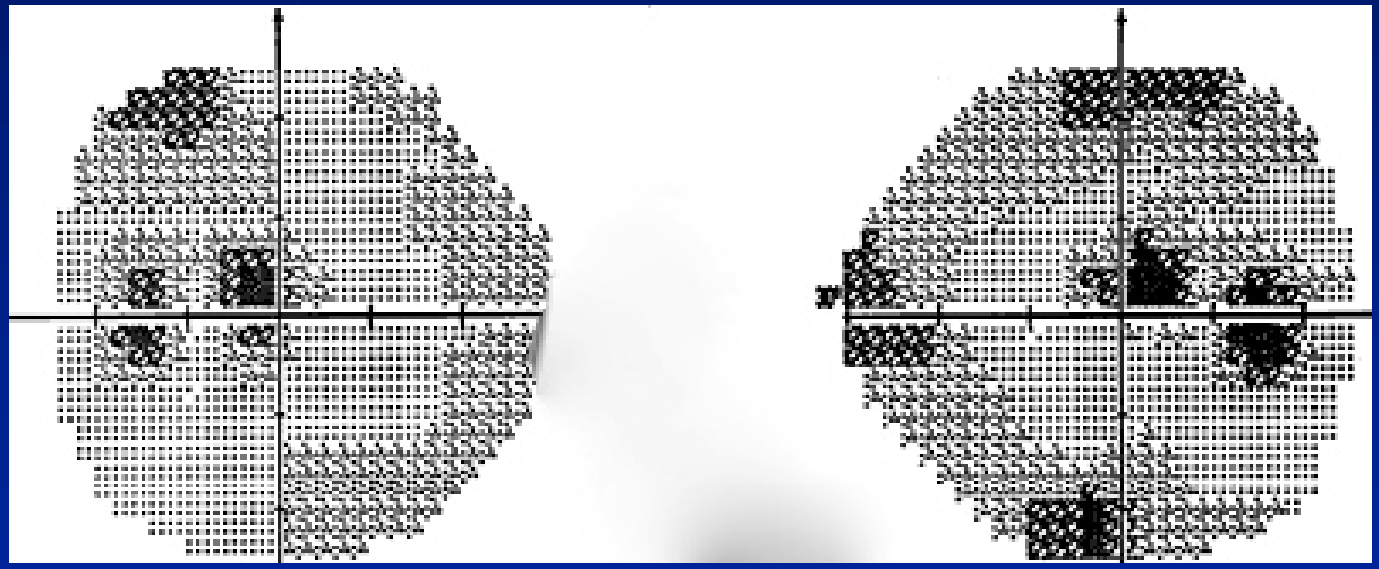
- mild to moderate vision loss
  - » final vision 20/20 to 20/200
  - » affects central vision, peripheral spared
- slowly progressive
- optic atrophy is usually symmetric
- classic bitemporal pallor, but diffuse in 50%
- occasionally associated with hearing loss

Dominant Optic Atrophy





Cecocentral Scotoma



Dominant Optic Atrophy

- expression is highly variable
  - » may have unrecognized family members
- spontaneous mutations not unusual
  - » may not have family members involved
- most commonly OPA1 gene

additional optic nerve conditions

- To Follow in Pediatric Cases not to be missed

Pediatric Pseudotumor Cerebri

- Definition:
  - Increased intracranial pressure
    - ? Lower normal ICP in children
  - Normal to small ventricular size
  - Normal CSF composition
  - ? Papilledema
  - Nonfocal neurological exam except CN6

Pediatric Pseudotumor Cerebri

- Symptoms
  - » Headache
    - – Often continuous, present on awakening
  - » Younger children may present with apathy or irritability
  - » transient visual obscurations
  - » pulse synchronous tinnitus
  - » visual loss

Pediatric Pseudotumor Cerebri

- Study from Dalhousie University, Halifax
- Retrospectively looked at population (2-15 years) of over 200,000 and found an annual incidence of 0.9 per 100,000
  - » Lower when looking at patients under 11
  - » Found higher incidence in females
  - » Majority of other studies found no sex predilection in preadolescent patients

Pediatric Pseudotumor Cerebri

- Pseudopapilledema is common
  - » Optic nerve head drusen present in 1-2% of population

Pseudotumor cerebri sine papilledema

- University of Utah Neuro-ophthalmology unit
  - » 353 patients with idiopathic intracranial hypertension between 1990-2003
    - – 5.7% without papilledema
    - – Of these 75% had SVP seen
    - – Mean opening pressure 309mm vs 373
    - – Visual field changes seen often nonphysiologic
- Krishna R, Kosmorsky GS, Wright KW
  - » Report 17 yo with headache, unilat 6th, no papilledema and LP opening pressure of 440 mm H2O

Pediatric Pseudotumor Cerebri

- Associations (partial list)
  - Tetracycline
  - Vitamin A intoxication
  - Steroid withdrawal
  - Systemic lupus
  - Growth Hormone use
  - Dural Sinus Thrombosis
  - Down's Syndrome
  - Sleep Apnea

Evaluation of Patient with Papilledema

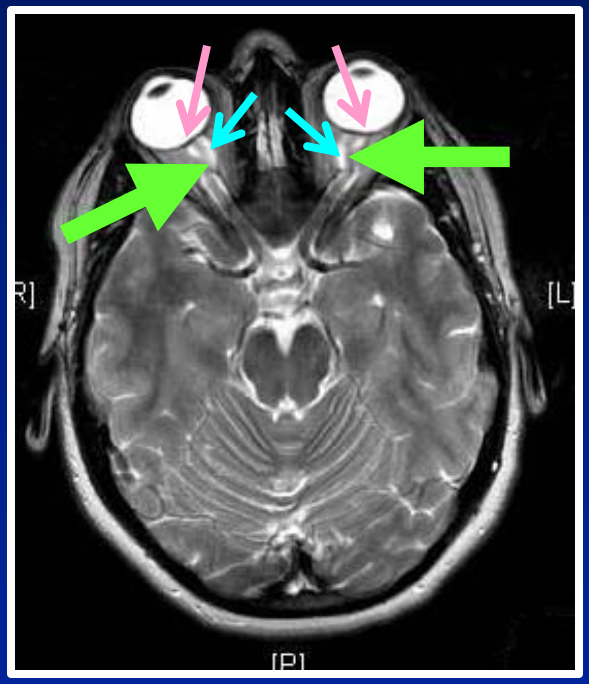
- Careful evaluation of pupils, color vision
- Visual Fields (if age appropriate)
  - Enlarged Blind spot
  - peripheral constriction
  - Inferonasal quadrant defect
- Disc Photos

Evaluation of patient with papilledema

- Neuroimaging
  - Usually MRI, although CT may be helpful if history suggestive of acute hemorrhage
- Lumbar Puncture
  - Measurement of opening pressure (specify) and composition r/o meningitis
- Other modalities
  - OCT
  - ultrasonography

Evidence of Intracranial Hypertension on MRI

Our Patient

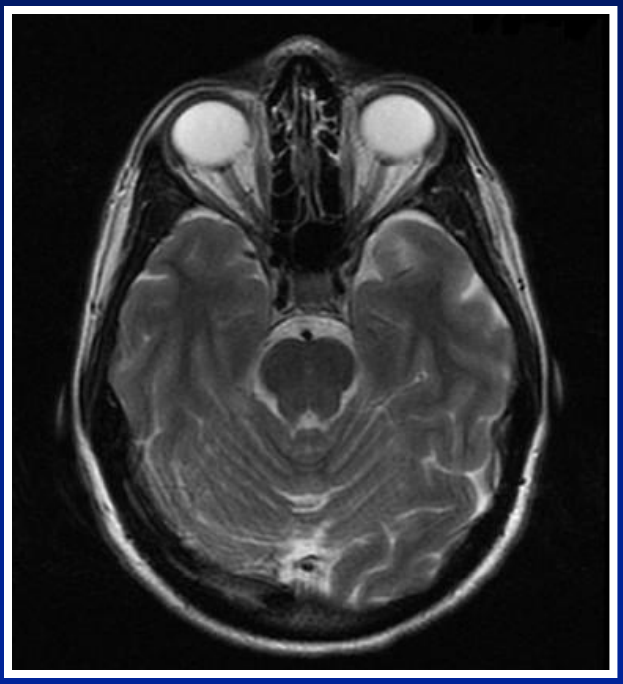


Axial T2 MRI
(PACS BIDMC)

Findings

- Increased fluid in optic nerve sheath bilaterally (bright signal)
- Mild flattening of posterior globes
- Tortuous optic nerves

Normal



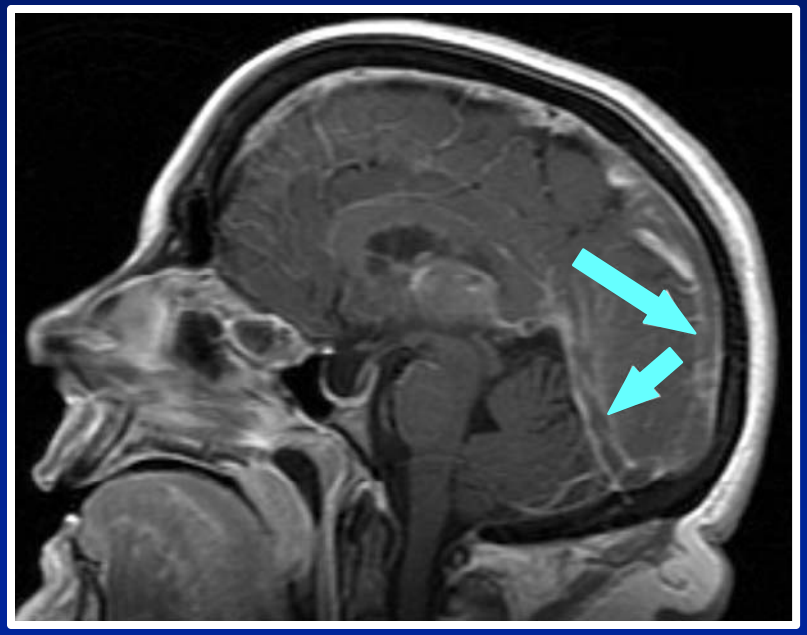
Axial T2 MRI
(PACS BIDMC)

Michael A. Dyer
Harvard Medical School, July 2009

Venous Thrombosis on MRI

Venous Thrombosis

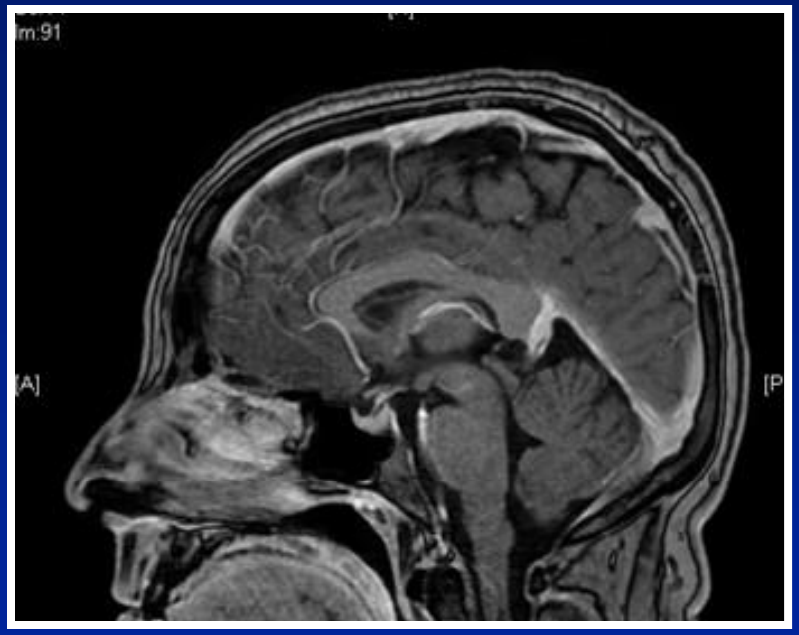
(filling defect in superior and straight sagittal sinuses)



Sagittal C+ T1 MRI

(Poon, CS, et al; AJROnline)

Index Patient: Normal



Sagittal C+ T1 MRI

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OCT in the evaluation of pediatric pseudotumor cerebri

- Duke University Medical Center
  - » Evaluated 11 clinically diagnosed PTC patients and 37 controls
  - » Increased nerve fiber layer thickness in the temporal and superior quadrants of Fast RNFL protocol centered on disc (inferior using Fast RNFL (3.4) protocol
  - » Increase macular thickness

OCT in the evaluation of pediatric pseudotumor cerebri

- Rebolleda G, Munoz-Negrete FJ
  - » Correlated peripapillary RNFL abnormalities in OCT patients with mild papilledema to visual field sensitivity loss
- ?What will happen to OCT in patients with severe papilledema that begin to get optic atrophy.

Ultrasonography

- 30 degree test
  - » If nerve diameter is increased on ultrasonography in primary position
  - » Patient is instructed to look 30 degrees temporally
  - » Decrease in diameter of 10% or more suggestive of fluid within nerve sheath
  - » Unchanged diameter suggests solid tissue
  - » May be difficult in pediatric population

Pediatric Pseudotumor Cerebri

- Treatment
  - » Elimination of cause if present
  - » Medication
    - – Diamox
    - – Lasix
    - – Steroids
    - – Heparin/Coumadin with sinus venous thrombosis

- Treatment
  - » Surgery
    - – Ventriculoperitoneal shunt
    - – Optic Nerve sheath fenestration
    - – ?Venous stenting (venous thrombosis)