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 abnormalities

» 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 abnormalities 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

1. 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

- Prematurity
  - 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
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

**Findings**

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

**Our Patient**

![Axial T2 MRI (PACS BIDMC)](image)

**Normal**

![Axial T2 MRI (PACS BIDMC)](image)

Michael A. Dyer
Harvard Medical School, July 2009
Venous Thrombosis on MRI

Venous Thrombosis
(filling defect in superior and straight sagittal sinuses)

Index Patient: Normal

Sagittal C+ T1 MRI
(Poon, CS, et al; AJROnline)

Michael A. Dyer
Harvard Medical School, July 2009
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)