Lawrence Kaufman, MD
Pediatric Ophthalmology for the Non-Ophthalmologist Symposium
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Optic Nerve Diseases

Optic Nerve Disease in Children

Congenital anomalies
Physiologic Cupping of the Optic Disc
The elevated disc
Optic neuritis
Optic atrophy

Congenital anomalies

Optic nerve aplasia
Optic nerve coloboma
Morning glory disc
Optic nerve hypoplasia
Peripapillary staphaloma
Megalopapilla
Optic pit
Congenital tilted disc
Optic disc dysplasia
Congenital optic disc pigmentation
Aicardi syndrome
Doubling of optic disc
Albinotic optic disc
V- or tongue shaped infrapapillary depigmentation

Optic nerve coloboma

- Results from failure of closure of posterior embryonic fissure
- Unilateral or bilateral
- Isolated or syndromic
- Associations: microphthalmia with cyst, CHARGE syndrome, Walker-Warburg syndrome and Aicardi syndrome.

Morning glory disc

- Optic disc appears enlarged within a larger excavation, and with overlying glial tissue obscuring the disc
- Associations: transsphenoidal basal encephalocele, and progressive constriction of the internal carotids (moyamoya disease).

Optic nerve hypoplasia

- Diagnosis based on clinical criteria
- Small optic disc
- +- atropy
- +- double ring sign

- Decreased vision
- 50/50 unilateral/bilateral
- Nystagmus in bilateral cases

- May occur as isolated ON finding or with CNS changes or Pituitary deficiencies
- Many associations: albinism, aniridia, FAS, maternal diabetes, maternal ingestion of dilantin/LSD, chromosomal anomalies
- W/U: MRI brain/orbit, attention to pituitary gland and optic stalk
  - Peds Neurology
  - Peds Endocrinology

Physiologic Cupping of the Optic Disc

- Large number of children with large cup/disc
- Higher incident in low birth weight, prematurity, periventricular leucomalacia,periventricular hemorrage, myopia
- Timing of brain insult may be important, cutoff at 28 weeks PCA
- Must distinguish from pathologic cupping due to glaucoma
- Serial exams: IOP, corneal clarity/size, anterior chamber depth, optic disc, refraction
- Disc photos
- Automated visual fields
- Other ancillary test
- Working Dx: Glaucoma suspect
- Exams q 6–12 months, until family fatigues and is lost to follow up, or enough time passes without progression to call physiologic (years)

The elevated disc

Congenital anomalies
Pseudopapilledema
Papilledema
ON compression
Ischemia
Inflammatory disease
Infiltrative disease
Toxic optic neuropathies
Hereditary optic neuropathies
Tumor
Vascular disease
Systemic disease
Shaken baby
Ocular hypotony

Pseudopapilledema

- An elevated disc appearance *not* due to elevated intracranial pressure; usually as a variation of normal or some benign process.
- Requires an ophthalmologist to make the call.
- The most important aspect is to inform the patient/family of the diagnosis so that in emergency setting such as in ER after head trauma that attending MD does not assume the disc changes represent elevated intracranial pressure.

Papilledema - Disc swelling due to elevated ICP

- *Hydrocephalus*
- Primary
- Space occupying intracranial lesions
- Cerebral edema
- Reduction in size of cranial vault
- Blockage of CSF flow
- Blockage of CSF resorbtion
- Increased CSF production
- *Pseudotumor cerebri*
- Primary
- Secondary
- Watch for false localizing lateral rectus palsy

Pseudotumor Cerebri in Children

- Primary: no identifiable cause
- Secondary:

Neurologic disease

Dural venous thrombosis, meningitis, meningioencephalitis, AVM

Systemic disease

SLE, Addison disease, anemia

Ingestions or withdrawals of exogenous substances

Malnutrition refeeding
Steroid withdrawal
Vitamin A
Tetracycline
Thyroxine
Nalidixic acid

Optic neuritis

- Different than optic neuritis in adults
- Usually bilateral (although unilateral cases may not be noted)
- 50/50 male/female
- Progression to MS unusual
- Usually anterior
- Rarely ischemic
- Infectious/Post Infectious:
  - Measles, mumps, chicken pox, rubella, pertusis, mononucleosis, cat scratch, Lyme, TB, syphilis,toxocara, toxoplasmosis
- Acute disseminated encephalomyelitis (post infectious encephalomyelitis): Diffusely distributed CNS demyelinization process, may include optic neuritis
- Neuromyelitis optica (Devic's disease**):** simultaneous or serial inflammation and demyelination of the optic nerve (optic neuritis) and the spinal cord (myelitis)
- Present with profound LOV over 1 – 2 days
- W/U: MRI brain/orbit, with and without contrast
- LP: R/O meningitis, neoplasm
- CXR
- Admit to hospital
- High dose IV steroid x 3 days then oral taper
- Commonly show complete recovery of VA over 2 weeks

Optic atrophy

- Compressive intracranial lesions
- Compressive bony disorders
- Craniosynostosis
- Fibrous dysplasia
- Hydrocephalus
- Post-papilledema optic atrophy
- Hereditary
- Leber hereditary optic neuropathy
- Dominant optic atrophy (Kjer)
- Recessive optic atrophy
- Behr optic atrophy
- DIDMOAD (Wolfram) optic atrophy
- Toxic/nutritional optic neuropathy
- Neurodegenerative disorders with optic atrophy
  - Krabbe disease
  - Canavan disease
  - Leigh disease
  - MELAS
  - Neonatal adrenoleukodystrophy
  - Metachromic leukodystrophy
  - Riley-Day syndrome
  - Lactic acidosis
  - Spinocerebellar degeneration
  - Mucopolysaccaridosis
- Ocular disorders
  - Glaucoma
  - Retinal disease : ROP, RP, RP(+)
  - Vascular disease
  - Uveitis
- Optic nerve hypoplasia
- Hypoxia
- Trauma
- Post-optic neuritis
- Radiation optic neuropathy
- Paraneoplastic syndromes
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Optic Atrophy Evaluation

History

- Detailed history of present illness
- LOV starting when?
- LOV stable or progressive?
- LOV unilateral/bilateral?
- Any other eye symptoms/signs?
- Prenatal, birth and postnatal history
- Growth and development
- Any neurologic regressions?
- ROS
- Skin
- Hearing

- Neuro
- Family ocular history
- Consanguinity?

Exam

- VA, VF, color, red desaturation, pupils, motility, iris, optic disk appearance, retina

W/U:

- ERG for suspected RP or RP(+)
- Peds Neurology
- Peds Genetics
- MRI brain/orbit, with and without contrast
- Laboratory as indicated by exam