Pediatric Optic Neuropathies

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Nothing to disclose
Pediatric Optic Neuropathies

- Pediatric Optic Neuritis
- Optic Atrophy
  - Compressive
  - Degenerative
  - Hereditary
  - Trauma
  - Infiltrative
- Optic Disc Anomalies
Optic Neuritis

- **Childhood Optic Neuritis**
  - Visual loss occurring over days
    - Severe
    - Bilateral in many cases (44-77%)
  - Ocular pain 40%
  - Headache 40%
  - Disc swelling 53-74%
  - Macular star (neuroretinitis) 10%
Case

Fundus Pictures
Case

MRI
Optic Neuritis

Childhood Optic Neuritis

- Associations
  - Febrile or Viral illnesses
    - Measles, mumps, varicella, mononucleosis, Lyme, URI, GI
  - Immunization
  - Systemic disorders
    - Lupus
Optic Neuritis

Childhood Optic Neuritis

- Management
  - Observation
    - Mild unilateral visual loss
  - High-dose IV steroids (methylprednisolone 15 mg/kg q/4 for 3 days, followed by prednisone, 1 mg/kg, tapered over 2 weeks)
    - Bilateral visual loss to any degree
    - Severe unilateral visual loss

- Visual prognosis
  - In most cases, excellent
Optic Neuritis

Childhood Optic Neuritis

– Relationship to Multiple Sclerosis

• Riikonen R (1988)
  – 8/14 (57%) had T2 high signal lesions
  5 of the 8 developed multiple sclerosis (5/21=24% of total)

• Kriss A (1988)
  – 30/39 (77%) childhood cases had no recurrence during a follow-up period of 3 mo to 29 years (mean 8.8 years)
  – 3/39 (8%) had recurrence of optic neuritis alone
  – 6/39 (15%) developed multiple sclerosis

• Lucchinetti CF (1997)
  – 13/79 (13%) with 10 yrs. of follow-up, 26% with 40 yrs. (Kaplan-Meier)
Optic Neuritis

  - 29 patients
    - 8 (28%) boys, 21 (72%) girls
    - Mean age 9.7 years (range: 4.3 - 16 years)
    - 62% ocular pain
    - 38% antecedent febrile or viral illness
    - 39/44 (89%) eyes with decreased visual acuity
    - 27/44 (61%) eyes had disc edema
    - 16/29 (55%) had bilateral optic neuritis
    - 3/29 (10%) had sequential optic neuritis
## Optic Neuritis Visual Acuity
### Adults vs. Children

<table>
<thead>
<tr>
<th>Visual acuity at presentation</th>
<th>ONTT (Adults) N=patients (%)</th>
<th>Pediatric Cohort N=eyes (%) (CHOP)</th>
</tr>
</thead>
<tbody>
<tr>
<td>20/40 or better</td>
<td>162 (35.4)</td>
<td>8 (17.8)</td>
</tr>
<tr>
<td>20/50-20/190</td>
<td>129 (28.2)</td>
<td>9 (20.0)</td>
</tr>
<tr>
<td>20/200 or worse</td>
<td>166 (36.3)</td>
<td>28 (62.2)</td>
</tr>
</tbody>
</table>
Optic Neuritis

  - 29 patients
    - 9/29 (31%) patients had relapses; 5 pts. had more than one relapse
    - 38/44 (86%) eyes had acuity better than 20/40 long term
    - 11/44 (38%) had one or more white matter lesions on baseline MRI
Optic Neuritis

  - 18/29 patients followed for more than 2 years; risk of development of MS studied in relationship to initial MRI findings
    - None of the patients (11/18) with a normal MRI developed MS
    - 7 patients had white matter lesions on MRI
      - 3 of these patients (3/18 = 17%) developed MS (3/7 vs. 0/11, \( p=0.04 \), Fisher’s exact test)
Case 2

- 8 year old daughter of a physician was asymptomatic until she was in her father's office looking at an eye chart. She realized when she covered her right eye, she could not see the chart clearly with her left.
- 20/20 OD, CF 4 ft. OS
- VF
  - Temporal defect OD, Central scotoma OS
- Fundi
Case 2

Left eye:
- Intact to I4e
- Out to V4e
- Out to III4e

Right eye:
- I2e
- III4e
- V4e
Case 2
Chiasmal-Hypothalamic Glioma
Optic Disc Atrophy in Childhood

- Compressive lesions of the pregeniculate visual pathway (optic nerve, chiasm, tract)
  - Optic pathway gliomas
  - Craniopharyngiomas

- Old optic neuropathy
  - History

- Congenital
  - Hereditary (Kjer’s)
    - OPA1
Optic Nerve Glioma
Craniopharyngioma
Leber’s Hereditary Optic Neuropathy
Optic Disc Atrophy in Childhood

- Mitochondrial
  - Leber’s hereditary optic neuropathy
  - Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy, Deafness (DIDMOAD)

- Spinocerebellar ataxias
  - SCA1
  - Friedrich’s ataxia

- Infiltrative

- Trauma
Optic Disc Anomalies

Definition

- Abnormally appearing optic nerve due to some embryologic mishap with resulting malformation

Presentation

- Unilateral
  - Strabismus/Amblyopia
  - Asymptomatic

- Bilateral
  - “My baby can’t see”
  - Nystagmus
Optic Nerve Hypoplasia
Optic Disc Anomalies

- Optic Nerve Hypoplasia
  - Most common disc anomaly
  - Any level of visual acuity; fixed deficit
  - Unilateral or bilateral
  - Ophthalmoscopy
    - Small disc with reduced diameter
    - Peripapillary pigmentary changes
    - Anomalous vessels
    - “Double ring” sign
Optic Nerve Hypoplasia

Double Ring Sign
Optic Nerve Hypoplasia

Double Ring Sign
Optic Nerve Hypoplasia

Double Ring Sign
Optic Disc Anomalies

- Optic Nerve Hypoplasia
  - Septo-optic dysplasia (de Morsier’s syndrome)
    - Optic nerve hypoplasia
    - Absence of the septum pellucidum
    - Also: Dysgenetic or absent corpus callosum
    - Pituitary dwarfism (hypopituitarism)

- Other MRI associations
  - Thin optic nerve
  - Pituitary ectopia
  - Schizencephaly and other cortical migrational abnormalities
Optic Nerve Hypoplasia

Absence of Septum Pellucidum
Thin Chiasm
Optic Nerve Hypoplasia

Agenesis of the Corpus Callosum
Pituitary Ectopia

Schizencephaly
Optic Disc Anomalies

- Optic Nerve Hypoplasia
  - Evaluation in young children
    - MRI of brain and orbits
    - Endocrinology
    - Pediatric ophthalmology
    - Pediatric neurology
Optic Disc Anomalies

铤 Morning Glory Disc Anomaly

– Ophthalmoscopy
  • Funnel shaped excavation
  • Enlargement of the disc
  • Gliosis and elevation of the surface
  • Spoke-like vessels, absent central vasculature
  • Pigment disruption

– Associations
  • Transphenoidal encephalocele
  • Moya-moya vessels