The Pediatric Optic Nerve

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Optic nerve abnormalities in children

- Poor vision and nystagmus / esotropia
- Systemic disease
- Early/accurate diagnosis important

- To present recent relevant information that may aid in the diagnosis of optic nerve abnormalities in children

Case 1

- 3 yo girl referred because of failed vision screening
- VA Allen pictures: 20/30 OD and 20/100 OS
- No strabismus
- + APD OS


Optic nerve hypoplasia

Double ring
How small is too small?

- DD/DM ratio: normal >0.35; hypoplasia <0.30

Small ON $\neq$ hypoplasia

Further work-up?

- A. none, this is unilateral hypoplasia
- B. none, no history of seizures, dev. delay jaundice, or hypoglycemia
- C. MRI brain first. If normal no other work-up necessary
- D. Endocrinology evaluation

Systemic associations

- Isolated (rare)
- Maternal diabetes: segmental superior ONH
- Septo-optic dysplasia (de Morsier Syndrome):
  - absence of the septum pellucidum,
  - pituitary abnormalities, (40-70%)
  - hypoplasia of corpus callosum
  - Other brain malformations (schizencephaly, leukomalacia, encephalomalacia)

MRI of brain

- Reliable predictor of pituitary gland function?
  - Retrospective study* 67 children with ONH
    - Sensitivity 88%
    - Neg Predictive Value $\Rightarrow (D' | \overline{T})$ 93% (PPV 100%)
  - Prospective study ** 47 children
    - 35% w/endocrine abnormalities had MRI abnormalities
    - No association between radiological abnormality and endocrinopathy ($p=1.00$)
  - Normal MRI does not completely rule out an endocrinological abnormality!

*Phillips et al. JAPOS 2011;5:275-80
Endocrinopathies

- Congenital hypopituitarism
  - Most common:
    - growth hormone (70%)
    - hypothyroidism (43%)
    - adrenal insufficiency (27%)
    - diabetes insipidus (5%)
    - multiple deficiencies

Pituitary dysfunction

- ↓GH → Impaired growth (not until age 4/PRL)
- ↓TSH → jaundice, developmental delay
- ↓ACTH → hypoglycemia, hypotension, seizures
- ↓FSH/LH → precocious or delayed puberty
- ↓ADH → Diabetes insipidus

- Test: provocative GH secretion, provocative cortisol secretion, water deprivation test, IGF-I, IGF-B3, T3, T4, TRH, TSH

Unilateral vs. Bilateral ONH

- Bilateral more common (75-85%)

- Similar risk of endocrine abnormality
  (80% bilateral/70% unilateral)

- Bilateral: poor vision/nystagmus
- Unilateral: strabismus, anisometropia
Case 2

- 2 year-old girl, referred for amblyopia OS
- Vision LEA: 20/30 OD, LP OS
- +APD OS
- Microphthalmia OS
- Posterior lens opacity OS with persistent hyaloid artery
- Left ON excavated

Congenital excavations of the ON

- Optic Nerve Coloboma
- Morning Glory Disc Anomaly
- Optic Disc Pit
- Peripapillary staphylomas
- Megalopapilla
- Bilateral Cavitary ON anomalies + Multiple Cilioretinal Vessels (Papillorenal syndrome)
- PVL

Morning Glory Disc Anomaly
Morning glory disc anomaly

MGDA vs. ON Coloboma

- Defect in posterior sclera and lamina cribosa with herniation of neural tissue
- Sporadic
- Unilateral
- Contractile elements
- Persistent hyaloid vasculature
- Risk of serous RD
- Hypertelorism, cleft lip/palate, basal encephalocele, corpus callosum agenesis, Moyamoya, PHACE

- Failure of embryonic fissure to close
- Unilateral or bilateral
- 20% AD, sporadic
- Coexistent ocular malformations
- Risk of serous RD, peripapillary CNV
- CHARGE (CHD7)/branchio-oculo-facial syndrome (TFAP2A)

Coloboma
Moyamoya Disease

MGDA Systemic associations

- Transsphenoidal basal encephalocele (herniation of sella contents into sphenoidal sinus)
- Moyamoya Disease (CNS vasculopathy with progressive stenosis of the internal carotids)
- MRI/MRA or CTA

Optic Nerve Pit

Scott RM, Smith ER. Moya Moya Disease. NEJM 2009;360:1226

Herniation of dysplastic retina into collagen pocket

6 year old male w/ GH deficiency
Both parents had normal exam
Optic nerve swelling

12 year-old with diplopia, HA, nausea, vomiting
VA 20/30 OD, 20/20 OS
15° ET/ 6° ET'

Brain MRI

Papilledema

14 year-old girl with syncope, blurry vision, HA, vomiting
VA 20/30 OD, 20/50 OS
Case # 3

- 22 month-old male
- Referred for intermittent crossing x 1 month
- PMH: Mild anemia. No recent ear infections, no fever, nausea, vomiting or headache.
- Meds: iron

- Variable angle L ET 0-30°
- Full EOM (no abduction deficit)
- No APD
- CRet: +4.00+0.50x90 OU
Lumbar puncture

- Under moderate sedation (propofol/sevo)
- Side w/ legs extended
- Opening pressure was 22 cm H₂O

- Normal OP in children → >20 cm H₂O

CSF opening pressure in children with optic nerve head edema

- Matched case-control study (N=41/group)
- Range of OP in children with/without ONHE

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<tr>
<th></th>
<th>ONHE</th>
<th>No ONHE</th>
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<tbody>
<tr>
<td>Mean</td>
<td>41.4</td>
<td>18</td>
</tr>
<tr>
<td>Range</td>
<td>22-56</td>
<td>9-29</td>
</tr>
<tr>
<td>&gt;28 cm H₂O</td>
<td>40</td>
<td>2</td>
</tr>
<tr>
<td>&lt;28 cm H₂O</td>
<td>1</td>
<td>39</td>
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NPV=97.5%  PPV=95.3%

Reference Range for Cerebrospinal Fluid Opening Pressure in Children

- Prospective study of 197 children 1-18 years (without papilledema)
- 11.5-28 cm H₂O (10-90th percentile)
- Moderate-deep sedation- higher OP

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<thead>
<tr>
<th>No Sedation</th>
<th>Minimal Sedation</th>
<th>Moderate to Deep Sedation</th>
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<tbody>
<tr>
<td>Frequency</td>
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A

B

97.6% >28 cm H₂O

2 patients >28 cm H₂O
• Both parents examined.
• Mother had dysplastic appearance of the left ON.

Conclusions

• Important to recognize subtle characteristics of the different pediatric optic nerve abnormalities
• Dictate work-up/ diagnose systemic disease
• Avoid unnecessary tests
• Guide treatment
• Complete eye exam, knowledge PMH/ROS, examination of both parents → Dx.
• Ancillary testing: ON photos, OCT, B-scan, MRI/MRA

Thank you!
Optic Nerve Head Drusen

OCT & Optic Nerve Head Drusen

From Differentiating Optic Disc Edema From Optic Nerve Head Drusen on Optical Coherence Tomography
Lenworth N. Johnson, MD; Meredith L. Diehl, MD; Chuck W. Hamm, COT, CRA, OCT C; Drew N. Sommerville, MD; Gregory F. Petroski, PhD
OCT papilledema

OCT drusen

Son-mNLF, papilledema, Drusen?
MNFL-mother

MRI other signs
Empty Sella