Morning Report

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Presentation – 37 day old F

- ED visit with unresponsiveness, hypothermia, respiratory failure
- Per mother – intermittent dilated and fixed pupils
- Constipation x3 days
- Consulted for possible NAT
• PMH:
  • Born at 40 wks gestation
  • Normal delivery
  • Discharged home in 2 days
  • Meeting milestones
  • No surgical history

• SHx:
  • Lives with parents
  • FOC on suboxone (buprenorphine/naloxone, treatment for opioid dependence)

• FHx:
  • negative
Exam

• VA sc: BTL both
• Pupils: 5 -> 4 mm both, sluggish, but no APD
• IOP: STP both
• EOM: full, no strabismus
• Penlight exam:
  – L/L: normal both
  – C/S: white and quiet both
  – K: clear both
  – AC: deep and quiet both
  – Iris: normal both
  – Lens: lens clear both
DFE

• BOTH: anomalous ONH with PPA, hypopigmented fundus, slightly enlarged venules, no hemorrhages
Differential

• Optic nerve hypoplasia
• De Morsier’s syndrome/septo-optic dysplasia
• NAT
• Optic Nerve Coloboma
• Morning Glory Optic nerve
• Hereditary Optic neuropathy (Kjer’s, Behr’s)
Workup

• Imaging
• Labs
No evidence of fractures or hemorrhage. ? Absent septum pellucidum
Marked bilateral hypoplasia of optic nerves, chiasm, optic tracts. Absent septum pellucidum; absent posterior pituitary bright spot.
• **Labs:**
  • Negative Urine toxic screen
  • Negative sepsis workup
  • Endocrine workup
    – BG 80s
    – normal electrolytes,
    – normal TSH, FT4
    – low IGF-1

• **Endocrine Consult**
9 days after presentation

- 42 days old
- Brought to ED with RLE edema/erythema – fractured femur
- Eye exam unchanged (no hemorrhages)
- Bone scan: bilateral humeral fractures, right femur fracture
bilateral humeral metaphyseal fractures, right femur fracture
Optic Nerve Hypoplasia (ONH)

- Decreased number of axons
- Pale or gray appearance
- Unilateral or bilateral
- +/- double ring sign
  - Outer ring corresponds to normal junction between sclera and lamina cribrosa
  - Inner ring – abnormal extension of the retina and pigment epithelium over the outer portion of the lamina cribrosa.
- +/- vessel tortuosity
- ON injury prior to complete development
  - Possibly with prenatal/perinatal injury
• VA:
  – One of the top 3 leading causes of childhood blindness
  – Normal to LP
  – Depends on integrity of macular fibers and often does not correlate with size of the disc
• VF: localized defects, constriction
• If bilateral: congenital sensory nystagmus
• If unilateral: frequently sensory strabismus
  – If vision loss is present may be 2/2 amblyopia
• Associations:
  – Maternal ingestion of phenytoin, quinine, cocaine, LSD, smoking
  – Fetal alcohol syndrome
  – Maternal type 1 DM
  – Young maternal age
  – First parity
  – Prematurity
Septo-optic dysplasia (SOD)/De Morsier syndrome

- Reeves (1941), De Morisier (1956)
- Triad: Midline CNS anomalies; unilateral (12%) or bilateral (88%) ONH; hypopituitarism
  - 30% will have all three at presentation
- Absence of septum pellucidum and/or corpus callosum
• Incidence 8-10/100,000
• 75-80% have ONH – presenting feature
• Homozygous mutations in HESX1 (uncommon)
• M = F
• Sporadic or familial (AD or AR)
• Variable phenotype
  – Short stature, obesity, sleep-wake cycle inversion
• Require endocrine workup
• Structural pituitary abnormalities – 15%
  – Absent posterior pituitary bright spot
  – Absent pituitary infundibulum
  – Hypoplastic anterior pit
  – Ectopic posterior pit

• Risk for hypothalamic and pituitary dysfunction
  – Growth hormone (58%), hyperprolactinemia (73%), hypothyroidism (39%), panhypopituitarism (62%), diabetes insipidus, adrenal insufficiency
• Cerebral hemisphere abnormalities (45%)
  – Schizencephaly
  – Periventricular leukomalacia
  – Encephalomalacia
  – Associated with neurodevelopmental defects

• Neonatal jaundice associated with hypothyroidism
• Neonatal hypoglycemia or seizures associated with panhypopituitarism
• Diabetes insipidus associated with thermal regulation problems
Reliability of Magnetic Resonance Imaging for the Detection of Hypopituitarism in Children with Optic Nerve Hypoplasia

Raghu H. Ramakrishnaiah, MD, Julie B. Shelton, MD, Charles M. Glasier, MD, Paul H. Phillips, MD

• Cross-sectional study
• 101 children with ONH
• Radiologists masked to funduscopic findings and endocrine status
• MRI was sensitive (96%) and specific (92%)

• Other recent studies (Ahmad et al, and Garcia-Filion et al)–significantly lower sensitivity of MRI (only 8/54 patients with hypopituitarism and ONH had MRI findings of pit abnormalities)
• Treatment: multidisciplinary (ophthalmology, neurology, endocrinology)
• Endocrine replacement therapy – lifelong monitoring
• KIGS data on 395 pts with long-term effects of GH substitution with good results
• No improvement of ONH with stem cell therapy
Bibliography


• Pediatric Ophthalmology and Strabismus, AAO, BCSC, Chapter 24, p. 308-9. 2013