Abstract: **Hypothalamic hamartomas are the most common cause of central precocious puberty and can present with visual field defects. Automated visual field testing in children can be completed with good reliability around age 8.**

"Growing Up Too Fast"

I. Case History
- 11yo white male
- First seen 2005, age 6
- CC: Failed pediatrician vision screening
- Adopted, Russia, age 4
- Birth mother – TB
- (-) POHx, PMHx
- NKDA, mutvitamins

II. Pertinent findings
2005 initial exam (age 6y, 1m)
- DVA OD: 20/55
  OS: 20/26
- Stereo: 188sec
- Wet Ret OD: +1.25-2.50x170
  OS: +1.25-1.00x040
- DCT: Ortho, NCT: 4xp
- EOMs, color, CS, OH: wnl
- Assessment: meridional amblyopia
- Plan: full time spectacle wear
  Final SRx OD: +0.75-2.50x170
  OS: +0.75-1.00x040
- 3m, 6mo, 1yr, 2yr f/u: improved VA & stereo, no patching, no Rx change
- DVA OD: 20/25
  OS: 20/26
- Stereo: 73sec
2008 exam (3yr f/u, age 9y)
- Slightly reduced VA OD, good stereo
- DVA OD: 20/28
  OS: 20/22
- Stereo: 60sec
- Updated SRx: OD +1.25-3.25x170
  OS +0.25-1.25x040

*Physical changes*
- Growth spurt (13inches), acne, increased body hair/odor, voice deepens
- Suspect precocious puberty
Medical Work-up (9y, 4m)
III. Differential Diagnosis

- Central precocious puberty (gonadotropin dependent)
- Precocious pseudopuberty (gonadotropin independent)
- Premature pubarche/thelarche (isolated events without other signs of puberty)
- Exogenous androgens (e.g. testosterone patch)

IV. Diagnosis & Discussion

- Diagnosis: Central precocious puberty secondary to hypothalamic hamartoma
- Causes of CPP
  1. Tumors (gliomas)
  2. Hypothalamic hamartoma
  3. Congenital adrenal hyperplasia
  4. CNS injury (inflammation, surgery, trauma)
  5. Congenital anomalies (hydrocephalus)
  6. Idiopathic
- Hypothalamus
  1. <1% of total brain
  2. Regulates sleep/dream, hunger/thirst, body temperature, stress, water balance, survival aggression, reproduction & sexual behavior
  3. GnRH --> LH & FSH from anterior pituitary gland
    a. Males: LH --> testosterone, sperm; FSH --> sperm maturation
    b. Females: FSH --> estrogen, ovulation; LH --> corpus luteum
- Hypothalamic hamartoma (HH):
  a. Hamartoma: “a benign (noncancerous) growth made up of an abnormal mixture of cells and tissues normally found in the area of the body where the growth occurs.”
  b. Non-progressive
  c. Located at base of hypothalamus
- HH Syndrome
  1. Gelastic seizures (laughing seizures)
  2. Developmental delays (motor, speech, cognition)
  3. Behavioral disturbances (rage, poor concentration, autistic-like)
  4. CPP
- Types of HH
  1. Pedunculated
    a. Parahypothalamic
    b. Attach to tuber cinereum
    c. Does not displace hypothalamus
    d. Neuro symptoms rare
    e. CPP common
2. Sessile
   a. Intrahypothalamic
   b. Attach to mammillary bodies
   c. Displaces hypothalamus
   d. Neuro symptoms present
   e. CPP 45%

V. Treatment
   • Medically - CPP w/o neuro symptoms (Patient AC)
     a. GnRH analog
     b. Subcutaneous injection
     c. Lasts 1mo or 3mo, take for several years
     d. Floods GnRH receptors, turns of hypothalamus
     e. Regresses puberty/bone growth, improves final ht outcome
   • Radiotherapy
     a. Small HH
     b. Bilateral mammillary attachment (high risk)
   • Surgery
     a. Large HH
     b. Seizures (excision vs disconnection)
     c. Early CPP Dx (avoid long-term hormone therapy)
   • Surgical complications
     a. Systemic
        1. Hemiparesis/hemiplegia
        2. Short-term memory loss
        3. Hyperthermia
        4. Hypersomnolence
        5. Decreased thyroxine
        6. Decreased growth hormone
        7. Hypernatremia
        8. Increased appetite, weight gain
        9. Stroke
     b. Ocular
        1. CN III palsy (temporary, permanent)
        2. Visual field defects (before and/or after surgery)

VI. Management
   • Baseline HVF 30-2 SITA standard OU
     a. At age 9y, 7m: poor reliability, high false positive/negative, midperipheral depression of sensitivity consistent with untrained pt. No defect consistent with optic chiasm compression.
     b. At age 11y, 1m: good reliability, same midperipheral depression of sensitivity consistent with untrained pt. no scotoma consistent with optic chiasm compression
   • Retino-striate pathway matures during first 2 yrs of life
• Non-physiological factors leads to unreliability
  1. Attention/concentration
  2. Learning effect
  3. Fatigue/boredom
• Use reliability indices & gaze tracker to monitor
• STATPAC: MD, PSD analysis compares against age-based norms
  a. Official normative database starts at age 18
  b. No perimeter has children’s normative database, no plans for one in future from Zeiss.
  c. Children are compared against 18yo, no projected normative age data
• Mean sensitivity: better than comparing against age-based norms
  a. Adult by 11-12yo
• ‘Reliable’ age (study review average): 8yo
• Duration per eye (study review average): 4.6min
• VF Pointers
  1. Pre-training
  2. Patience, encouragement
  3. Take breaks
  4. Shorter test time (SITA fast)
  5. Custom HVF
  6. Child chin rest / saddle seat

VII. Conclusion
• Hypothalamic hamartoma – benign, non-progressive
• HH Syndrome: gelastic seizures, developmental delays, CPP
• HH can involve CN III palsy & VF defects
• No official perimetry normative database for children
• Children can perform visual fields reliably around age 8
• Automated visual fields can be successfully and reliably completed in children and should be attempted when indicated.

References