What is “Infantile Spasms”? Why the Urgency?

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October 24, 2017
What is Infantile Spasms?

- An early, devastating form of epilepsy (disorder characterized by chronic seizures)
- Also known as West Syndrome
  - Described in 1841 by Dr. William West watching the movements in his own 4-month-old son
- IS starts in infancy
  - Peak onset 4-6 months
  - 95% children before the age of two
  - Can very rarely start right at birth
  - Occurs in 1 in 2000-4000 births
Description of Spasms

- "The bowing and relaxing of the head and trunk would be repeated alternatively at intervals of few seconds and from ten to twenty or more times at each attack that would continue not more than two or three minutes ...sometimes two, three or more attacks in the day. The child appeared frightened and screamed.... he neither possessed the intellectual vivacity or the power of holding himself upright or using his limbs, and his head falls without support."
Infantile Spasms
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Days 4 of Seizures
2 min once a day
Diagnosis

- Clinical description of spasms
- EEG with hypsarrhythmia pattern
  - Chaotic background with epileptiform discharges (electric sparks that disrupt normal brainwave patterns and cause seizures)
  - A routine (~30 minute) EEG may not capture a spasm, and may miss hypsarrhythmia because they are not present at all times
  - Video-EEG is needed to ensure capturing spasms and hypsarrhythmia
Hypsarrhythmia versus normal EEG
Causes (Etiology)

- Not one single disease, but a type of epilepsy with multiple causes
- Brain Malformations (Lissencephaly, focal cortical dysplasia)
- Genetic Syndromes (Trisomy 21, ARX, CDKL5, Rett, Aicardi)
- Metabolic Disease (Mitochondrial, amino acidurias)
- Brain injury (hypoxia, trauma, infection)
- Neoplasm
- Neurocutaneous disorders (TS, Sturge Weber)
- Unknown – presumed genetic
Prognosis

- Remission rate (rate of going away) if untreated is 30% based on natural history, before treatments discovered
- Remission rate with treatment is 50 – 58%
- Success depends on age of onset, how quickly treatment is given and cause of spasms
- The question after remission, is “what comes next?”
- 90% of children with spasms will have some degree of developmental abnormalities, including developmental delay, autism, learning disability or intellectual disability
Why the urgency?

- We worry most about a child’s development
- The minute spasms start (and often before), there is developmental stagnation and / or regression
  - This may be why earlier onset has a worse prognosis, because development is disrupted so early
  - Therefore, stopping spasms as soon as possible may allow for better development
- There is some evidence that earlier treatment may lead to better long-term developmental outcomes
References


References
