Case Presentation

LILY TRAN, MD
PEDIATRIC EPILEPTOLOGIST – CHOC CHILDREN’S SPECIALISTS
INFANTILE SPASMS SYMPOSIUM
OCTOBER 25, 2017
Case #1

- 9 month old boy
- CC: Episodes of head dropping & developmental regression
- HPI:
  - Normal development until age of 6 months, then appeared to stop progressing developmentally
  - Over past three months, did not develop any new skills, only sitting unsupported
    - Started crawling at 6 months but then stopped
    - Was able to look at her and track mom, but now only intermittently keeps eye contact
    - Does not recognize faces when looking at a mirror.
    - Started to coo and say mama/dada around 7 months, no longer saying any words.
  - Over last week, increasing brief episodes of head bobbing forward and eyes rolling back, as if baby has lost head control.
  - No associated emesis, loss of consciousness, or recent trauma. No changes in PO intake, voiding, or stooling.
Case #1

- **PMHx:**
  - 8/2016: History of episode diagnosed as ALTE

- **Surgical Hx:**
  - None

- **Birth Hx:**
  - NSVD at term, uncomplicated delivery and nursery stay and went home with mom

- **Development:**
  - By 6 months: cooing, sat unsupported, started to roll over.
  - Past three months has not rolled over, is not able to pull to stand, less social, smiling less, does not respond to name, feeding self less. Used to say "mama" but has not for a few weeks.
IZN: Up-to-date

Family Hx:
- Father - celiac disease, diabetes mellitus
- Mother - Hashimoto's
- Maternal GF - leukemia at age 40
- Older brother with h/o post-infectious demyelinating disorder
- No family history of epilepsy

Social Hx:
- Lives with mother, father, and 3 siblings
Case #1

Exam:
- General exam: Normal
- Neurological exam:
  - Cognition and Attention:
  - Attention: Normal for age.
  - Cranial Nerves: Normal
  - Motor: Normal bulk and tone 5/5 bilateral upper and lower extremities.
  - DTR: 2+ throughout the biceps, bracioradialis, patella, and ankle.
  - Sensation: Intact to light touch throughout with no evidence of extinction.
  - Coordination: No dysmetria with reach for toys or objects.
Case #2

- 8 month old girl with tuberous sclerosis complex

- **CC:** Clusters of seizure-like activity x 1 day

- **HPI:**
  - Brief episodes of shoulders shrugs and bilateral arms extension
  - Occurs 1-3 times in a clusters
  - Episodes not associated with awakening from sleep. No eye deviation.
  - Not babbling as much and not as social
  - Otherwise been well, no fevers/cough/congestion/vomiting/diarrhea/weight loss.
Case #2

- **PMHx:**
  - Tuberous sclerosis work up started when PMD noticed multiple hypopigmented spots on exam. Per mom genetics testing sent and positive for TS. Imaging of brain completed and showed lesion in frontal lobe, renal imaging showed lesion in the kidney as well.

- **Surgical Hx:** None

- **Birth Hx:** Full-Term, NSVD, baby had irregular heart beat in utero, mom on bedrest for incompetent cervix. Otherwise no complications.

- **Developmental Hx:**
  - Sat by 6 months of age. She is currently pulling to stand and cruising. She turns pages in books and eats herself finger foods. She uses both hands equally. She babbles "mama" "dada" and "nana" (grandma). Mom reports that she is not babbling as often as she was prior to onset of spells, but she has not lost any words. Similarly, she was more social prior to onset, in that she laughed, smiled, and was more, but still laughs and smiles.
Case #2

- **IZN**: Received 4 month vaccines, needs 6 month vaccines and flu.

- **Family Hx:**
  - Dad with tuberous sclerosis, otherwise cousin with seizure disorder. No other developmental delay, brain tumors.
  - Seizures: cousin.
  - No Developmental Delay.
  - No Neuromuscular Disease.
  - No Psychiatric Illness.

- **Social Hx:**
  - Lives at home with mom, dad, half sister, niece.
Case #2

Exam:

- General: Normal
- Neurological Exam:
  - Mental Status: Age appropriate.
    - Observed State: Alert, Attentive to environment
    - Speech: babbling during exam.
  - Cranial Nerves: Normal
  - Motor Examination: Normal bulk and tone. Strong and symmetric strength in all four extremities.
  - Coordination: reaches for objects, uses pincer grasp, brings to mouth.
  - Reflexes: DTRs: 2+ throughout Toes: Upgoing
  - Sensory exam: Pain and soft sensation: Intact throughout.