Conflicts of Interest

Nothing to declare
Objectives

• How accurate is our diagnosis of epilepsy in children and what are the consequences of misdiagnosis?

• How do we diagnose and classify epilepsy in children?

• Why is electroclinical syndrome and constellation important?
Accuracy of Diagnosis
12-Year-Old Nicole

- Recurrent episodes of passing out since age 8 yrs
- Semiology: nearly all triggered (anxiety, pain, heat); feels unwell, nauseated, looks pale and falls limply to ground
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Misdiagnosis is Common

- BMJ 2002 – 1/3 of 214 children treated for epilepsy by a British consultant were felt to not have epilepsy: “Such a rate may not be unusual”

- First seizures: 24-35% referred to first seizure clinics had non-epileptic or “disputable” events (van Donselaar et al: 1989; Hamiwka et al: 2007)

- Refractory epilepsy: 39% of children referred to Danish tertiary epilepsy center for refractory epilepsy did not have epilepsy based on video-EEG (Uldall et al: 2006)
Risk of Diagnosing Non-Epileptic Events as Epilepsy

- Failure to treat underlying condition
  - Cardiac
  - GI – Sandifers
  - Psychiatric
  - Other
- Side effects of multiple AEDs
- Unnecessary lifestyle restrictions
- “Stigma” of having epilepsy
3-Month-Old Boy

- 2-month history of spells, diagnosed as colic
- Parents concerned as not using left hand well, poor visual attention and little developmental progress
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Dx: Ohtahara’s Syndrome and Epileptic Spasms
Consequences of Not Diagnosing Real Epilepsy

- Ongoing cognitive sequelae in certain epilepsy syndromes
- Risk of physical injury – falls, bike accidents, burns
- Risk of SUDEP/accidental death – fortunately rare in population-based studies (<1/4000 person-yrs) but more common in intractable, poorly controlled epilepsy
Diagnosis and Classification of Epilepsy
History is Paramount

- **SEMILOGY** – ask the child, parent, any witness
  - DDx of epilepsy from non-epileptic events
  - Seizure type
  - Localization in focal onset seizures
- Triggers, pre-ictal and post-ictal states
- Past history: predisposing factors for epilepsy or non-epileptic events
  - Perinatal problems, CNS infections, febrile seizures, head injury, developmental delay
- Family history
Physical exam

• Head circumference
• Dysmorphic features
• Focal neurological findings
• Neurocutaneous lesions
• Developmental assessment
• General physical exam
  • Cardiac findings
  • Organomegaly
Investigations

EEG

- **Consensus**: indicated in all children with new onset epilepsy or first afebrile seizure but does not make the diagnosis!
- 30-50% show EA on first EEG
- 3-4% without seizures show EA on EEG
- Normal variants (wickets, 14 and 6, sleep etc) often misread as epileptiform

Bloodwork

- No evidence of value but little evidence of no value ([AAN practice parameter](https://www.aan.com/content/aan/sections/practiceparameters/))

Imaging

- MRI is indicated after a first seizure except in idiopathic epilepsies (**Consensus**)
Classification of Epilepsy

- **Mode of onset**
  - Generalized/bihemispheric
  - Focal/unihemispheric
  - Spasms
  - Unknown

- **Etiology**
  - Genetic
  - Structural/Metabolic
  - Unknown

- **Electroclinical syndrome**

- **Constellation** = “surgical syndrome”
Why is Electroclinical Syndrome and Constellation Important?
Two 4-Year-Old Girls, Previously Well

• Case 1:
  – single early morning seizure, choking sound, rhythmic twitching of L face and arm, then secondary generalization; normal exam
  – over next 2 yrs, 3 more spells, but remains normal

• Case 2:
  – single seizure beginning with clonic activity in left face and arm with secondary generalization; exam initially normal;
  – over next 2 years, increasingly frequent left sided seizures and left hemiparesis
Case 1

Diagnosis
• Benign epilepsy of childhood with centrotemporal spikes

Outcome
• If AEDs needed, seizures usually easily controlled
• 100% remission
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Case 2

? Diagnosis

- Rasmussen encephalitis
- Responds poorly to AEDs
- Ultimately will need hemispherectomy
Two 4-Year-Olds with Staring Spells Diagnosed as Absence Seizures

Case 1

- Normal cognition
- 2 month hx of staring spells, clear onset and offset, 40x/d
- No postictal phase
Two 4-Year-Olds with Staring Spells Diagnosed as Absence Seizures

Case 2

- Well until 6 mo earlier
- Staring spells have less clear onset and offset
- Also has twitching events, sometimes which can lead to falls
- Occasional GTCS
Two 4-Year-Old Children with Absence Seizures

Case 1

Dx: childhood absence epilepsy

Case 2

Dx: myoclonic atonic epilepsy
Electroclinical Syndrome/Constellation

- Consider age at onset, seizure type(s), presumed etiology, EEG features, other

- How common can these be identified?
  Wirrell et al: 2011
  - Electroclinical syndrome identified in 29%
  - Constellation identified in 3%

- Provides clearer information
  - Response to therapy/choice of best therapy
  - Likelihood of remission
  - Developmental outcome
Conclusions
Diagnosis of Pediatric Epilepsy

• History is paramount – there are no “confirmatory” tests

• Non-epileptic paroxysmal events are commonly misdiagnosed as seizures

• EEG can provide additional information but does not make the diagnosis
  • Normal variants misinterpreted as “abnormal”
  • Spikes ≠ epilepsy (3-4% of normal kids have spikes on EEG)
Conclusions
Diagnosis of Pediatric Epilepsy

• Epilepsies should be classified into mode of onset, etiology, and where possible electroclinical syndrome or constellation
  • Guides decisions regarding best treatment option
  • Provides clearer picture of natural history and long-term outcome