Specific Aims

- How do we define epilepsy?
- Do seizures equal epilepsy?
  - What are seizures?
- Seizure medications, “does one size fit all?”
  - Does my son/daughter have to take medication?
- Why do we want to perform electroencephalograms (EEG), brain scans (Magnetic Resonance Imaging or MRI), and ask you so many questions when we see you?
- Is there more to epilepsy than just seizures?
Definition of Epilepsy

- Epilepsy is defined as two or more unprovoked events (seizures) that are produced by abnormal synchronization of cortical neurons that result in a change in perception or behavior.
Definition of Epilepsy

Potential Diagnosis of Epilepsy

Breath holding, syncope, Sleep paroxysmal events

Seizures

Provoked

Yes

Febrile convulsion, symptomatic

No

EPILEPSY

Solitary seizure
Seizures are repetitive events that are produced by abnormal synchronized cortical discharges.

- Most patients have a limited repertoire of events.

Distinct from:
- Tics
- Involuntary movements (i.e. dystonia)
- Behaviors (i.e. day dreaming)
- Sleep paroxysmal events (i.e. sleep walking)
- Tremors
- Typical migraine headaches
Epidemiology

- If you live long enough, approximately 3% of the population will have epilepsy (Cumulative Incidence).
- The highest frequency of afebrile seizures is during the first years of life and at the end of life.
Seizures

- Seizures can begin:
  - In one part of the brain—Focal onset or focal seizures (partial seizures)
    - EEG demonstrate a focal onset.
    - The patient stopped and stared: partial complex seizure.

Patient, suddenly stopped and stared.
Seizures

- Seizures can begin from multiple sites in the brain.
- When seizures begin on both sides of the brain at the same time, we call these generalized seizures.
  - The EEG demonstrates a generalized onset.
  - The patient stopped and stared—a generalized seizure.

Doctor, she just stopped
And stared.
Seizures

- Did you pick up how the two seizures were described on the EEG and by the parent?
  - The EEG showed a focal seizure and generalized seizure, yet these were described the same.
    - Stopped their behavior and stared.
  - Does the difference matter?
Medications

- What are the decisions to be considered before starting medication therapy?
  - Risks of further seizures outweigh the risks of treatment.
    - From seizures themselves
      - Injury to self
      - Injury to others
      - Psychosocial consequences
  - The effectiveness of medications to prevent recurrences.
    - Almost no child should be treated with medication after the first seizures.
      - The overall recurrence rate within 2 years is about 40% - 50%.
      - After the second seizure, the rate increases to 80% - 90%.
Medications

• So, when we think we have to treat seizures:
  • What are our choices?
  • Why do we make the choices we do?
  • What happens when everything fails?

• What is your response to what we use to treat your child?
  • Does it matter to us?
  • How do we need to work together to get things right?
Medications: Choices

- ACTH*
- Carbamazepine*
- Clobazam
- Ethosuximide*
- Felbamate
- Gabapentin
- Lamotrigine
- Lacosamide
- Levetiracetam
- Oxcarbazepine
- Phenobarbital*
- Pregabalin
- Primidone*
- Rufinamide
- Stiripentol
- Topiramate
- Valproic Acid*
- Vigabatrin
- Zonisamide
Questions to you: Why?

• The truth: “History”- why we ask such funny questions.
  • We do not see your sons/daughter’s seizures.
  • We rely on how you describe the seizures.
    • Look for clues to see if we can determine if it begin focally or generalized.
      • How did the seizure look when it first began?
      • How long did it last?
      • What did your son/daughter do during the event?
        o One side moved, both sides, rhythmically or just stiffening.
  • We think it makes a difference how we begin treatment.
  • Gives us options of which medications.
Questions to you: Why?

- Family history:
  - Your son/daughter’s seizures may be due to genetics.
  - We are finding that many epilepsies are due to genes.
  - If we find a gene at fault it might give us direction for treatment.
- For example: Dravet syndrome
  - Due to a mutation in the sodium channel.
  - We know this produces a very difficult seizure to control with medications.
    - Valproic acid, Clobazam, Stiripentol
  - We can tell you the development problems to look for and what the future may hold for your son/daughter.
Next Steps

- Remember our two cases, both had a behavioral arrest and staring, but the EEG….
- This is why we usually ask you to get an EEG on your child.
Next Steps

• What does the EEG tell us:
  • Focal versus Generalized
  • Possible epilepsy syndrome.
• Why would knowing the epilepsy syndrome be important?
  • Just as in finding out possible genetics it tell us:
    • What drugs might work.
    • Allows us to give you a prognosis for the future.
    • Allows us to help you with other co-morbidities that are associated with that particular epilepsy syndrome.
Next Steps:

- We will probably ask you to get an MRI scan of your son/daughter. Why?
  - To look for structural causes for the seizures.
  - Again to help us define an epilepsy syndrome.
  - Gives us an expanded view for possible therapy.
Medications

- So, what do we have at this point?
  - History
  - What the seizure looks like
  - EEG
  - MRI
  - Possibly some labs
    - If the history indicates that we might have:
      - Genetic
      - Metabolic
So what are the epilepsy syndromes?

- Neonatal period
  - Benign familial neonatal epilepsy
  - Early myoclonic encephalopathy
  - Ohtahara syndrome
- Infancy
  - Epilepsy of infancy with migrating focal seizures
  - West syndromes
  - Myoclonic epilepsy in infancy (MEI)
  - Benign infantile epilepsy
  - Dravet syndrome
  - Myoclonic encephalopathy in nonprogressive disorders
So what are the epilepsy syndromes?

- Childhood
  - Febrile seizures plus (FS+ can start in infancy)
  - Early onset benign childhood occipital epilepsy (Panayiotopoulos type)
  - Epilepsy with myoclonic atonic seizures
  - Benign childhood epilepsy with centrotemporal spikes
  - Autosomal dominant nocturnal frontal lobe epilepsy
  - Late onset childhood occipital epilepsy (Gastaut type)
  - Epilepsy with myoclonic absences
  - Lennox-Gastaut syndrome
  - Epileptic encephalopathy with continuous spike and wave during sleep
  - Landau Kleffner syndrome
  - Childhood absence epilepsy
So what are the epilepsy syndromes?

- Adolescence-Adult
  - Juvenile absence epilepsy
  - Juvenile myoclonic epilepsy
  - Progressive myoclonus epilepsies
  - Autosomal dominant epilepsy with auditory features
  - Other familial temporoal lobe epilepsies
  - Epilepsy with generalized tonic-clonic seizures alone
Why is this important?

• Syndromes
  • Literature on:
    • Treatment
    • Prognosis
    • Co-morbidities

• Our aims are to see if your son/daughter fit into a syndrome so can more focussed on treatments and letting you know what lies ahead.
Reality

• Unfortunately, not everyone fits into a syndrome.
• What does this mean?
  • The literature tells us: If no syndrome is known:
    • First medication works about 45% of the time to stop seizures.
    • Second medication works about 25% of the time to stop seizures.
    • Third and beyond medications work about 3 – 5% of the time to stop seizures.
• What if medications do not work?
What Next?

- When medications do not work:
  - Vagus nerve stimulator
  - Ketogenic diet/Modified Atkins Diet
  - Epilepsy surgery
Why should we care?

- Emotional/Psychological aspects:
  - Learning/memory
  - Self-worth
    - Seizures in front of peers
    - I am “less” than my peers because I have seizures
  - Psychological aspects of not knowing when the next seizure will occur.
  - Development of “dependent” relationships
  - SUDEP
To be continued:

- Thanks for listening.
- Questions?