Syndromes
Definitions, Clinical Presentations, and EEG Patterns

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Dravet Syndrome
Severe myclonic epilepsy of infancy (SMEI)

- Also known as severe myclonic epilepsy of infancy (SMEI)
- Appear in the 1\textsuperscript{st} year of life as multiple febrile seizures
- Progresses to other types of seizures including myoclonus, tonic clonic, and status epilepticus
Dravet Syndrome: Cause

- 30-80% of cases believed to be caused by a gene defect
- Most cases the mutations in the sodium channel gene SCN1A
Dravet Syndrome: Treatment

- Antiepileptics: Tegretol, Sabril, Trileptal, Dilantin, fosphenytoin, and lamotrigine are not effective due to interaction with the sodium ion channel.
- VNS
- IVIG Therapy
- Ketogenic Diet
- Marijuana
Dravet Syndrome: Prognosis

- Cognitive impair directly correlates with seizure frequency
- Teenages tend to be dependant on caregivers
4 YOF w sz since 3 months, regression, staring episodes, drop attacks, GTC, twitches
Progression of seizure
Note the slowing and attenuation
Kleine Levin Syndrome
Sleeping Beauty Syndrome

- Primarily affects adolescent boys 2:1
- Hypersomnia
- Hypersexual
- Compulsive behaviors
- Mood disorders/irritability
- Increase food intake
- Derealization, hallucination and delusion

- Cognitive disturbances
- Median age 15 years old
- 16.9 ± 8.5 years
- 81% of onset occurred in second decade of life

Picture courtesy of http://cartoonesia.com
Kleine Levin Syndrome: Cause

- Precipitated by infection 2:3
- Head trauma 9%
- Alcohol consumption 5.4%
- Marijuana use
- Physical exertion/ psychological distress
- Surgery
- Lactation/menses
- Possible link among infection, head trauma and alcohol consumption is an increase in blood brain barrier permeability
Kleine Levin Syndrome: Prognosis

- Median duration of episodes 10 days
- Median length between episodes 3.5 months
- Duration median 8 years
- Mean of 4 new cases reported per year
- A complete recovery and good prognosis
- Resolves on its own
Kleine Levin Syndrome: EEG patterns

- 70% Non-specific diffuse slowing of background activity
- Less often, low frequency high amplitude delta or theta wave occurred in isolation or sequences in the bilateral temporal or temporofrontal areas
- Short REM sleep latency in two case
- Proportional increase in sleep stages
- Decrease sleep efficiency
- EEG, brain flow SPECT, or neuropathological data showed that frontal, temporal, less common the occipital and parietal lobes can be involved as well as the thalamus
11 YOF W altered mental status
Note the bilateral frontotemporal delta
Sturge Weber Syndrome (SWS)

- Port wine stain
- Developmental delay/mental retardation
- Learning problems
- Attention deficit-hyperactivity disorder
- Macrocephaly
- Ocular manifestations

- Soft-tissue hypertrophy
- Hemiparesis
- Visual loss
- Hemianopsia

Sturge Weber: Cause

- The etiology of SWS is unclear
- Some correlation with the inversion of chromosome arm 4q and trisomy 10
Sturge Weber: Treatment

- Seizure control
- Aspirin therapy
- Early surgical treatment may prevent neurological deterioration
Factors predicting a poor outcome (or indicating the need for surgery) in SWS include the following:

- Early seizure onset
- Extensive leptomeningeal angioma (LA)
- Medically refractive seizures
- Relapsing or permanent motor deficits
- Headaches or mild trauma associated with transient motor deficits
- Evidence of progressive neurologic damage
Sturge Weber: Prognosis

- Increasing seizure frequency and duration
- Increasing duration of postictal deficits
- Increasing focal or diffuse atrophy
- Progressive atrophy or calcifications
- Development of hemiparesis
- Deterioration in cognitive functioning (loss of intellectual abilities)
Sturge Weber: EEG patterns

- Focal seizures with subsequent generalization
- Discharges seen from area under port wine stain
- Seizures seen in 72-93%
2 YOF w left sided SWS
Note left side attenuation and slowing
Angelman Syndrome

- Complex genetic disorder
- Developmental delay (by 6-12 months of age)
- Speech impairment
- Ataxia
- Epilepsy
- Microcephaly
- Happy/excited demeanor
- Fair skinned/light hair
- Hyperactive
- Difficulty sleeping
- Scoliosis
- 1 in 12,000 to 20,000
Angelman Syndrome: Cause

- Maternal chromosome 15 containing this gene is deleted (70%)
- Mutation in the maternal copy of the UBE3A gene (11%)
- Inherits two copies of chromosome 15 from father
- Unknown cause (10%-15%)

Picture courtesy of http://www.cureangelman.org
Angelman Syndrome: Treatment

- Anti-seizure medication
- Physical therapy
- Communication therapy
- Behavior therapy
Angelman Syndrome: Prognosis

- Live a normal life span
- May become less excitable with age
Angelman Syndrome: EEG Pattern

- Seizures often begin when a child is between 2 and 3 years old
- Slowing of the background
- IMSD
8 YOM w hx of Angelman’s being seen for episodes of yelling out and chewing

Note: 3 hz spike and wave; multifocal sharps at baseline
Rett Syndrome

- There are four stages
- Hypotonia, difficulty feeding, jerky limb movements, compulsive hand wringing/washing movements, loss of speech, autistic like behaviors, grinding teeth, apnea
- Neurological disorder affecting almost exclusively girls with normal early development, loss of purposeful hand movements, slow brain and head growth, ataxia, seizures, intellectual disabilities

Picture courteous http://diseasepictures.com/rett-syndrome/
Rett Syndrome: Cause

- Occurs in 1 in 10,000-15,000 live births
- Mutation of the MECP2 gene found on the X chromosome
- It is possible but unlikely to be a asymptomatic carrier
- If the mutated gene has dominate expression over the normal gene, the more severe and the earlier the onset
Rett Syndrome: Cause

- Boys that have the MECP2 defect lack a second X chromosome to compensate for the mutation. Therefore boys have severe problems and die shortly after birth not having time to develop clinical signs of Rett’s
Rett Syndrome: Treatment

- No cure for Rett’s; only treat symptoms
- Medication for seizures, breathing problems, and motor difficulties
- Occupational therapy
- Physical and hydrotherapy for prolonged mobility
Rett Syndrome: Prognosis

- Prognosis is difficult to determine due to rarity
- Cases reports of patients living into 40’s and 50’s
6 YOF w Rett’s
Bifrontal discharges
13 YOF w Rett’s
Intermixed theta and delta
EMG that correlates with hand movements
Lennox Gastaut

- severe form of epilepsy that presents by the age of 4
- developmental delay
- intellectual, behavioral and information processing disturbances
- Seizures may include tonic, atonic, atypical absence, myoclonic

Picture courteous
Lennox Gastaut: Cause

- Brain malformation
- Prenatal aphixia
- Severe head injury
- CNS infection
- Inherent degenerative condition
- Metabolic conditions
- 30-35% undetermined
Lennox Gastaut: Treatment

- Clobazam
- Antiepileptics such as valproate, lamotrigine, felbamate, or topiramate
Lennox Gastaut: Prognosis

- Varies
- No cure
- Complete recovery and normal development very unlikely
- Death possible if status epilepticus not resolved
Lennox Gastaut: EEG Pattern

- Multiple seizure types
- Status epilepticus
11 YOM w Lennox Gastaut
Note the chaotic background at baseline
Same patient with sensitivity at 15uV/mm
Seizure in a Lennox Gastaut
GPFA (generalized paroxysmal fast activity)
References

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