Common Intellectual & Psychiatric Developmental Disabilities Associated with CYE

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Objectives

• Define intellectual disability, developmental disabilities and their relationship with Epilepsy
• Discuss common Epilepsy Syndromes associated with ID, DD – incidence, causes, and treatments
• Explore psychiatric comorbidities associated with epilepsy in children and adolescence
Famous Quote

“The human spirit is stronger than anything that can happen to it.”

C.C. Scott
Definitions

• **Intellectual disability** - characterized by significant limitations both in **intellectual functioning** (reasoning, learning, problem solving) and in **adaptive behavior** (everyday social & ADL skills that originates before the age of 18. Usually relates to cognitive-thought processes. Examples include PDD, Autism & ASD

• **Developmental disability** - severe, chronic can be cognitive or physical or both, disabilities appear before the age of 22, lifelong. Some DD’s are largely physical issues, such as cerebral palsy or epilepsy. Some individuals may have a condition that includes a physical and intellectual disability, for example Down syndrome

• Because intellectual and other developmental disabilities often co-occur, intellectual disability professionals often work with people who have both types of disabilities

Definitions from AAIDD (American Association of Intellectual and developmental Disabilities)
Spectrum of Epilepsy

• Can range from frequent seizures with intellectual developmental disability to full seizure control with no cognitive or developmental deficits

• Most CWE fall within the normal range of IQ, but more have scores in the low-normal range than in the general population

• Cognitive difficulties can be subtle or transient and therefore not easily recognized

• However in some situations, frequent seizures or epileptiform discharges result in substantial cognitive decline in children. The term epileptic encephalopathy frequently is used to describe these conditions.
Epilepsy Risk Factors increasing risk of Learning & Intellectual Disabilities

- frequent/poorly-controlled seizures
- high number of lifetime tonic clonic seizures
- status epilepticus
- early onset of seizures
- epilepsy caused by perinatal insult
- focal cortical dysplasia
- structural/metabolic (symptomatic) epilepsies, and temporal lobe (particularly left) epilepsy
Epilepsy Syndromes & Comorbidities

Epilepsies typically associated with intellectual/developmental disability & significant learning problems:

• Epileptic Encephalopathies
  – Infantile spasms (West Syndrome)
  – Lennox-Gastaut syndrome
  – Landau-Kleffner syndrome and CSWS
  – Dravet Syndrome
• Rasmussen’s syndrome
• Tuberous sclerosis
• Progressive myoclonic epilepsy
• Many others
Infantile Spasms (West Syndrome)

• Typically begins between 3-8 months of age, (all before 12 months), and stops by age 4
• Rare occurrence 1/3000 roughly
• Contributing factors = conditions affecting brain development (genetics, body metabolism, brain malformations, anoxia, brain malformations or injury
• Dx by hx of symptomology, physical and neurological exam
• MRI, Blood and urine tests
• EEG shows interictal hypsarrhythmia pattern
West Syndrome continue

- Seizures consists of a sudden stiffening of the body, arms and legs and head bends forward (jackknife effect). Lasting 2-1 seconds, occurring in clusters, occurring most common upon waking, rarely during sleep.
- TX = Steroid therapy (ACTH) by injection, or prednisone PO and the AED medicine (vigabatrin). Problem potential SE is permanent retinal damage, loss of peripheral damage.
- Other AEDs = Depakote® (valproate), Topamax® (topiramate), pyridoxine (vitamin B6), Zonegran® (zonisamide), Onfi® (clobazam) or Klonopin® (clonazepam). (Early dx and Treatment are vital)
- Adjunctive therapies (with limited response to AEDS) = Ketogenic Diet, Epilepsy Surgery with causative brain malformations.
- Most children have developmental disabilities later in life.
- Many children develop other kinds of epilepsy (1/5 develop LGS), some develop autism (ranges 9-35%)
Lennox-Gastaut Syndrome

- Incidence – 26/100,000 persons, 1/10 with seizures beginning in the first 5 years of their life have LGS, more common in boys then girls
- Seizures usually begin 26 – 28 months of age, usually prior to age
- Children with ID and DD have a greater chance of being dx with LGS
- Diagnosis can be difficult because course of symptoms can take years to develop
- Causes = 25% children have no known cause (cryptogenic, but initial research hint toward gene abnormalities) 75% are symptomatic causes like brain injury due to brain infections (encephalitis, meningitis), tuberous sclerosis, brain malformations, injury at birth (causing decreased blood and oxygen to the brain), trauma, and injuries to the frontal lobes of the brain.
- Lifelong, persistent from childhood, to adolescence to adulthood
LGS Diagnostics

• 3 key diagnostic features
  – Multiple seizure types (tonic, atonic-drop, atypical absence, generalized tonic-clonic) starting in childhood and developing over time
  – Complex characteristic EEG patterns while awake, during REM & non-REM sleep, and during seizures (focal to generalized, as well as slower and irregular brain waves throughout the brain)
  – Cognitive Impairment, behavioral abnormalities, and developmental delay (majority develop after seizure occurrence and get progressively worse with age)

Additional factors adding to DD, and ID in LGS include frequent seizures, and needed use of polytherapy AED side effects such as disturbances in mood, alertness, memory etc.
LGS Treatment

- Difficult, usually refractory to AED treatment, 2 or more meds required to treat multiple seizure types, partial seizure relief obtained by valproic acid, lamotrigine, topiramate, felbamate, clonazepam, rufinamide, clobazam and occasionally other medications.
- Use of rescue meds crucial to prevent clustering &/or status
- Diet therapies (Ketogenic, Modified Atkins, LGI)
- Corpus callosotomy only when diet or VSN is not successful in decreasing frequency of drop seizures and subsequent injuries
- VNS most frequently implanted device for LGS clients (not RNS)
Family Factors in LGS

• LGS care requires 24 hr, 7 days a week involvement putting strain on family and marriage relationships (parents of LGS children have a 75% divorce rate)
• Respite care - needed to prevent caregiver burnout, referral to social workers and case managers is a necessity to aide in finding placement
• Education – early evaluation for is needed for IEP accommodations (behavioral plans, medical plans as well as educational goal placement) and rehabilitation services (OT, PT, ST). Neuropsychiatric evaluation is vital
• Transitional resources and education needed – Licensed Social workers vital in providing and finding resources for adult day care, work programs, assisted living, group home and guardianship issues

(17% of adults with cognitive problems who live in supported facilities and are unable to live independently have LGS.)
Dravet Syndrome

- Rare genetic epileptic encephalopathy (not inherited but a new gene mutation developed in infancy). Gene mutations affect sodium ion channels involved in brain cell function.
- Occurs within the first year of life in healthy infants.
- Incidence rates vary from 1 in 20,000 - 40,000.
- 3-8% children having their 1st seizure prior to 12 months of age have increased risk of Dravet Diagnosis.
- Risk factors- seizures lasting more than 10 minutes, seizures occurring on one side of the body, seizures triggered by warm water baths prior to 12 months of age.
Dravet Syndrome Seizure Types

- Occur within the first year of life, occurring with fever, are usually tonic-clonic convulsive, or myoclonic on one side of the body
- Myoclonic seizures develop between ages 1-5 in 85% of Dravet clients
- Many types of seizures can develop (myoclonic, tonic-clonic, absence, atypical absence, atonic, partial and non-convulsive status epilepticus)
- Seizure triggers include illness, fever, external temperature changes, stress, excitement and photosensitivity
- Seizures are usually prolonged &/or repetitive and development of status epilepticus (seizures > 30 minutes) is an increased risk
- Seizures are usually refractory to routine AED treatment
Developmental and Other Issues Associated with Dravet

include:

- Low motor tone – can lead to painful foot problems
- Unsteady walking
- Older children and adults may develop a crouched gait
- Chronic infections
- Low humoral immunity
- Growth and nutrition problems
- Problems with the autonomic nervous system
- Behavioral or developmental problems such as autism spectrum disorder
Dravet Syndrome Treatment

• Early diagnosis provides better outcomes
• Seizures are usually resistant to medication treatment. Goal of AED treatment provide best seizure control possible, improve developmental delays & decrease mortality risk from status or SUDEP
• Increased seizure occurrence and status increases severity of DD, Use of seizure plan and appropriate use of rescue medications is essential
• Poly-AED therapy to treat multiple seizure types
• Avoid sodium channel blockers AEDS (phenytoin (Dilantin), fosphenytoin (Cerebyx, Prodidilantin), carbamazepine (Tegretol), oxcarbazapine (Trileptal), lamotrigine (Lamictal) and rufinamide (Banzel). Avoid other AEDS: vigabatrin (Sabril), and tiagabine (Gabatril) they increase the frequency of myoclonic seizures
• Adjunct treatments that may be helpful include ketogenic diet and VNS (surgery is usually not helpful)
Autism & Epilepsy

- Epilepsy is more common in individuals with autism than the general population (estimates include 20-30%).
- Some estimate that more than 15% of people with epilepsy have autism traits such as impaired social interactions, communication impairments.
- Studies have showed that epileptic seizures short-circuit the neurological function in the brain that affects socialization and involves the same traits as seen in autism – impairment of normal social interaction (eye contact, conversation, enjoying the act of sharing with someone else) and tightly regimented or repetitive cycles of behavior.
- Some researchers believe that epilepsy medication might ease some symptoms of autism and PWE can benefit from autism therapies that focus on improving socialization skills.
ADD/ADHD

- Classified as a mental behavioral disorder
- Defined as "a persistent pattern of inattention and/or hyperactivity-impulsivity that interferes with functioning or development".
- Incidence (Cleveland Clinic) = 3-7% of general youth population versus 12-57% of children diagnosed with epilepsy
- Diagnosed by psychiatric examination
- AEDS side effect of attention problems = topiramate, phenobarbital, benzodiazepines, tiagabine and zonisamide
- If suspected causative factor may need to change AED or decrease dosage
- Treatment includes psycho-therapy and ADHD medications, Neuropsychiatric evaluation for school IEP qualification and accommodation
- Behavioral Disorders associated with ADHD = intermittent explosive disorder (IED) and oppositional defiant disorder (ODD)
- Untreated ADHD = school performance issues = self esteem and socialization problems = increase risk of depression
- ADHD have a higher rate of sleep problems. Patients with electrical status epilepticus of sleep (ESES) have more problems with attention and hyperactivity. Patients with nocturnal frontal lobe epilepsy also can have more problems with inattention and sleep
Psychiatric Co-morbidities and Childhood Epilepsy

2007 survey: 977 of 91,605 reported epilepsy/seizures

- Children with epilepsy/seizures
  - Depression (8 vs 2%)
  - Anxiety (17 vs 3%)
  - ADHD (23 vs 6%)
  - Conduct problems (16 vs 3%)
  - DD (51 vs 3%)
  - ASD (16 vs 1%)

Depression & Epilepsy

• Depression is under-recognized in youth with epilepsy

• Depression prevalence in youth with epilepsy is up to 30% (11% in General Population)

• Some mistakenly view depression as an expected result of the negative impact of seizures on youth

• Depression may more negatively affect quality of life more than epilepsy
Depression & Epilepsy

- Depression not linked to seizure type or severity in youth
- Associated with negative attitude toward epilepsy and problems in family
- Depression is a risk factor for developing epilepsy
- Studies show children and adults 1.7X more likely to have pre-seizure depression

Treatment includes use of SSDIs and CBT
S&S of Depression & Epilepsy

• Some of the important symptoms observed in children with depression:
  Problems falling asleep or staying asleep, sleeping too much, changes in appetite, greater sensitivity to rejection, poor self-esteem, **hopeless thoughts, thoughts of suicide**
  
• Reported 3.5-5.8 X higher risk of death by suicide in PWE

• Higher risk of suicide with temporal lobe epilepsy

• 2008 FDA issued a black box warning about the increased risk for thoughts of suicide in the clinical trials of 11 AEDS.

• Early DX, acceptance, and TX are vital, psycho-therapy, SSDIs, CBT
Conclusion

• Cognitive difficulties are common in children with epilepsy, some can be subtle or transient however others associated with epilepsy syndromes are progressive and associated with DD &ID

• Diagnosis and Treatment can be difficult and complicated with multiple IEDs, and adjunctive therapies

• Psychological co-morbidities are common in CWE

• Nurses and SW are an important key in early recognition, referral to epilepsy specialists, important psycho-social supports and educational resources

• Epilepsy Foundation of Michigan can help
I'm not disabled, I'm Differently Able!

Montel Williams
Case Study

• Case A six year old boy with normal premorbid development, seizures occurring since the age of 18 months. Seizure hx included generalized tonic-clonic convulsion, several episodes in a day; over a period of five years he developed multiple types of seizure: (myoclonic jerks, drop attacks and absence seizures). He had been treated without success with multiple anti-epileptic drugs (phenobarbitone, phenytoin, carbamazepine and valproate). At the time of presentation, he was having 7-8 episodes of seizure a day and was on valproate 25mg/kg, carbamazepine 6mg/kg and clonazepam 0.08 mg/kg.

• General and Systemic exams were normal, including MRI and metabolic profile

• Child walks with a stooping gait with short steps,

• Teachers and parent are noting that the child is having an inability to keep up with classmates and severe problems with inattention and acting out.

• Routine EEG showed sharp slow waves