Idiopathic epilepsy syndromes

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Classification

- 1964
- 1970
- 1981
- 1989
- 2001
- 2006
- 2010
- 2013
- 2016

Identify of epilepsy syndromes based on:
- Typical age onset
- Seizure types
- Specific EEG characteristics
- Other features or related symptoms
- Implications for treatment, management, and prognosis

Definition of idiopathic epilepsy syndromes
- A syndrome that is only epilepsy, with no underlying structural brain lesion or other neurological signs or symptoms. These are presumed to be genetic and are usually age-dependent.

Idiopathic generalized epilepsy
- CAE
- JAE
- JME
- GTCSA
Childhood absence epilepsy

**Clinical**: Onset 2-12 yrs (peak 5-6), both sexes. Antecedent and birth history is normal. 15-20% of cases – History of febrile convulsion, development, cognitive and neuro-exam-normal

**Seizures**: Absence, frequent (multiple daily), brief (~ about 10 seconds), awareness and responsiveness impaired. No another types

**Genetic**: 10% GLUT1 def. SLC2A1, GABRG2 and CACNA1A

**Caution**: Seizures>45 seconds=>focal dyscognitive seizures, GCS before adolescence=>other epilepsy syndrome (JME)

CAE & EEG Findings: 3 Hz spike waves

**Background**: Normal, OIRDA (Occipital intermittent rhythmic delta activity)

**Interictal**: Generalized spikes and waves often becomes fragments with sleep deprivation or in sleep

**Activation**: Provoked by hyperventilation, may not seen if poorly performed

**Caution**: Hyperventilation is performed well for three minutes, no GSW=>CAE is unlikely

CAE: EEG 3 Hz spike waves

CAE: Related family epilepsy syndromes

- Juvenile absence epilepsy
- Juvenile myoclonic epilepsy
- GEFS+ (Generalized epilepsy with febrile seizure plus): rare

**Gene**: Complex inheritance, linked to GABRG2 and CACNA1A

**NOTE**: Absence onset <4 y/o=> GLUT1 deficiency, age onset>8 y/o, infrequent sz=> JAE

Juvenile absence epilepsy

**Clinical**: Age onset 8-20 y/o (peak 9-13), absence seizure not very frequent (if onset 8-12y/o, DDX CAE by frequency of absence seizure)

- Adolescents may present with generalized convulsive seizures prior to onset of absence
- Development and cognitive and neuro-exam-usually normal

**Seizures**: Absence (not frequent, not severe, awareness), GCs (80% of cases, upon awakening-within 30 minutes of waking)

**Genetic**: GABRG2, CACNA1A and others

**Treatment**: Required for life

JAE: EEG 3-6 Hz GSW/PSW

**Background**: Normal, OIRDA may be seen

**Interictal**: GSW, fragments of GSW or PSW

**Ictal**:

- Absence: Regular 3-6 Hz GSW or PSW
- GCs: EEG obscure by artifact, generalized fast rhythmic spikes-tonic phase, after coming slow waves are synchronous with clonic jerks and postictal period of irregular slow activity follows generalized convulsions
**Juvenile myoclonic epilepsy**
- **Clinical**: age onset 8-25 y/o, myoclonic seizures and generalized convulsions, 5% of cases evolve from CAE, 5-10% from febrile seizures.
- **Seizures**: myoclonic (mandatory), especially on awakening (within 30min-1hr of wakening).
- May have GEs (>90% of individuals) preceded by series of myoclonic, absence (1/3 of cases, briefer<3 seconds).
- **Genetic**: may be complex or Mendelian CACNB4, GABRA1, CLCN2, GABRD and EFHC1. Microdeletions, such as the 15q13.3 microdeletion and others.
- **Exclusion**: if other types of seizures.

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**Epilepsy with generalized tonic clonic seizure alone**
- **Clinical**: age onset 15-40 y/o, peak 11-23, infrequent generalized convulsive seizures from the second decade of life, typically provoked by sleep deprivation, previous history of childhood absence epilepsy.
- **Developmental, cognitive and neuro-exam**: normal.
- **Seizures**: GEs especially on awakening (within 1-2 hrs of wakening).
- **Genetic**: complex inheritance, CLCN2 and others.
- **Exclusion**: all other types of seizures.

**GTCSA**: EEG GSW/PSW
- **Background**: Normal (no generalized slowing).
- **Interictal**: GSW/PSW usually at 3.5-6 Hz, fragments can appear focal or multifocal but not consistently seen in one area.
- **Activation**: hyperventilation may provoked absence, <10% sz induced by visual stimuli.
- **Ictal**: single generalized PSW correlates with myoclonic seizures.

**Idiopathic focal epilepsy**
- **BECTS**
- **PS**
- **COE-G**

**JME**: EEG 3.5-6 Hz GSW/PSW
- **Background**: Normal (no generalized slowing).
- **Interictal**: GSW/PSW usually at 3.5-6 Hz, fragments can appear focal or multifocal but not consistently seen in one area.
- **Activation**: hyperventilation may provoke absence, <10% sz induced by visual stimuli.
- **Ictal**: single generalized PSW correlates with myoclonic seizures.

**GTCSA**: EEG GSW/PSW
- **Background**: Normal (no generalized slowing).
- **Interictal**: GSW/PSW (1/3 of cases seen only during sleep).
- **Activation**: PT may provoked intermittent photoparoxysmal response, GSW often becomes fragmented with sleep deprivation or in sleep.
- **Ictal**: GEs: ictal EEG patterns.
- **Note**: No generalized slowing or SSWC.
BENIGN CHILDHOOD EPILEPSY WITH CENTROTEMPORAL SPIKES

- Benign focal epilepsy of childhood (BFEC), Rolandic epilepsy (BRE)
- **Clinical**: age onset 3-14 y/o (peak 8-9), hemifacial seizures that may secondarily generalize if occur nocturnally, 5-15% of febrile seizures
- Prior history unremarkable, Development and cognitive prior onset of seizure is normal
- **Genetic**: Complex inheritance, GRIN2A gene
- Self-limited, usually resolved by age 13 years (occasionally occur up to age 18 years)

BEfT: EEG CENTROTEMPORAL SPIKES

- **Background**: normal
- **Interictal**: High amplitude centrotemporal spikes or sharp-and-slow wave complexes, maximum negativity in C3/C4 and T3/T4 and maximum positivity frontally, increased during drowsiness and sleep, unilateral or bilateral
- May be seen focal spikes outside CT region (midline, parietal, frontal and occipital)
- **Activation**: marked increase in drowsiness and sleep and wider field and may be bilateral synchronous, after 10 y/o may be photoresponsive, 10-20% by sensory stimuli of fingers or toes
- Ictal: rare to obtained ictal recording

Late onset childhood occipital epilepsy (Gastaut type)

- Self-limiting
- **Clinical**: age onset 5 months and 19 years (peak 8-9), prior history: normal, usually easily controlled (50-60% remission in 2-4 years after onset)
- 90% dramatic response to carbamazepine
- **Seizures**: seizures with visual aura occur from awake states, brief (typical seconds, most <3 minutes, rarely up to 20 minutes)
- **Genetic**: unknown
Late onset childhood occipital epilepsy (Gastaut type)

- **Visual aura**: Multi-colored circles in peripheral vision increased involved and moving horizontally to the other side, these may be followed by deviation of eyes or head turning (ipsilateral)
- **Other occipital features**: Ictal blindness, complex visual hallucinations, visual illusions, orbital pain, eyelid fluttering or repetitive eye closure, ictal headache or N/V
- **May spread outside the occipital lobe** resulting in hemiparesthesia, dyscognitive features, hemiclonic

**COE-G: EEG Occipital Spikes**

- **Background**: Normal
- **Interictal**: Occipital spikes or spike-and-wave (may only occur during sleep), 20% of cases may co-exist with CT, frontal or GSW
- **Activation**: By sleep deprivation and by sleep, 20-90% of cases – induced by fixation-off sensitivity
- **Ictal**: During oculo-clonic seizure or ictal blindness: BG activity reduction and then occipital faster rhythms with spikes of low amplitude; these may be slower SW

Panayiotopoulos syndrome

- **Clinical**: Age onset 1-14 years (peak 3-6), autonomic seizure (polyautonomic SE), infrequent
- **Prior and birth history**: Normal
- **Developmental and exam**: Normal
- **5-17% history of FS
- **Seizures**: Autonomic features mainly emetic (nausea, retching, vomiting), pupillary (mydriasis), circulatory (pallor, cyanosis), heart and respiratory change. Apnea and asystole can occur (severe case). Prolong duration, but without residual neuro deficit, some of case- fronto-parietal opercular
- **Genetic**: Unknown gene, complex (report in sibling)
- **Self-limiting, resolve by age 11-13 years

Panayiotopoulos syndrome

- **Background**: Normal
- **Interictal**: Multifocal SPK/SW 90%, 10% normal single EEG, occipital spikes seen on EEG in 60% of patients. Low voltage spikes and generalized discharges may be seen in a minority of cases
- **Activation**: Eye closure (elimination of central vision and fixation-off sensitivity) may activate occipital spikes; EEG abnormality is enhanced by sleep deprivation and by sleep
- **Ictal**: Unilateral, often having posterior onset, with rhythmic slow (theta or delta) activity intermixed with small spikes

Panayiotopoulos syndrome

- **PS: EEG Multifocal Spikes**

Example of multifocal spikes in Panayiotopoulos syndrome

- **PS: EEG Occipital Spikes**

Example of occipital (left) spikes of high amplitude in Panayiotopoulos syndrome