Idiopathic epilepsy syndromes

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Classification

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

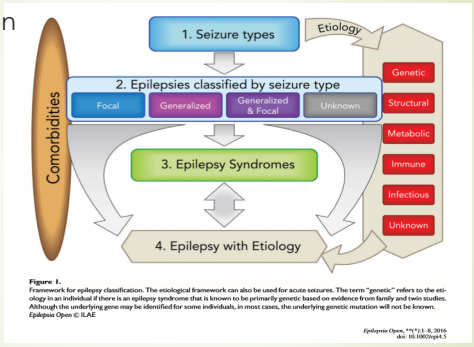

Figure 1.
Framework for epilepsy classification. The etiological framework can also be used for acute seizures. The term "genetic" refers to the etiology in an individual if there is an epilepsy syndrome that is known to be primarily genetic based on evidence from family and twin studies. Although the underlying gene may be identified for some individuals, in most cases, the underlying genetic mutation will not be known.
Epilepsia Open © ILAE

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doi: 10.1002/epi4.5

Identify of epilepsy syndromes based on:

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- Typical age onset
- Seizure types
- specific EEG characteristics
- Other features or related symptoms
- **Implications for** treatment, management, and prognosis

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Definition of idiopathic epilepsy syndromes

- A syndromic 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.

Epilepsy syndrome

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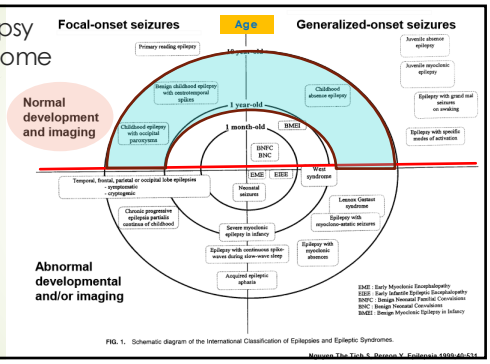

FIG. 1. Schematic diagram of the International Classification of Epilepsies and Epileptic Syndromes.

Idiopathic generalized epilepsy

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- CAE
- JAE
- JME
- GTCSA

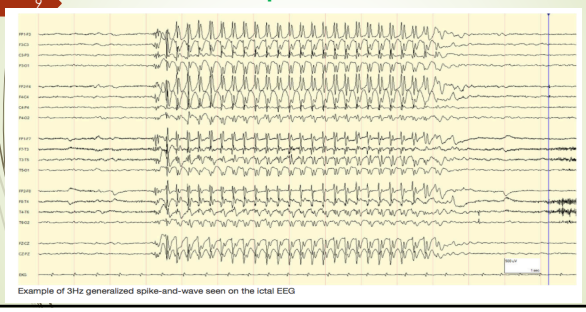
7 Childhood absence epilepsy

- **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
- Self-limited
- **Genetic**: 10% GLU-T1 def. SLC2A1, GABRG2 and CACNA1A
- Caution: seizures>45 seconds=>focal dyscognitive seizures, GC before adolescence=>other epilepsy syndrome (JME)

8 CAE& EEG Findings: 3 Hz spike waves

- Backgrounds: 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

9 CAE: EEG 3 Hz spike waves



Example of 3Hz generalized spike-and-wave seen on the ictal EEG

10 CAE: related family epilepsy syndromes

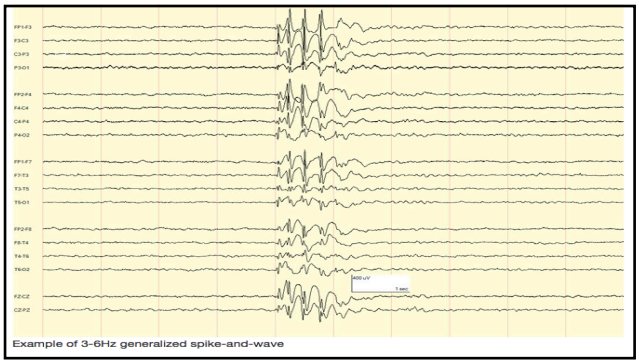
- Juvenile absence epilepsy
- Juvenile myoclonic epilepsy
- GEFS+ (Generalized epilepsy with febrile seizure plus): rare
- Gene: complex inheritance, linked to GABRG2 and CACNA1A
- **NOTE** : absence onset <4 y/o=>GLUT1 deficiency, age onset>8 y/o, infrequent sz=> JAE

11 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

12 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, spike and after coming slow waves are synchronous with clonic jerks and postictal period of irregular slow activity follows generalized convulsions


Example of 3-6Hz generalized spike-and-wave

Juvenile myoclonic epilepsy

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- **Clinical**: age onset 8-25 y/o myoclonic seizures and generalized convulsions, 5% of cases evolve from CAE, 5-10% -febrile seizures
  Developmental, cognitive and neuro-exam-normal
- **Seizures**: myoclonic (mandatory), especially on awakening (within 30min-1hr of wakening)
- May have GCs (>90%of individuals) preceded by series of myoclonic , absence (1/3 of cases, briefer<3 seconds)
- **Genetic**: may be complex or MendelianCACNB4, GABRA1, CLCN2, GABRD and EFHC1, Microdeletions, such as the 15q13.3 microdeletion and others
- Exclusion: if other types of sz

JME: EEG 3.5-6 Hz GSW/PSW

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- 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

Epilepsy with generalized tonic clonic seizure alone

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- **Clinical**: age onset 5-40 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**: GCs especially on awakening (within 1-2 hrs of wakening)
- **Genetic**: complex inheritance, CLCN2 and others.
- Exclusion: all other types of sz

GTCSA: EEG GSW/PSW

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- Background: Normal (no generalized slowing)
- Interictal: GSW/PSW (½ of cases seen only during sleep)
- Activation: PT may provoked intermittent photoparoxysmal response, GSW often becomes fragmented with sleep deprivation or in sleep
- Ictal: GCs: Ictal EEG patterns
- Note: No generalized slowing or SSWC

Idiopathic focal epilepsy

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- BECTS
- PS
- COE-G

BENIGN CHILDHOOD EPILEPSY WITH CENTROTEMPORAL SPIKES

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- 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

BENIGN CHILDHOOD EPILEPSY WITH CENTROTEMPORAL SPIKES

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- **Seizures**: seizure with fronto-parietal opercular features – hemifacial (lip,mouth and tongue),clonic movements (with may be unilateral), laryngeal symptoms, articular difficulty (aphasia), swallowing or chewing movements and hyper-salivation, brief (<5 minutes),
- Few in number of seizure, may have secondarily generalize (typically nocturnal events)
- Exclusionary: GCs during wakefulness

BECTS: EEG CENTROTEMPORAL SPK

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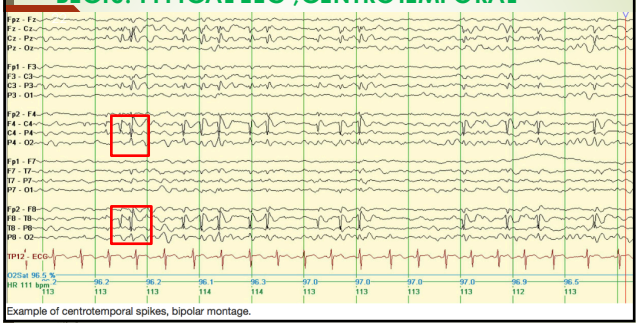
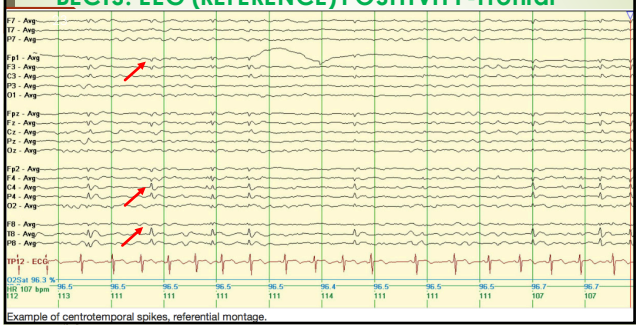
- Background: normal
- Interictal: High amplitude centrotemporal spikes or sharp-and-slow wave complexes, maximum negativity in CT (C3/C4 and T3/T4) and maximum positivity frontally, increased during drowsiness and sleep, unilateral or bilateral
- May be seen focal spikes outside CT region (midline, parietal, frontal and occipital)
- Activation: marked increase in drowsiness and sleep and wilder 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

BECTS: TYPICAL EEG ;CENTROTEMPORAL



Example of centrotemporal spikes, bipolar montage.

BECTS: EEG (REFERENCE) POSITIVITY-Frontal



Example of centrotemporal spikes, referential montage.

Late onset childhood occipital epilepsy(Gastaut type

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- Self-limiting
- **Clinical**: age onset 5 months and 19years (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 20minutes)
- **Genetic**: unknown

Late onset childhood occipital epilepsy (gastaut type)

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- 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)
- May 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

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- 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 (elimination of central vision)
- 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

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- **Clinical**: age onset 1-14 years (peak 3-6), autonomic seizure (25%may autonomic 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

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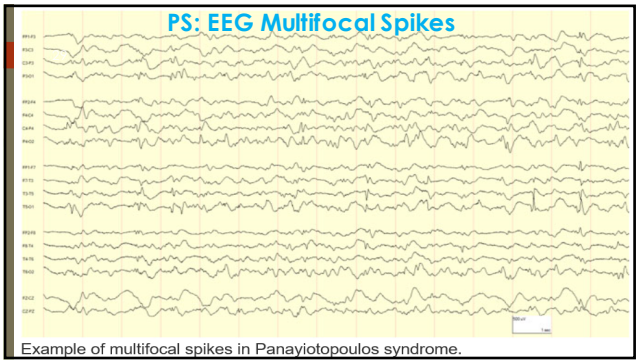
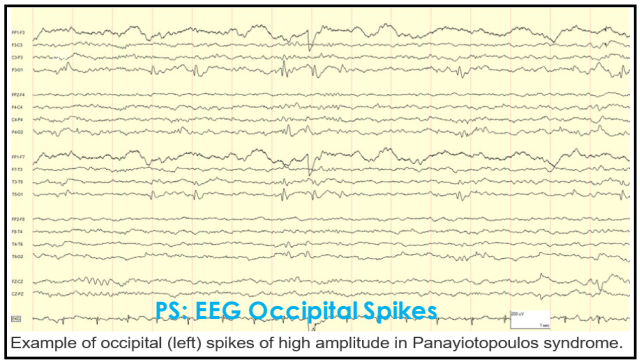
- Background: normal
- Interictal: Multifocal SPK/SW 90%, 10% normal single EEG, occipital spikes seen on EEG in 60% of patients. Low voltage spikes and generalized discharges may be seen in a minority of cases.
- Activation: Eye closure (elimination of central vision and fixation off sensitivity) may 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

PS: EEG Multifocal Spikes



Example of multifocal spikes in Panayiotopoulos syndrome.



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