Idiopathic epilepsy syndromes

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Epilepsy course
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Classification

- 1964
- 1970
- 1981
- 1989
- 2001
- 2006
- 2010
- 2013
- 2017

Seizure types
- Focal onset
- Generalized onset
- Unknown onset

Epilepsy types
- Focal
- Generalized
- Generalized & Focal

Non epileptic events
Semiology & EEG Finding

Epilepsy Syndromes
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

No approved ILAE epilepsy syndromes

https://www.epilepsydiagnosis.org
Easy guide for Epilepsy syndrome
“Pattern diagnosis”

Seizures
Epilepsy

Age of onset
Development
Seizure types
IEDs

Epilepsy syndrome

Epilepsy syndrome

Focal-onset seizures

Generalized-onset seizures

Age

1 month-old
1 year-old
6 years-old

Normal development and imaging

Abnormal developmental
and/or imaging

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

Idiopathic Epilepsy Syndromes

- Idiopathic/Genetic Generalized Epilepsy
- Idiopathic/Self-Limited Focal Epilepsy
‘Idiopathic/Genetic Generalized Epilepsies’

- Childhood Absence Epilepsy
- Juvenile Absence Epilepsy
- Juvenile Myoclonic Epilepsy
- Generalized Tonic-Clonic Seizures Alone

Childhood absence epilepsy

| Age of onset | 2-12 years (peak 5-6 years) |
| Seizure type | Absence only (multiple daily, brief, LOA) |
| EEG          | IEDS: 3 Hz Generalized spikes and waves; Normal background, OIRDA |
| Tests: Genetic | SLC2A1, GABRG2 and CACNA1A |
Childhood absence epilepsy
Juvenile absence epilepsy

**Age of onset**: 8-20 years (peak 9-13 years)

**Seizure type**
- Absence (not frequent, not severe, awareness)
- GTC at onset -> Absence in *adolescent* GCs (80% of cases, upon awakening)

**EEG**
- IEDS: **3-6 Hz** Generalized spikes/polyspikes and waves, normal background, OIRDA (may)
- Ictal: *Absence*: Regular 3-6 Hz GSW or PSW
- GCs: EEG obscure by artifact, generalized fast rhythmic spikes-tonic phase, spike and slow waves and postictal period slowing

**Tests: Genetic**
- GABRG2, CACNA1A and others
### Juvenile absence epilepsy

**Age of onset**
8-25 years (peak 9-13 years) 5% of cases from CAE

**Seizure type**
- Myoclonic (mandatory), especially on awakening (within 30min-1hr)
- GTCs (>90%) preceded by series of myoclonic
- Absence (1/3 of cases, briefer<3 seconds)

**EEG**
- IEDS: 3.5-6 Hz GSW/P SW, normal background, fragments
- Hyperventilation may provoked absence, <10% sz induced by visual stimuli
- Ictal: single generalized PSW correlates with myoclonic seizures

**Tests: Genetic**
- Complex or Mendelian CACNB4, GABRA1, CLCN2, GABRD
- EFHC1, Microdeletions, such as the 15q13.3 microdeletion and others
### Epilepsy with GTCs alone

<table>
<thead>
<tr>
<th>Age of onset</th>
<th>5-40 years (peak 11-23 years)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Seizure type</td>
<td>GCs especially on awakening (within 1-2 hr of wakening) infrequent, typically provoked by sleep deprivation, PH of <em>childhood absence epilepsy</em></td>
</tr>
<tr>
<td>EEG</td>
<td>IEDS: GSW/PSW (½ of cases seen only during sleep) fragmented, intermittent photoparoxysmal response, normal BG (no slowing) Ictal: GCs: ictal EEG patterns</td>
</tr>
</tbody>
</table>

**Tests: Genetic**
- Complex inheritance, CLCN2 and others.

### ‘Self-Limited Focal Epilepsies’

- **Benign Epilepsy with Centro Temporal Spikes (BECTS)**
- **Self-Limited Occipital Epilepsy of Childhood: Panayiotopoulos type (early-onset):PS**
- **COE-G Gastaut type (late onset)**
- **Self-limited frontal/temporal/parietal lobe epilepsies**
### Benign Childhood Epilepsy with Centrotemporal Spikes

<table>
<thead>
<tr>
<th><strong>Age of onset</strong></th>
<th>3-14 years (peak 8-9 years)</th>
</tr>
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</table>
| **Seizure type** | 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 hypersalivation, brief (<5 minutes), Few, (may) secondarily generalize (typically nocturnal events) (not GTC during awake)  
Self-limited usually resolved by age 13 years (occasionally occur up to age 18 years) |

### EEG

| **IEDs:** | High amp. Centrotemporal Spikes or Sharp-and-slow wave complexes, max. negativity in CT (C3/C4 and T3/T4) and max. positivity F, increased during drowsiness and sleep, unilat or bilat, (may) SPK outside CT region (midline, parietal, frontal and occipital), (may) photoresponsive (age.10 yrs), 10-20%-by sensory stimuli of fingers or toes |
| **Tests: Genetic** | Complex inheritance, GRIN2A gene |

Ictal: rare to obtained ictal recording
Benign Childhood Epilepsy with Centrotemporal Spikes
Benign Childhood Epilepsy with Centrotemporal Spikes

Example of EEG in same patient, showing activation in sleep
### Panayiotopoulos Syndrome

| **Age of onset** | 1-14 years (peak 3-6 years)  
| **Self-limiting, resolve by age 11-13 years** |
| **Seizure type** | 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 (25% may autonomic SE), infrequent |
| **Tests: Genetic** | unknown gene, complex (report in sibling) |

### EEG

| **IEDs:** | Multifocal SPK/SW 90%  
| Normal single EEG 10%  
| Occipital spikes 60% of patients  
| Low voltage SPK and Gen d/c 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 posterior onset, with rhythmic slow (theta or delta) activity intermixed with small spikes |
Panayiotopoulos syndrome

Example of multifocal spikes in Panayiotopoulos syndrome.

Panayiotopoulos syndrome

Example of occipital (left) spikes of high amplitude in Panayiotopoulos syndrome.
Late onset childhood occipital epilepsy (Gastaut type)

<table>
<thead>
<tr>
<th>Age of onset</th>
<th>5 months-19 years (peak 8-9 years)</th>
</tr>
</thead>
</table>
| Seizure type | Seizures with **visual aura** occur from awake states, brief (typical seconds, most < 3 minutes, rarely up to 20 minutes **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, dycognitive features, hemiclonic |

Late onset childhood occipital epilepsy (Gastaut type)

| EEG         | **IEDs**: Occipital spikes or SW (may) only during sleep, 20% of cases may co-exist with CT, frontal or GSW, BG normal
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 |
| Tests: Genetic | Unknown |
| Prognosis   | **Self-limiting**
Easily controlled (50-60% remission in 2-4 years after onset) 90% dramatic response to carbamazepine |
### Summary of IGE

<table>
<thead>
<tr>
<th>IGE</th>
<th>CAE</th>
<th>JAE</th>
<th>JME</th>
<th>GTCSA</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age onset</strong></td>
<td>childhood</td>
<td>Juvenile</td>
<td>Juvenile</td>
<td>Juvenile</td>
</tr>
<tr>
<td><strong>Seizure type</strong></td>
<td>Absence</td>
<td>Absence</td>
<td>Myoclonic</td>
<td>GTCs</td>
</tr>
<tr>
<td><strong>EEG</strong></td>
<td>3 Hz GSW</td>
<td>3-6 Hz GSW</td>
<td>3.5-6 Hz GSW</td>
<td>GSW/PSW</td>
</tr>
</tbody>
</table>

### Summary of SFE

<table>
<thead>
<tr>
<th>SFE</th>
<th>PS</th>
<th>BECTS</th>
<th>COE-G</th>
</tr>
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<tbody>
<tr>
<td><strong>Age onset</strong></td>
<td>Infantile 1-14 (3-6) yrs</td>
<td>Childhood</td>
<td>Childhood</td>
</tr>
<tr>
<td><strong>Seizure type</strong></td>
<td>Autonomic (Emetic)</td>
<td>Perisylvian</td>
<td>Occipital</td>
</tr>
<tr>
<td><strong>EEG</strong></td>
<td>Multifocal 90% Occipital 60%</td>
<td>Centrottemporal</td>
<td>Occipital</td>
</tr>
</tbody>
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