Clinical syndromes

Pediatrics Primary Generalized Epileptic Syndromes

Ictal phenomenology

50 30 20 10 0

Age (years)

Incidence of epilepsy

Syndrome Type Def:

- Idiopathic epilepsy syndrome: A syndrome that is only epilepsy, with no underlying structural brain lesion or other neurological signs or symptoms. Etiology presumed to be genetic. Usually age-dependent
- Symptomatic epilepsy syndrome: Epileptic seizures are result of an identifiable structural lesion
- Probably symptomatic epilepsy syndrome: Epileptic seizures are believed to be symptomatic, but no aetiology has been identified
- Benign epilepsy syndrome: Epileptic seizures are easily treated or need no treatment and remit without sequelae

Etiology of epilepsy

PROPOSED DIAGNOSTIC SCHEME FOR PEOPLE WITH EPILEPTIC SEIZURES AND WITH EPILEPSY

Axis 1 - Ictal phenomenology
- detailed description of symptoms during the seizure

Axis 2 - Seizure type or types
- according Ictal phenomenology and EEG

Axis 3 - Syndrome
- list of syndromes, syndromic diagnosing is not always possible

Axis 4 - Etiology
- genetic defect, or specific pathological substrates for symptomatic focal epilepsy

Axis 5 - Impairment
- disability caused by epilepsy
ILAE Classification of Epilepsy

<table>
<thead>
<tr>
<th>Localization-Related (named by location)</th>
<th>Generalized (named by disease)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Idiopathic</td>
<td>Generalized Epilepsies</td>
</tr>
<tr>
<td>Early myoclonic encephalopathy</td>
<td>Benign Neonatal Convulsions (+/– familial)</td>
</tr>
<tr>
<td>Early infantile epileptic encephalopathy</td>
<td>Benign Rolandic epilepsy (Benign childhood absence epilepsy with centro-temporal spikes)</td>
</tr>
<tr>
<td>Cortical abnormalities</td>
<td>Benign occipital epilepsy of childhood</td>
</tr>
<tr>
<td>1. Focal Epilepsies</td>
<td>Idiopathic Focal Epilepsies</td>
</tr>
<tr>
<td>1. Temporal lobe</td>
<td>Temporal lobe</td>
</tr>
<tr>
<td>2. Frontal lobe</td>
<td>Frontal lobe</td>
</tr>
<tr>
<td>3. Parietal lobe</td>
<td>Parietal lobe</td>
</tr>
<tr>
<td>4. Occipital lobe</td>
<td>Occipital lobe</td>
</tr>
<tr>
<td>4.4. Manifestations</td>
<td>Manifestations</td>
</tr>
<tr>
<td>4.4.4. Motor epilepsy</td>
<td>Motor epilepsy</td>
</tr>
<tr>
<td>4.5. Migraine</td>
<td>Migraine</td>
</tr>
<tr>
<td>4.6. Status Epileptic Epilepses</td>
<td>Status Epileptic Epilepses</td>
</tr>
<tr>
<td>4.6.1. Epilepsies</td>
<td>Epilepsies</td>
</tr>
<tr>
<td>4.6.1.1. Absence</td>
<td>Absence</td>
</tr>
<tr>
<td>4.6.1.1. Myoclonus</td>
<td>Myoclonus</td>
</tr>
<tr>
<td>4.6.1.1. Tonic-clonic</td>
<td>Tonic-clonic</td>
</tr>
<tr>
<td>4.6.1.1. Atypical absence</td>
<td>Atypical absence</td>
</tr>
<tr>
<td>4.6.1.1. Tonic, atonic</td>
<td>Tonic, atonic</td>
</tr>
<tr>
<td>4.6.1.1. Tonic-clonic</td>
<td>Tonic-clonic</td>
</tr>
</tbody>
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Diagnostic Features of Focal Epilepsies
- Age of onset - throughout lifetime
- Seizure type – simple/complex partial and/or secondarily generalized tonic-clonic
- Underlying structural etiology may be found and MRI should be performed
- Focal EEG finding may be present, but interictal EEG may be also normal
- Response to “carbamazepine”-like AEDs

Diagnostic Features of Generalized Epilepsies
- Age of onset - late childhood to early adult life
- Seizure type - absence, myoclonus, tonic-clonic
- Lack of underlying structural etiology although microscopical/QMRI changes may be present
- Specific EEG finding (3Hz spike and wave/photosensitivity)
- Genetic basis and positive family history
- Diurnal pattern of seizure occurrence (awakening)
- Excellent response to "valproate"-like AEDs

Idiopathic vs Symptomatic Generalized Epilepsies

<table>
<thead>
<tr>
<th>Idiopathic</th>
<th>Symptomatic</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Etiology</td>
<td>Genetic, Acquired/Genetic</td>
</tr>
<tr>
<td>2. Seizure</td>
<td>Absence, Atypical absence</td>
</tr>
<tr>
<td>3. Exam</td>
<td>Normal, Intellectual disability</td>
</tr>
<tr>
<td>4. EEG</td>
<td>Normal background, Spikes, Spike-wave 3Hz</td>
</tr>
<tr>
<td>5. Imaging</td>
<td>Normal, Often focal or diffuse</td>
</tr>
<tr>
<td>6. Prognosis</td>
<td>Good, Poor</td>
</tr>
</tbody>
</table>

From gene defect to epilepsy phenotype

- Single gene defects
- Mosaic gene defects
- Complex, progressive defects
- Epilepsy phenotypes
- Idiopathic epilepsies
- Partial
- Tonic-clonic
- Symptomatic epilepsies
- Benign
- Progressive neurodegeneration
- Intellectual disability
- Abnormal brain development

Genetically defined epileptic syndromes and specific diseases
- Epilepsies in inborn errors of metabolism
- Progressive myoclonic epilepsies (PME)
- Epilepsies and chromosomal disorders
- Epilepsy and malformations of the cerebral cortex
- New epilepsy syndromes or subsyndromes with single gene inheritance
- Rasmussen’s syndrome
- The mesio-temporal lobe epilepsy syndrome
Generalized Epilepsy with Febrile Seizures plus (GEFS+).
A genetic disorder with heterogeneous clinical phenotypes.

- Family with members with generalized epilepsies of various types and FS
- FS + febrile seizures lasting over age of 6 years
- FS+ absences or myoclonic sz’s or atonic sz’s, myoclonic-astatic epilepsy

GEFS+

<table>
<thead>
<tr>
<th>GENE LOCUS</th>
<th>GENE</th>
<th>GENE PRODUCT</th>
</tr>
</thead>
<tbody>
<tr>
<td>19q13</td>
<td>SCN1B</td>
<td>Voltage-gated Na channel (~ alpha 1 subunit)</td>
</tr>
<tr>
<td>2q24</td>
<td>SCN1A</td>
<td>Voltage-gated Na channel ~ alpha 2 subunit</td>
</tr>
<tr>
<td>2q24</td>
<td>SCN2A</td>
<td>Voltage-gated Na channel ~ gamma 2 subunit</td>
</tr>
<tr>
<td>5q34</td>
<td>GABRG2</td>
<td>GABA receptor</td>
</tr>
</tbody>
</table>

Known idiopathic epilepsy genes

<table>
<thead>
<tr>
<th>Genes</th>
<th>Locus</th>
<th>Type of Mutation</th>
<th>Expression</th>
</tr>
</thead>
<tbody>
<tr>
<td>SCN1A</td>
<td></td>
<td>De novo truncating</td>
<td>CAE and FS</td>
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<tr>
<td>SCN1B</td>
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<td>Inherited missense</td>
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<tr>
<td>SCN2A</td>
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<td>De novo truncating</td>
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<tr>
<td>GABRG2</td>
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Locus Heterogeneity and Variable Expressivity

- GEFS+
- Dravet syndrome (SMEI)
- Benign familial neonatal-infantile seizures (BFNIE)
Benign familial neonatal seizures

Early neonatal absence epilepsy

Ohtahara syndrome

West syndrome/Infantile spasms

Benign neonatal epilepsy

Benign familial infantile seizures

Benign infantile seizures (non-familial)

Dravet’s syndrome (SMEI)

Hemiconvulsion Hemiparesis syndrome

Migrating partial seizures of infancy

Myoclonic status in nonprogressive encephalopathies

Benign childhood epilepsy with centrotemporal spikes (BECTS)

Early onset benign childhood occipital epilepsy (Panayiotopoulos type)

Late onset childhood occipital epilepsy

Startle epilepsy

Benign myoclonus of early infancy (benign nonepileptic Infantile spasms)

- Onset day 2+3, up to 10+20 Sz/day, Autosomal. Dominant Chr. 20+>Fam Hx(+), Stop in 1+6 m/o.
- 10% turn epielptic
- Common up to 5% of FT Sz. Multifocal clonic, can have apnea, status epielpticus. Sz stop in 1 to max. 15 days. Poss. Zn def as etiology.

Benign Familial Neonatal Seizures (BFNS)

- Insidence: 1.4/10 000 live births
- Age of onset: 80% D2-3 (rest up to 3 mth, usually premature)
- Etiology: EBN1 locus 20q13.3 gene KCNQ2
- Seizures: start with a diffuse tonic component, followed by various autonomic and motor (clonic)changes
- EEG: interictal normal
- Therpay: short-term PB or VPA up to 6 months
- Prognosis: favorable, risk of febrile seizures 5%, subsequent epilepsy 11%, especially BECTS, no mental retardation, no neurological abn., no severe epilepsy

Benign Idiopathic Neonatal Seizures (BINS)

- Prevalence: 2-7 % of all neonatal seizures
- Age of onset: 97% D3-D7
- Etiology: idiopathic
- Seizures: clonic partial, and/or apneic, never tonic
- EEG: interictal: normal or bursts of theta rhythms on Rolandic areas
- Therpay: no treatment or short-term AEDs
- Prognosis: favorable, however, probably up to 50% have some abnormalities as child; febrile seizures, other seizures, BECTS, minor neurological impairment

Early infantile epileptic encephalopathy with suppression-bursts (EIEE, Ohtahara’s syndrome)

- Insidence: no data
- Age of onset: within first 3 months
- Etiology: cerebral dysgenesis, anoxia, cryptogenic
- Seizures: tonic-spasms, focal motor, hemiconvulsions, generalized seizures
- Background EEG: suppression burst ** in both awake and sleep
- Ictal EEG: diffuse synchronization, cluster of fast activity
- Therapy: ACHT, B6-vit., VPA, other AEDs, surgery
- Prognosis: static impairment to severe mental retardation, quadriplegia and bed-ridden, evolution to West and Lennox Gastaut syndrome, high incidence of death

Benign familial infantile seizures

- Insidence:
- Age of onset:
- Etiology:
- Seizures:
- EEG:
- Therapy:
- Prognosis:

Ohtahara : Dx criteria

- Aicardiand Ohtahara 2002:
  1. Onset in early infancy, within the first 3 months, mainly within the first 10 days of life
  2. Main seizure pattern: tonic spasms
  3. Other seizures: partial seizures, rare myoclonic seizures
  4. Suppression bursts in EEG, during both waking and sleeping states
  5. Poor prognosis: severe psychomotor retardation and frequent death during infancy
  6. Intractable seizures and frequent progression to West syndrome
  7. Polyetiology, but majority of cases are associated with structural brain damage
Early (neonatal) myoclonic encephalopathy (EME)

Incidence: no data
Age of onset: neonatal
Etiology: inborn errors of metabolism, familial, cryptogenic
Seizures: erratic or fragmentary myoclonus, massive myoclonus, simple partial seizures, infantile spasms, tonic
EEG: suppression-burst in sleep, discharges of slow waves/spikes and fast activity in awake
Therapy: ACTH ineffective, pyridoxine may be tried
Prognosis: progressive impairment to vegetative state, infantile spasms, high mortality in infancy

IEM & EME

• inborn error of metabolism, can produce the clinical and EEG picture typical of early myoclonic encephalopathy.
• nonketotic hyperglycinemia (Several authors)
• DB glyceric acidemia (Grandgeorge et al 1980), propionic acidemia (Vigevano et al 1982; Lombroso 1990)
• molybdenum cofactor deficiency (Aukett et al 1988)
• **methylmalonic acidemia (Lombroso 1990).**
• **Pyridoxine dependency**
• Wang and colleagues reported a patient with a clinical picture of early myoclonic encephalopathy and an atypical suppression-burst pattern, with full recovery after administration of pyridoxine (Wang et al 1998).
• CNS malformation – early myoclonic encephalopathy (Martin et al 1981), but more often they produce Ohtahara syndrome.

severe neonatal epilepsies with suppression-bursts pattern

| NEONATAL EPILEPTIC SYNDROMES CHARACTERIZED BY PERSISTENCE OR APPEARANCE OF BURST/SUPPRESSION BEYOND 3-4 WEEKS OF AGE |
|-----------------|-----------------|-----------------|
| **CLINICAL FEATURES** | **BURST/SUPPRESSION SEIZURES** | **CLINICAL FEATURES** |
| Major clinical signs | Myoclonus and tonic convolution | "Tonic spasms" |
| Major EEG signs | Inborn error of metabolism | Bilateral structural lesions or malformation or destructive focal lesion |
| Major common cause | Inborn error of metabolism | Bilateral structural lesions or malformation or destructive focal lesion |
| Outcome | Variable, usually poor | Poor |

Ohtahara syndrome

• EEG Wu Burst suppression generalized 3-4 weeks EEG seizure, Lt. occipital, Right temporal w/ multiregional sharp waves
• อาการของผู้ป่วยอาจสัมพันธ์กับ 12 วัตถุ 165 B6 จำนวน 100 mg ซึ่งจะช่วยลดตัวเลือกและมีผลิตภัณฑ์ต่างๆที่ไม่ได้ใช้ในสัตว์
• ผู้ป่วยได้รับ vigabatrin 150 mg/kg ทำให้การชักลดลง แต่ผู้ป่วยยังคงมีอาการชัก
• ผู้ป่วยยังคงไม่ดี ยังไม่ดีอยู่ ควรศึกษาโรคที่อาจสัมพันธ์

Ohtahara syndrome
**Epileptic syndromes starting in infancy and early childhood**

- infantile spasms and West syndrome
- migrating partial seizures in infancy
- benign myoclonic epilepsy in infancy
- severe myoclonic epilepsy in infancy (Dravet syndrome)
- myoclonic atonic epilepsy
- Lennox-Gastaut epilepsy
- myoclonic status in non-progressive encephalopathies
- febrile seizures
- idiopathic and/or benign localization-related epilepsies
- non-idiopathic localization-related epilepsies

**Myoclonic atatic epilepsy (MAE, Doose)**

**Prevalence:** 2% of childhood epilepsies

**Age of onset:** between 18 and 60 months (94% under 5 yrs)

**Etiology:** idiopathic, found also in GEFS+-families

**Seizures:** generalized epilepsy syndrome with multiple seizure types including myoclonic-atatic, absences, tonic-clonic, eventually tonic seizures

**EEG:** background normal or 4-7 Hz theta activity, interictaly and with myoclonic jerks and atonic component: bursts of 2-3 Hz generalized (poly)spike-and-wave discharges, tonic component: 10-15 Hz polyspike discharges

**Therapy:** VPA, VPA+LTG, +ESM, +BZD, TPM,LEV

**Prognosis:** Favourable with seizure control in 3 yrs and normal cognitive outcome or unfavourable with resistant epilepsy and cognitive deterioration (if tonic seizures, myoclonic status)

**Severe myoclonic epilepsy in infancy (SMEI, Dravet syndrome)**

**Prevalence:** 7% of pts whose seizures start before age f 3 yrs

**Age of onset:** before age of 1 year (peak 5 months)

**Etiology:** genetic, GEFS+ spectrum (SCN1A-mutation), others

**Seizures:** prolonged, generalized or unilateral clonic seizures triggered by fever, focal myoclonic jerking may precede, later: multiple seizure types: GTCSs,GCSs, myoclonic, atypical absences, complex focal seizures, tonic seizures

**EEG:** paroxysms of generalised polyspikes and slow waves, 2 Hz spike-wave or both, elicited by photic stimulation and facilitated by sleep, background deteriorates progressively

**Therapy:** VPA, BZDs, PB, ESM, TPM, LEV

**Prognosis:** Epilepsy resistant, psychomotor retardation, progressive ataxia and pyramidal symptoms appear within a year from onset (as the result of severe seizures), mortality high (15%)

**Epileptic syndromes starting in childhood**

- epilepsy with centro-temporal spikes
- idiopathic childhood occipital epilepsies
- non idiopathic focal epilepsies of childhood
- the HHE syndrome (hemiconvulsions, hemiplegia, epilepsy)
- electrical status epilepticus during slow sleep (ESES or CSWS) including acquired epilepticus aphasia (Landau-Kleffner)
- childhood absence epilepsy and related syndromes
- the syndrome of myoclonic absences

**Benign epilepsy with centrotemporal spikes (BECTS, Rolandic epilepsy)**

**Prevalence:** 15% of children with seizures (1-15 years)

**Age of onset:** 1 to 14 years, peak at 8-9 years

**Etiology:** idiopathic

**Seizures:** unilateral facial sensorimotor symptoms, oropharyngolaryngeal manifestations, speech arrest and hypersecretion, 1/3-2/3 have nocturnal sGTCs

**EEG:** interictal EEG shows centrotemporal spikes maybe unilateral, but often are bilateral

**Therapy:** not necessarily, AEDs for frequent seizures

**Prognosis:** remission in 2-4 years and before 16 years of age, some have linguistic problems, < 2% epilepsy adults, < 1% evolution to CSWS or Landau Kleffner

**Early onset benign childhood occipital epilepsy (Panayiotopoulos type)**

**Prevalence:** at least 6% of pts with seizures below the age of 13 yrs

**Age of onset:** 3-6 yrs (80%), peak 5 yrs, range 1-13 yrs

**Etiology:** idiopathic

**Seizures:** infrequent autonomic and behavioural disturbances, ictal emesis, ictal syncope, deviation of the eyes, rare visual hallucinations, autonomic status epilepticus, convulsions

**EEG:** variable, occipital/frontal/multifocal spikes, generalized discharges, spikes similar to BECTS

**Therapy:** not necessarily needed, AEDs for frequent seizures

**Prognosis:** remission in 1-2 yrs after onset, 20% may develop other type of infrequent seizures, 13% BECTS, atypical rare cognitive evolutions like in BECTS have been described
### Late onset childhood occipital epilepsies (Gastaut type)

- **Prevalence:** 0.2-0.9% of all epilepsies
- **Age of onset:** 3-16 years, mean 8 years
- **Etiology:** idiopathic
- **Seizures:** frequent visual seizures manifested with elementary visual hallucinations or ictal blindness or both, ocular pain, eye deviation, eyelid fluttering may progress to hemiconvulsions or GTCs
- **EEG:** interictal: occipital paroxysms eyes closed (fixation-off sensitivity), random occipital spikes, ktaxocpital rapid spikes or discharges
- **Therapy:** CBZ (response 90%), other AEDs
- **Prognosis:** remission in 50-60 % in 2-4 years, others continue to have seizures, atypical evolution to CSWS has been described

### Electrical status epilepticus during slow sleep (ESES or CSWS)

- **Insidence:** rare, exact numbers not available
- **Age of onset:** seizures before ESES start at 2mths-12 yrs (peak 4-5 yrs)
- **Etiology:** 30% have preceding encephalopathy, pre-perinatal problems, congenital hemiparesis, rest cryptogenic
- **Seizures:** 1) motor 2) unilateral partial motor seizures or GTCs, absences 3) atypical absences with atomic or tonic szs
- **EEG:** ESES develops 1-2 yrs after seizures: during non-REM sleep continuous bilateral and diffuse slow wave SWs mainly at 1.5-2 Hz persisting through all slow sleep stages
- **Therapy:** seizures VPA, BZDs, ESM, electrographic abnormalities respond poorly to treatment
- **Prognosis:** neuropsychological impairment during ESES, afterwards seizures disappear, EEG improves, 50% cognitively impaired

### Acquired epileptic aphasia (Landau-Kleffner)

- **Insidence:** rare, exact numbers not available
- **Age of onset:** 2-8 years (peak 5-7 years)
- **Etiology:** epileptogenic functional lesion in the speech cortex
- **Seizures:** sz’s present in 70-80%: atypical absences, myoclonic sz, focal sz’s w/ 2° generalization, variable prognosis
- **Clinical:** verbal auditory agnosia ➔→→ acquired aphasia ➔→→→→ behavioural and psychiatric problems
- **EEG:** bilateral symmetrical/asymmetrical multifocal spikes and SW in temporal and parieto-occipital regions, sleep enhances spiking up to CSWS (85% of slow wave sleep)
- **Therapy:** VPA, BZDs, ESM, TPM (steroids, surgery, immunoglobulin)
- **Prognosis:** aphasia usually improves with EEG normalization before adulthood, 10-20 % may achieve complete normalization, others are left with permanent sequelae

### The syndrome of myoclonic absences (EMA, Tassinari syndrome)

- **Prevalence:** 1% of the epilepsies
- **Age of onset:** 11mths-12 yrs
- **Clinical:** 45% mentally retarded before diagnosis, imaging abnormal 17%
- **Seizures:** impairment of consciousness variable from mild to complete loss (absence) and bilateral myoclonias (shoulder,arms,legs)
- **EEG:** GSWDs 3/s
- **Therapy:** VPA, ETS, LTG, BZD, LEV
- **Prognosis:** 70% mentally retarded, 50% seizures persist over the age of 20 yrs

### Childhood absence epilepsy (CAE)

- **Prevalence:** 10-12% of children with epilepsy
- **Age of onset:** between 4 and 10 years, peak at 5-7 years
- **Etiology:** idiopathic (normal neurological state and development)
- **Seizures:** brief (4-20 s) and frequent (tens per day) absence seizures with abrupt and severe impairment (loss) of consciousness. Automatisms are frequent but have no significance in the diagnosis
- **EEG:** ictal discharges of generalized high-amplitude spike and double (maximum occasional three spikes are allowed) spike- and slow-wave complexes, rhythmic at around 3 Hz, duration 4-20 s
- **Therapy:** ESM, VPA, LTG, BZDs, TPM, LEV?
- **Prognosis:** remission in 33-78% at age of 3-19 yrs, risk for later GTCs 36-60%, psychosocial problems in 30%

### Childhood Absence Epilepsy

- **Onset:** 4-8 Y (3-12 Y range)
- **Clinical:** Brief blank staring / impairment in school performance
- **Up to 50 % has one GTC.
- **EEG:** 3 Hz SWC on Normal BG
- **DOC:** Ethosuximide / VPA
  - Use VPA if the patient has other type of seizure
  - Can be followed by EEG with Hyperventilation
Epileptic syndromes starting in older children and adolescence

- complex reflex epilepsies: reading epilepsy and praxis induction
- isolated focal seizures of adolescence
- juvenile absence epilepsy
- juvenile myoclonic epilepsy
- epilepsy with generalized tonic-clonic seizures only

**Juvenile absence epilepsy (JAE)**

**Prevalence:** 2-3% of adults with epilepsy

**Age of onset:** 7-17 years, peak at 10-12 years

**Etiology:** Idiopathic

**Seizures:** Frequency of absence seizures lower and impairment of consciousness less severe than in CAE, manifests in most patients also with infrequent tonic-clonic seizures and sporadic, infrequent myoclonic jerks

**EEG:** Background normal, generalized symmetric SW discharges with frontal accentuation, faster than 3 Hz (3-4.5 Hz)

**Therapy:** VPA, LTG, BZDs, TPM, LEV?

**Prognosis:** Life long disorder, although seizures can be controlled in 70-80%

**Juvenile myoclonus epilepsy (JME, Janz)**

**Incidence:** 5-10% of all epilepsies

**Age of onset:** between 8 to 26 years, mostly between 12 to 18 years

**Etiology:** Idiopathic

**Seizures:**
1) myoclonic jerks (characteristic spontaneous, brief, involuntary, sudden, synchronous and symmetric)
2) typical absence seizures (in one third)
3) tonic-clonic seizures (in majority)

**EEG:** Bilateral polyspike-wave discharges, synchronous and symmetric, include 5-20 spikes, 12 to 16 Hz. Interval between apex of spike and myoclonic jerk in EMG is short 20-50 ms indicating cortical myoclonia

**Therapy:** VPA, LTG, TPM, LEV, BZDs

**Prognosis:** Life long, 15% resistant with increased risk of SUDEP, 30% has psychological or subtle frontal lobe dysfunctions

**Generalized polyspike wave (psw) in JME**

(Janz 1969)
Continuum of 4 epileptic syndromes

Epilepsy with GTCS only

Prevalence: not available
Age of onset: between 6 to 47 years, peak at 16-17 years
Etiology: idiopathic, high incidence of other IGEs
Seizures: includes GTCSs occurring on awakening, diffusely while awake or during sleep or randomly
EEG: generalized discharges of spike/multiple spike slow waves
Therapy: VPA, LTG, TPM, LEV, BZDs, (sodium channel-blockers)
Prognosis: life-long disease with a high (83%) incidence of relapse on withdrawal

EEG features of absence seizures in idiopathic generalized epilepsy: Impact of syndrome, age, and state

*Levente Grant Sander, Ingrid E. Scheffer, Sherry Smith, Benedict Carstens, Kevin Farrell, and Mary B. Connolly

Discussion: The EEG features of absence seizures are influenced by a complex interaction of age, epilepsy syndrome, level of arousal, provoking factors, and other intrinsic factors. Epilepsy syndrome alone cannot predict specific features of GSEW; however, JME is more frequently associated with polypharmacy and disorganization of the paroxysm.
## Seizures

**EEG:** interictal EEG often normal, reading provocation results in other language related activities, rare: visual/oculomotor, dyssynergia, high risk of sGTC

**Therapy:** VPA, CLN

**Prognosis:** drug response good, complete remission rare

## Etiology

**Age of onset:** between 12-25 years

- Dentato(rubro)pallidoluysian atrophy
- Gaucher type III
- MERFF
- onset

## Progressive myoclonus epilepsies (PME)

- Neuronal ceroidlipofuscinoses
- MERFF
- Gaucher type III
- Lafara’s disease
- Unverricht-Lundborg’s disease (EPM1)
- Dentato(rubro)palidoluysian atrophy

## Insistence:

**Age of onset:** between 7-18 years, 86% starts between 9-13 years

**Prevalence:** about 200 patients in Finland

**Incidence:** unknown (especially the silent forms)

**Characteristics:**
- Progressive, many patients need wheelchair at least occasionally, no major cognitive decline

## Autosomal Dominant Nocturnal Frontal Lobe Epilepsy (ADNFLE)

- **Etiology:** autosomal dominant gene that encodes cystatin B (CSTB), a cysteine protease inhibitor
- **Age of onset:** between 7-18 years
- **Prevalence:** about 200 patients in Finland
- **Incidence:** unknown (especially the silent forms)

## Prognosis:

**EEG:** spontaneous spike-wave discharges, photosensitivity and polyspike discharges during REM sleep, the almost continuous, small amplitude jerks are often not time-locked to EEG discharges, only the large-amplitude ones often are

**Therapy:** VPA, LTG, TPM, LEV, piracetam, CLN, CLB, PB

**Prognosis:** variable, but progressive, many patients need wheelchair at least occasionally, no major cognitive decline

## Ring chromosome 20 syndrome (r20S)

- **Insistence:** unknown (especially the silent forms)
- **Age of onset:** seizures usually begin in childhood
- **Etiology:** the formation of the ring is associated with loss of telomeric material on both arms of chromosome 20, the severity of mental retardation correlates with the percentage of the abnormal lymphocytes while epilepsy does not

## Autosomal Dominant Nocturnal Frontal Lobe Epilepsy (ADNFLE)

- **Characteristics:**
  - Generalized tonic-clonic seizures
  - Throwing hyperkinetic activity
  - Secondary GTC

## Progressive myoclonus epilepsy (EPM1, ULD)

- **Prevalence:** about 200 patients in Finland
- **Age of onset:** between 7-18 years
- **Incidence:** unknown (especially the silent forms)
- **Characteristics:**
  - Progressive, many patients need wheelchair at least occasionally, no major cognitive decline

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- **Characteristics:**
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- **Prevalence:** about 200 patients in Finland
- **Age of onset:** between 7-18 years
- **Incidence:** unknown (especially the silent forms)
- **Characteristics:**
  - Progressive, many patients need wheelchair at least occasionally, no major cognitive decline

## Ring chromosome 20 syndrome (r20S)

- **Insistence:** unknown (especially the silent forms)
- **Age of onset:** seizures usually begin in childhood
- **Etiology:** the formation of the ring is associated with loss of telomeric material on both arms of chromosome 20, the severity of mental retardation correlates with the percentage of the abnormal lymphocytes while epilepsy does not

## Autosomal Dominant Nocturnal Frontal Lobe Epilepsy (ADNFLE)

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  - Generalized tonic-clonic seizures
  - Throwing hyperkinetic activity
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- Occurrence of nocturnal paroxysmal dyskinesia is often accompanied by nocturnal dystonia.
- DOC is often used for focal seizures, including carbamazepine and phenytoin, with minimal other therapeutic options.
- EEG shows interictal discharge with rhythmic sharp and slow waves, and ictal discharge with rhythmic sharp and slow waves, which suggests cortical involvement.

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