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Epileptic syndrome in children: understanding developmental and genetic factors

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Epileptic syndrome

Neonatal/infantile onset
Childhood Onset
Adolescent/Adult Onset

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Neonatal/infantile onset

Self-limited

- Self-limited (familial) neonatal epilepsy (SeLNE)
- Self-limited familial neonatal infantile epilepsy (SeLFNIE)
- Self-limited (familial) infantile epilepsy (SeLIE)
- Genetic epilepsy with febrile seizures plus (GEFS+) spectrum
- Myoclonic epilepsy in infancy

Developmental and epileptic encephalopathy

- Early-Infantile DEE (EIDEE)
- Epilepsy of Infancy with Migrating Focal Seizures (EIMFS)
- Myoclonic encephalopathy
- West syndrome
- Dravet syndrome

A full-term newborn boy developed brief focal tonic seizures on day 3 of life,

He was alert between episodes with normal feeding and no signs of encephalopathy.

His father had neonatal seizures that resolved spontaneously. Neurological exam and brain MRI were unremarkable.

Differential diagnosis

- Focal seizures
  - Metabolic disturbance (hypoglycemia)
  - Stroke
  - Birth Trauma
  - Brain malformation
  - Epileptic syndrome
    - Self-limited (familial) neonatal epilepsy (SeLNE)
    - Self-limited familial neonatal infantile epilepsy (SeLFNIE)

- Seizure type
- Time
- Family history

Self-limited (familial) neonatal epilepsy (SeLNE)

Onset: 2-7 days of life

Seizure: Focal motor seizures with tonic or clonic features, Sequential seizures, Autonomic seizure

EEG: Normal background or minor non-specific abnormality

Prognosis: Seizures usually remit by 6 months of age

Self-limited (familial) neonatal epilepsy (SeLNE)

an autosomal dominant familial focal epilepsy syndrome with onset in the neonatal or infantile period in different family members

Onset: Focal tonic seizures from day 1 to 23 months of life

Prognosis: seizures remit by age 12–24 months in all individuals

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Self-limited (familial) infantile epilepsy (SeLIE)

Onset: 3-20 months of life (peak 6 months)

Seizure: Focal seizures with Behavioral arrest, Autonomic features (e.g., cyanosis), Impaired awareness, Automatisms

EEG: Normal background or interictal EEG, may have midline spikes during SWS

Prognosis: usually remit within 1 year from the onset

Gene distribution in benign familial syndrome

Benign familial epilepsy syndromes	BFNE	*KCNQ2*	Subunit of voltage-gated K+ channel	AD
		KCNQ3	Subunit of voltage-gated K+ channel	AD
	BFNIE	*SCN2A*	Subunit of voltage-gated Na+ channel	AD
	BFIE	*PRRT2*	Protein-rich transmembrane protein 2	AD
		SCN2A	Subunit of voltage-gated Na+ channel	AD
		SCN8A	Subunit of voltage-gated Na+ channel	AD
	GEFS+	*SCN1A*	Subunit of voltage-gated Na+ channel	AD
		SCN1B	Subunit of voltage-gated Na+ channel	AD, AR
		GABRG2	Subunit of GABAa receptor	AD
		STX1B	Syntaxin 1B	AD
	Myoclonic epilepsy in infancy	Unknown	-	-

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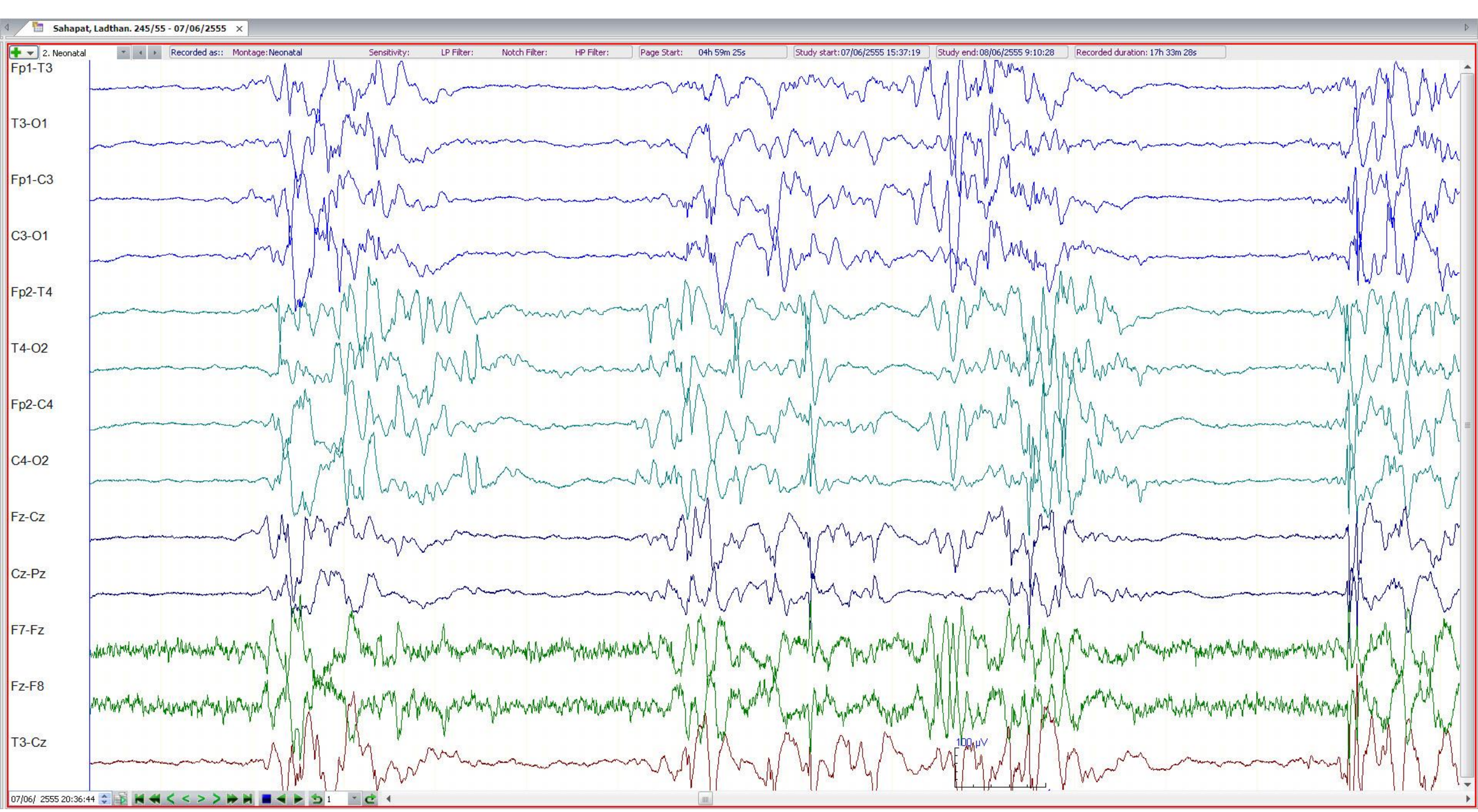
A full-term newborn girl began having brief focal tonic seizures on 3rd day of life.

She also had multiple seizure types, including myoclonic and subtle seizures.

PE: revealed signs of encephalopathy.

She showed poor response to multiple anti-seizure medications (PHB, PHT, TPM, LEV, B6)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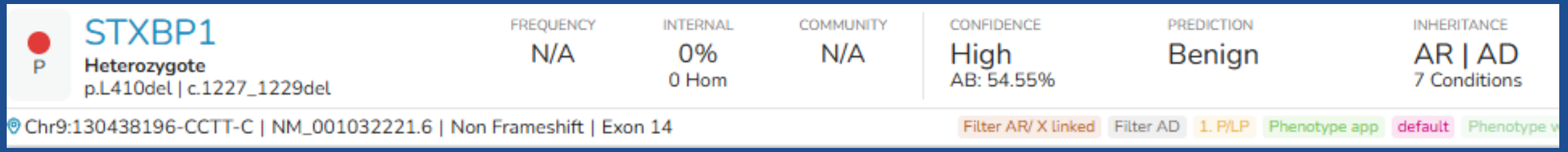
Differential diagnosis

- Focal seizures
  - Metabolic disturbance (hypoglycemia)
  - Stroke
  - Birth Trauma
  - Infection
  - Brain malformation
  - Epileptic syndrome
    - Early-Infantile DEE (EIDEE)



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Early-infantile developmental and epileptic encephalopathy syndrome (EIDEE)

Onset: < 3 months

Seizure: Tonic and/or myoclonic seizures, focal clonic, and epileptic spasms

- Abnormal neurological examination findings

EEG: burst- suppression pattern, diffuse slowing, or multi-focal discharges

Underlying etiologies including genetic, metabolic, and structural

Prognosis: usually remit within 1 year from the onset

Don’t forget

- Seizure mimic
- Treatable cause

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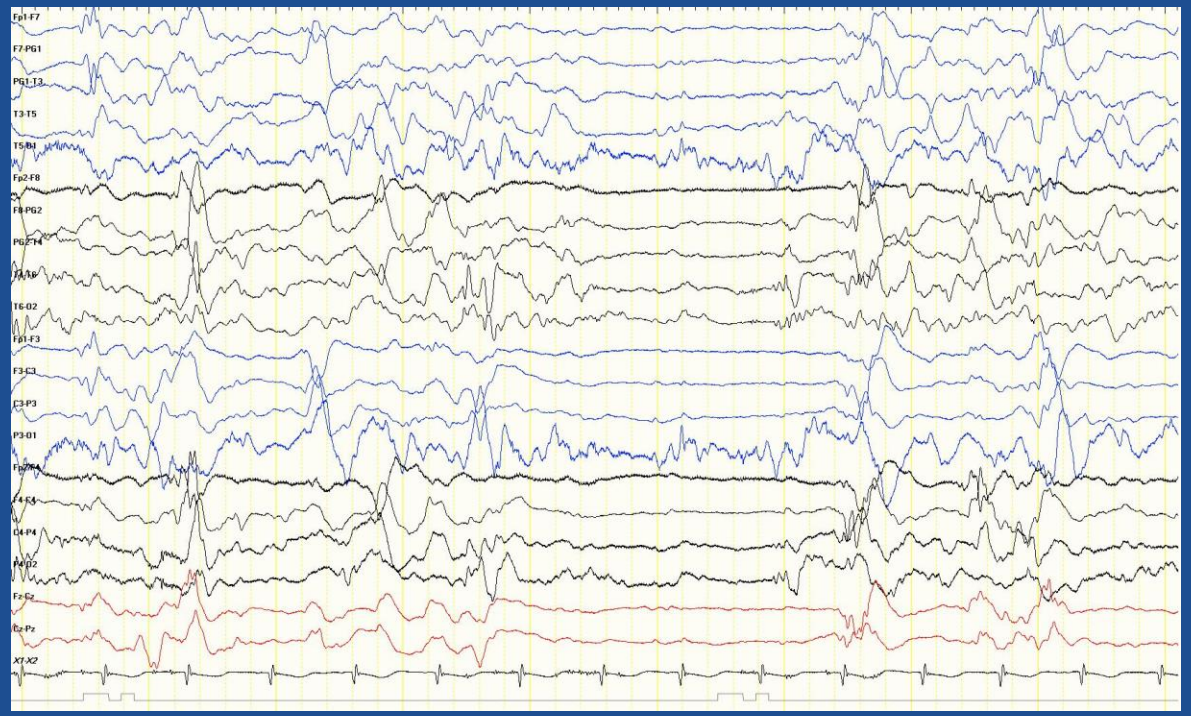
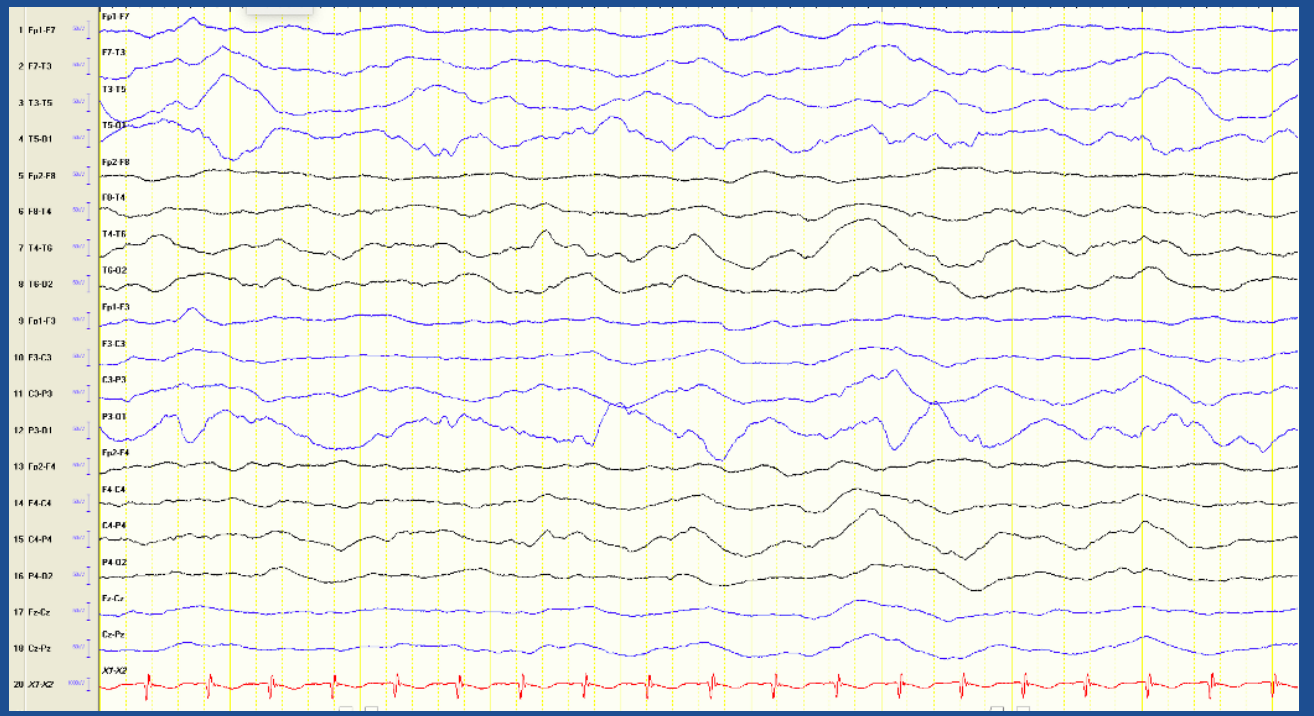
3-month-old girl presented with refractory status epilepticus

Failed ASM: PHB, LEV, MDZ, Pentobarbital



After first dose of pyridoxine treatment



Pyridoxine dependent epilepsy

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Epilepsy of infancy with migrating focal seizures

- Onset: 4 weeks (range day 1 to 6 months)
- Seizures
  - First phase: focal motor seizure accompanied by autonomic manifestations; many migrated from one side of the body to the other
  - Stormy phase: (3 weeks to 10 months): seizures become very frequent occurring in clusters for days or weeks
  - Third phase(1-5 years): seizure free
- Causative variants (69%): KCNT-1(27%), SCN2A(7%), KCNQ2, GABRB3, etc.

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West syndrome

2–3 per 10,000 live births

Onset: 2–12 months (peak 4–7 months)

Cause: Genetic (e.g., STXBP1, CDKL5), Structural-genetic (TSC1/2, ARX), Structural-congenital, Structural-acquired, Metabolic, or Unknown

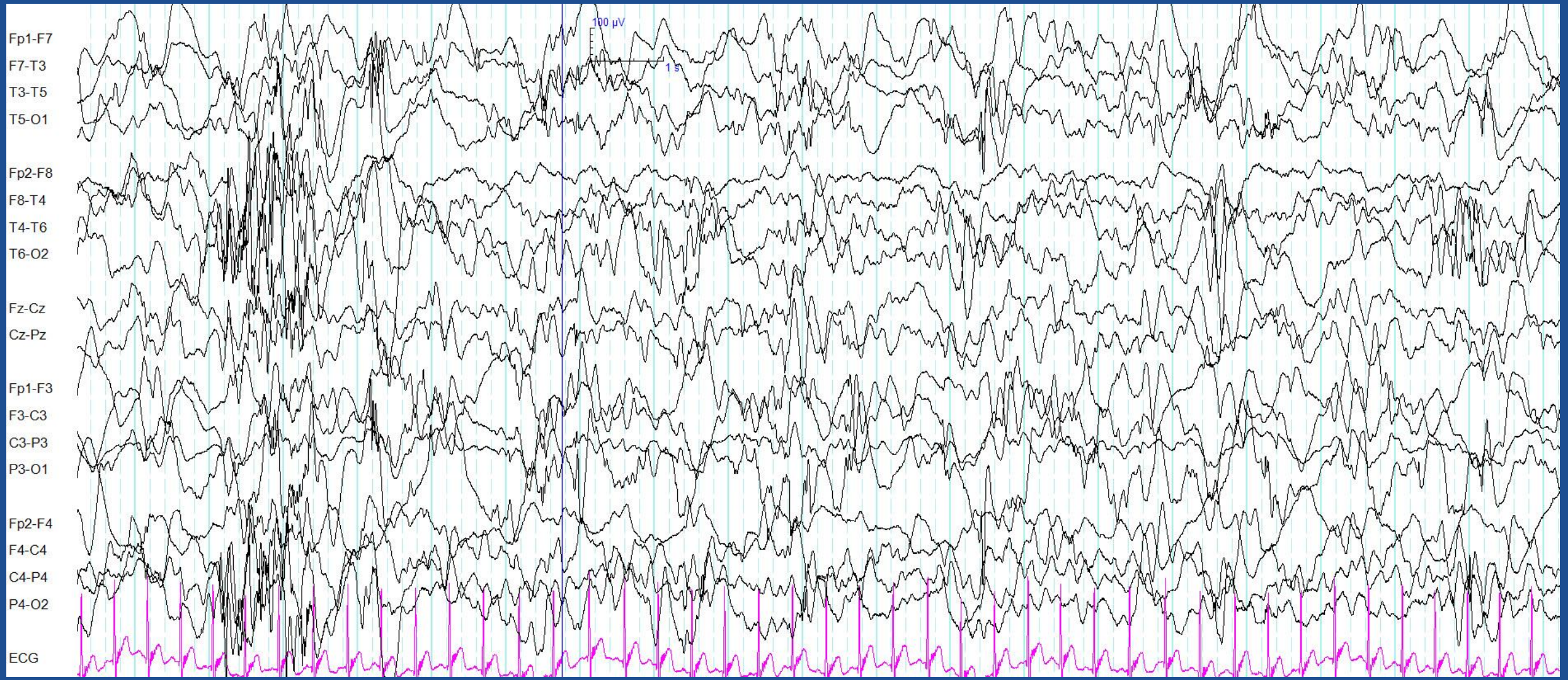
Seizures: Epileptic spasms

EEG: Hypsarrhythmia pattern

Comorbid: Developmental regression or delay

Treatment: ACTH, Prednisolone+ Vigabatrin, Prednisolone, Vigabatrin

Prognosis varies by etiology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ECG

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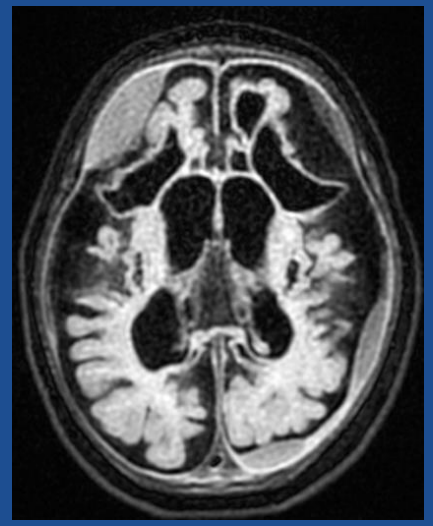
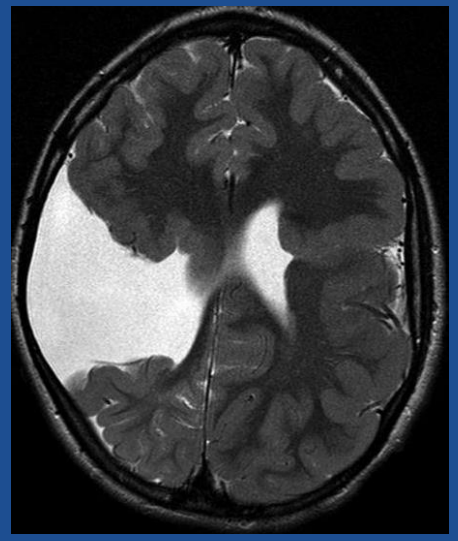
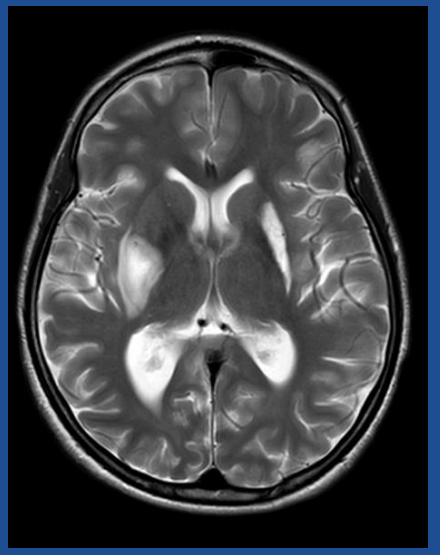


Structural acquired



Structural congenital



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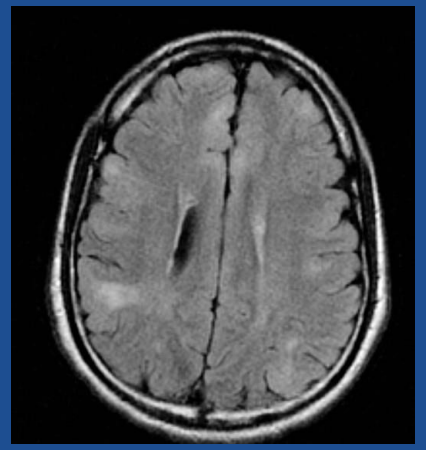


Structural genetic

Genetic

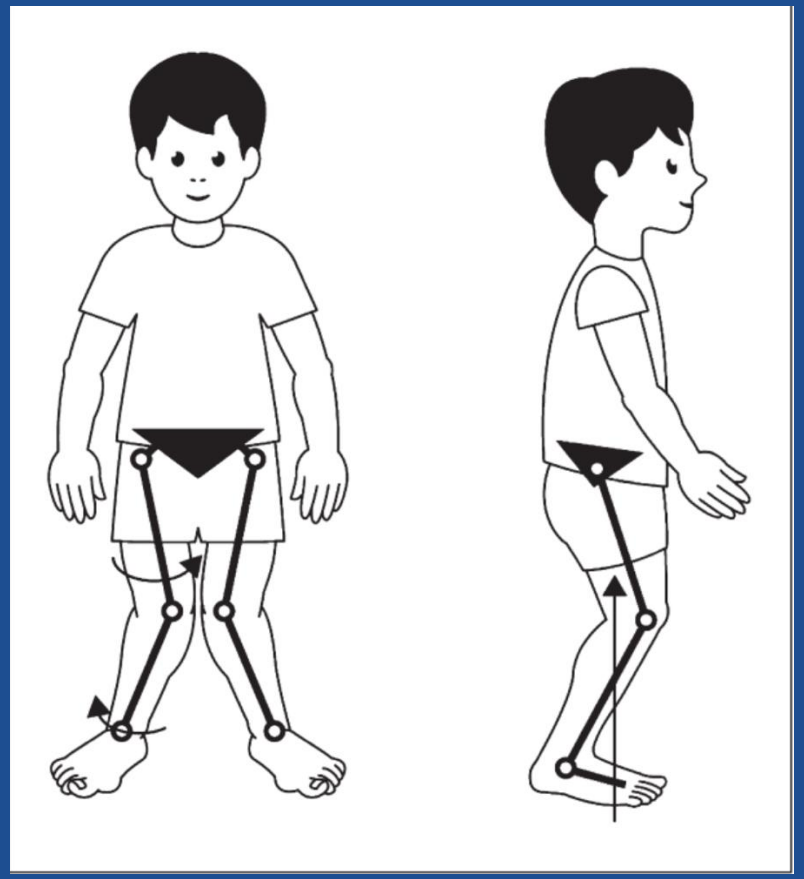
Idiopathic

- 6-month-old girl presented with recurrent prolonged or cluster GTC—following a febrile illness.
- Over the next several months, she had recurrent febrile and afebrile seizures, including hemiclonic and myoclonic seizures
- Often triggered by fever or warm temperatures.
- Development plateaued following seizure onset

Dravet syndrome

- Severe infantile-onset developmental and epileptic encephalopathy (DEE)
- Loss of function in *SCN1A* gene (80% of patients)
- Onset: 4-7 months (mean 5 months)
- Seizure
  - Febrile seizures, prolong or cluster
  - Multiple type seizures (GT, hemiclonic Sz, myoclonic seizure)
- Triggering Factors: Fever, Hot bath, Vaccine
- Developmental delay
- Drug resistance

Crouch gait



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Childhood onset

Self-limited

- Self-limited Epilepsy with Centrotemporal Spikes (SeLECTS)
- Self-limited epilepsy with autonomic seizures (SeLEAS)
- Childhood occipital visual epilepsy (COVE)
- Photosensitive occipital lobe epilepsy (POLE)

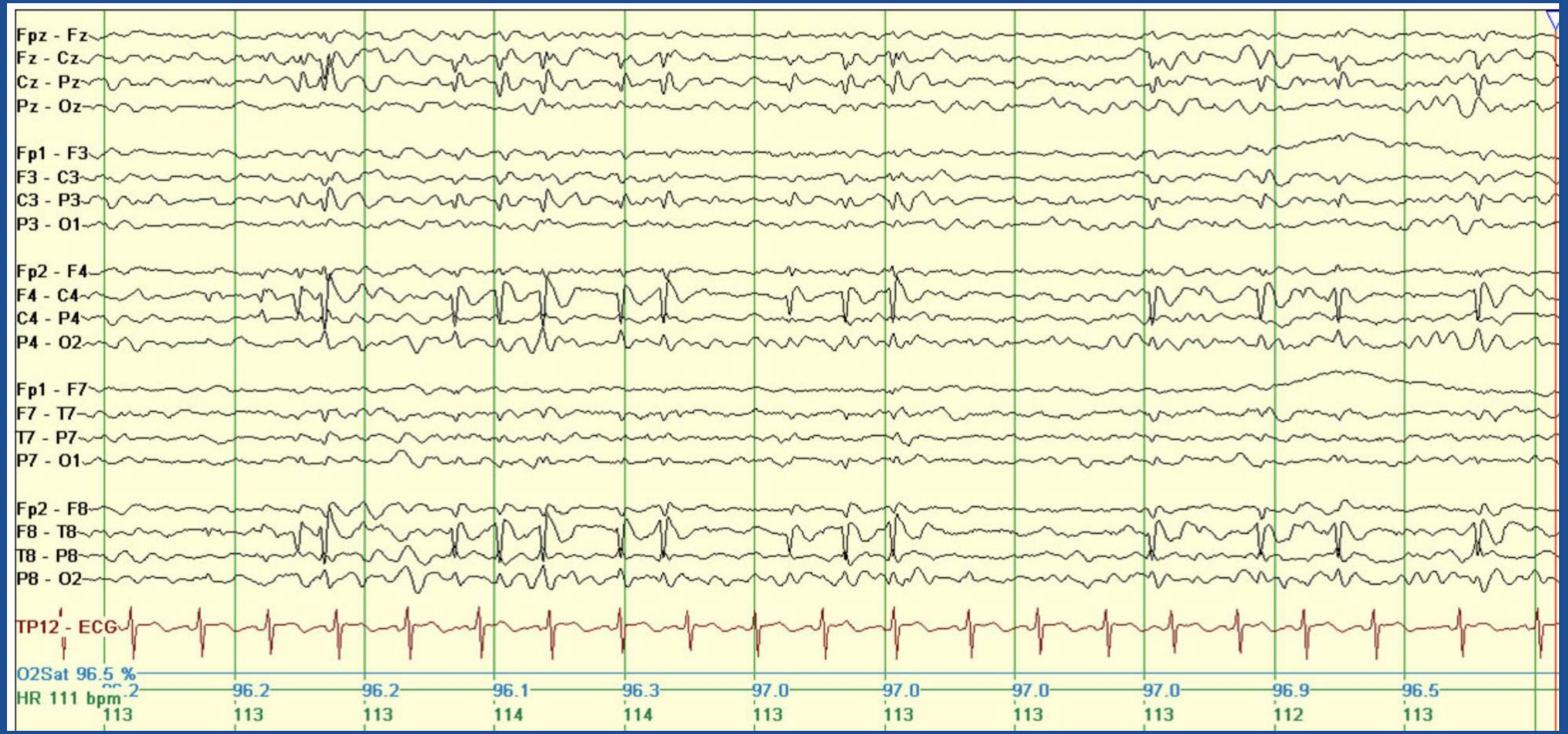
Genetic generalized epilepsy

- Childhood Absence epilepsy (CAE)
- Epilepsy with eyelid myoclonia (EEM)
- Epilepsy with myoclonic absence (EMA)

Developmental and epileptic encephalopathy

- Lennox–Gastaut syndrome(LGS)
- Epileptic encephalopathy with spike-and-wave activation in sleep
- Landau–Kleffner syndrome

- 7-year-old right-handed boy presented with facial twitching on the right side, drooling, and speech arrest, lasting approximately 2 minutes during sleep
- Normal development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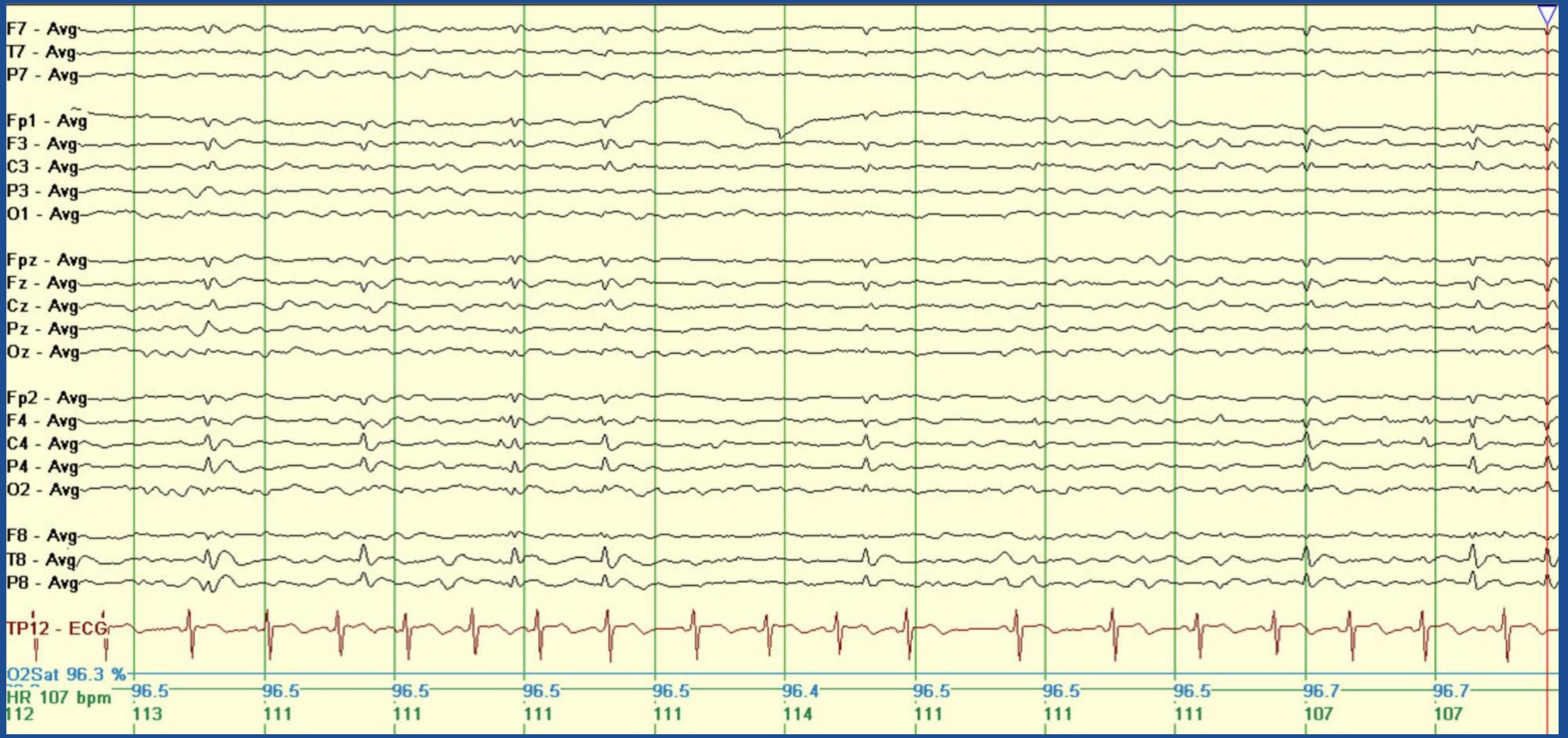
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Self-limited Epilepsy with Centrotemporal Spikes (SeLECTS)

- 6–7% of childhood epilepsies (6.1 per 100 000 children aged <16 years per year)
- **Onset:** 3–14 years (peak ~7 years)
- **Seizure:** focal clonic or tonic activity of the throat/tongue and the lower face during sleep
- **EEG:** Centrotemporal spikes (sleep-activated), with a transverse dipole
- Remits by adolescence
- Excellent prognosis

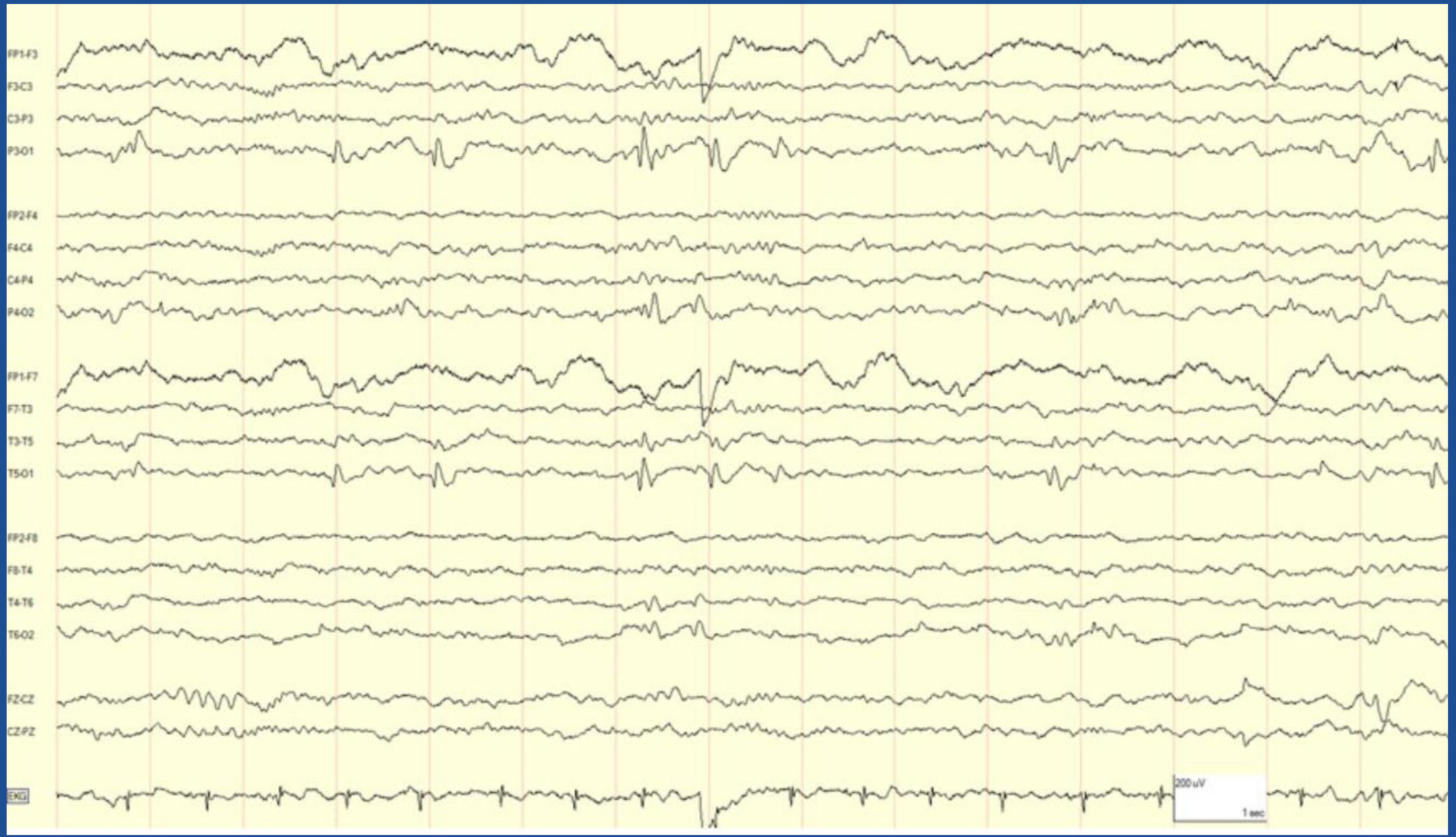
Differential diagnosis

- DEE- SWAS or EE- SWAS
- Focal seizures due to structural brain abnormality
- Other SeLFEs

Caution

- Onset:<3 years or >14 years
- Seizure type: GTC during wakefulness
- Neurocognitive regression with a continuous spike-and wave pattern in sleep (suggests EE-S WAS)
- Causal lesion on brain MRI

- A 6-year-old healthy girl presented with nocturnal seizure. The episode began with nausea and vomiting, followed by unresponsiveness and rightward eye deviation lasting about 10 minutes.
- Normal development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T5-O1
FP2-F8
F8-T4
T4-T6
T6-O2
FZ-CZ
CZ-PZ
EKG
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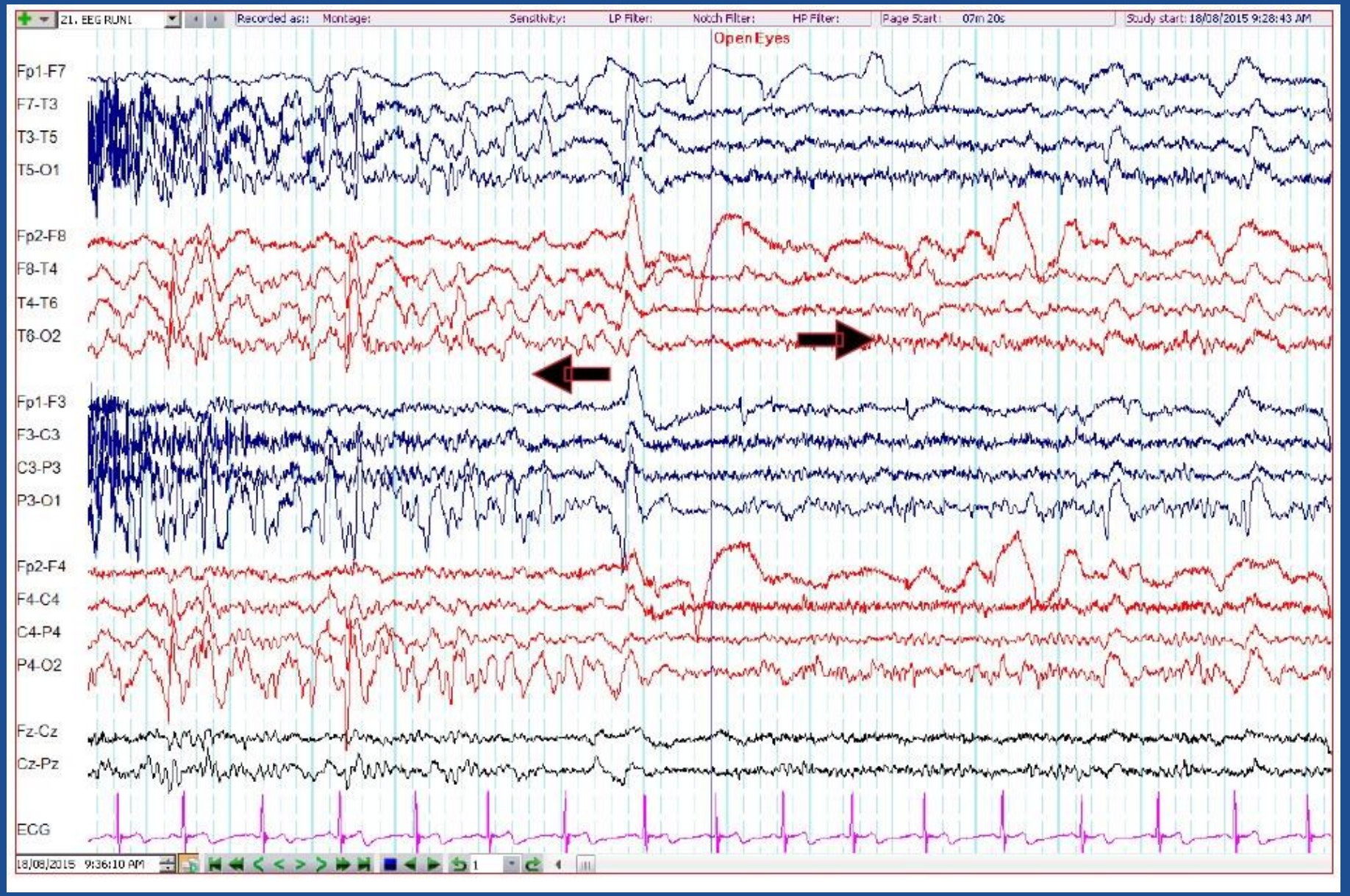
Self-limited epilepsy with autonomic seizures (SeLEAS)

- Prevalence:13% of childhood epilepsies
- Onset: 3–14 years (peak ~7 years)
- Seizure: Focal autonomic seizures (**Vomiting (80%)**, pallor, flushing, nausea, malaise, or abdominal pain), with or without impaired awareness
- EEG: Multifocal spike/sharp wave over the posterior region, fixation-off sensitivity (not pathognomonic)
- Prognosis and comorbid: remit within 1– 2years, with normal neurodevelopment

- An 8-year-old previously healthy boy presented with recurrent early morning episodes of visual hallucinations, such as seeing flashing lights and colored shapes
- Normal development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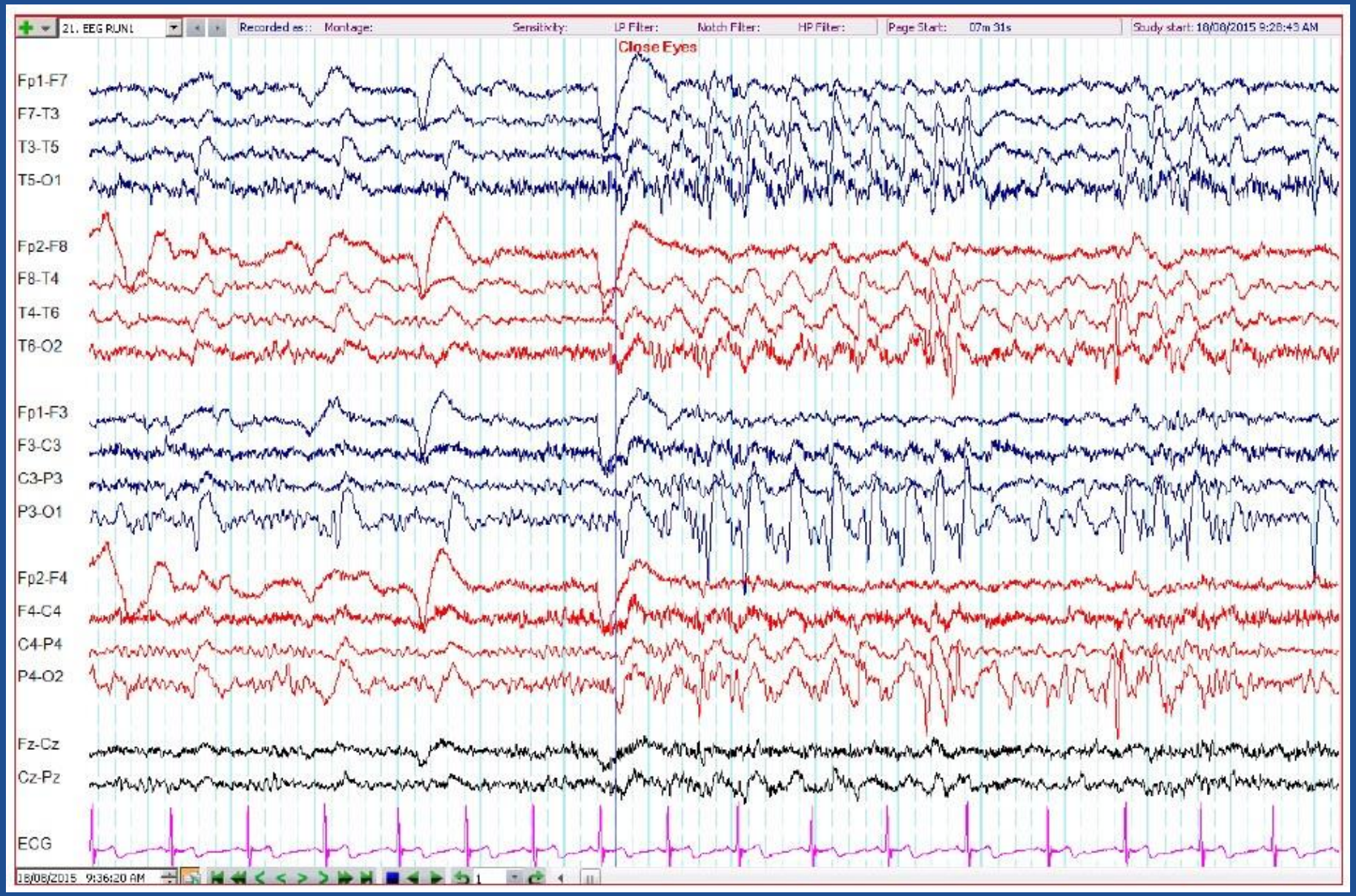
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Childhood occipital visual epilepsy (COVE)

- Prevalence: 0.3% of children epilepsies
- Onset: 8-9 years (1-19 years)
- Seizure characteristics
  - Focal sensory visual seizures during wakefulness, described as small multicolored circles, ictal blindness, complex visual hallucinations or illusions
- EEG: Occipital discharge, Fixation-off sensitivity (not pathognomonic), **not triggered by photic stimuli**
- Remission occurs in 50%– 80% of patients by puberty with or without administration of ASM

Photosensitive occipital lobe epilepsy (POLE)

- Prevalence: 0.7% of childhood epilepsies
- Onset: 4 and 17 years (mean = 11 years)
- Seizure characteristics:
  - Visual sensory symptoms include lights, colored spots, formed visual hallucinations
- EEG: Occipital discharge aggravated by eye closure and intermittent photic stimulation
- Prognosis: varies

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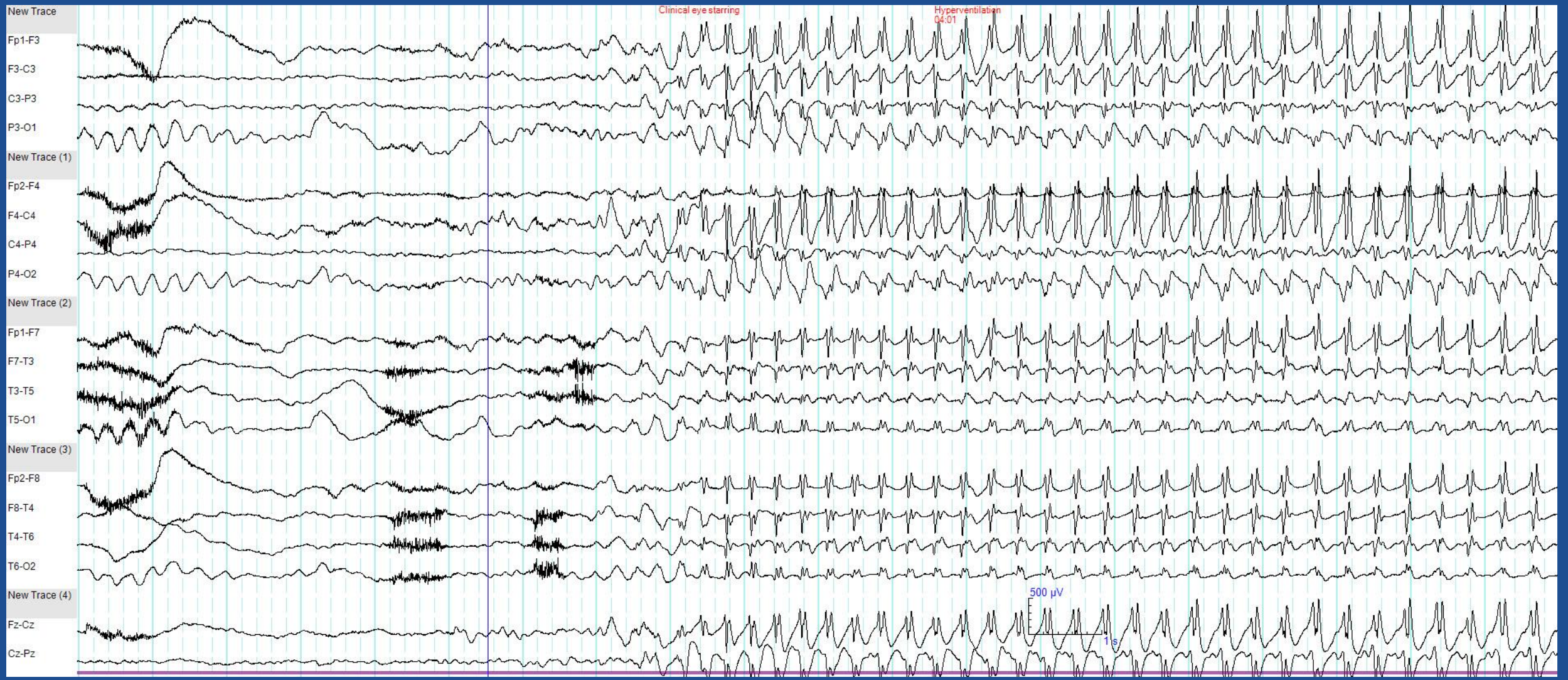
- Landau–Kleffner syndrome

- A 6-year-old girl was brought in for evaluation of frequent brief staring spells noticed by her teacher, occurring multiple times daily.
- Normal development

Childhood Absence Seizure

- Prevalence: approximately18% of epilepsy in school-aged children
- **Onset**: 4– 10 years (range = 2– 13 years)
- **Etiologies**: Consider genetic etiology, but genetic testing is not part of current routine genetic diagnosis
- **Seizure: Absence seizure**
- **EEG**: Generalized slow spike and wave complexes of 3 Hz Hz, OIRDA (21%)
- **Treatment**: Valproate, Ethosuximide
- Prognosis: Drug responsive, remits by early adolescence in 60% of patients, possible report LD/ADHD

Clinical and Laboratory Characteristics	Absence	Complex Partial	Daydreaming
Frequency/day	Multiple	Rarely >1–2	Situation-dependent
Duration	Frequently <10 sec; rarely >30 sec	Average duration >1 min; rarely <10 sec	Seconds to minutes
Aura	Never	Frequent	Never
Clonic component	Common; eyeblinking common	Rare	Never
Postictal impairment	Never	Frequent	Never
Seizures activated by			
Hyperventilation	Frequent	Rare	Never
Photic stimulation	Frequent	Rare	Never
EEG			
Interictal	Generalized spike wave	Focal spikes, sharp waves	Normal
Ictal	Generalized spike wave	Rhythmic spikes, sharp waves, or slow waves	Normal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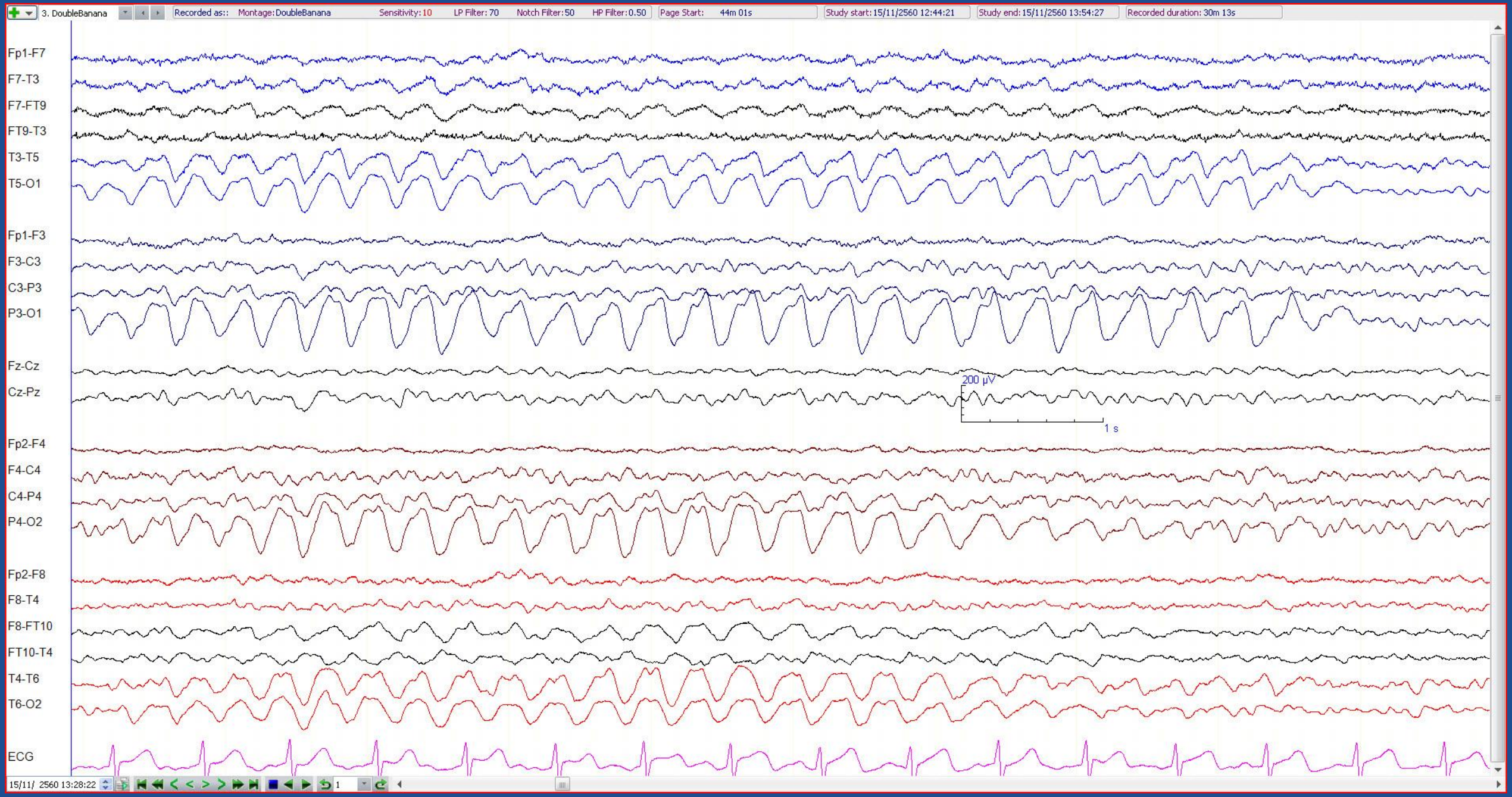
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Feature	CAE	JAE
Age at onset		
Usual	4–10 years	9–13 years
Range	2–13; caution if diagnosing at <4 years of age	8–20 years; exceptional cases may present in adulthood
Development	Typically normal, but may have learning difficulties or ADHD	Typically normal, but may have learning difficulties or ADHD
Absences		
Frequency	At least daily to multiple per day but may be underrecognized by family	Less than daily
Duration	Typical duration = 3–20 s	Typical duration = 5–30 s
Impaired awareness	Severe loss of awareness	Less complete impairment of awareness
Other seizure types		
Febrile	Occasional	Occasional
Generalized tonic–clonic seizure	Rarely precede or occur during period of frequent absences but may occur later with evolution to other IGE syndrome	May precede and commonly occur during the period of frequent absences
Myoclonic	Prominent myoclonus exclusionary	Prominent myoclonus exclusionary
EEG background	OIRDA in 21%	Normal
Interictal epileptiform discharge		
Awake	2.5–4-Hz generalized spike-wave	3–5.5-Hz generalized spike-wave
Asleep	Polyspike and wave may be seen in drowsiness and sleep only	Polyspike and wave may be seen in drowsiness and sleep only
Irregular generalized spike-wave	Uncommon	More common than CAE Discharges are more frequent than in CAE
Photoparoxysmal response	Rare IPS triggers generalized spike-wave in 15%–21% but does not induce seizures	Rare IPS triggers generalized spike-wave in 25% but does not induce seizures
Hyperventilation induction	87%	87%
Ictal EEG	Regular 3-Hz (range = 2.5–4 Hz) generalized spike-wave; 21% may have absences starting at 2.5-Hz spike-wave, and 43% may have absences starting at 4 Hz; if no generalized spike-wave is seen with hyperventilation for 3 min in an untreated patient, CAE can be excluded Disorganized discharges[a] less frequent	Regular 3–5.5-Hz generalized spike-wave If no generalized spike-wave is seen with hyperventilation for 3 min in an untreated patient, JAE can be excluded Disorganized discharges[a] 8 times more frequent than CAE

Epilepsy with myoclonic absence

- Prevalence: Unknown
- **Onset**: 7 years (range = 1– 12 years)
- **Etiologies**: polygenic
- **Seizure: Absence seizures are associated with rhythmic 3- Hz jerks of the upper limbs**
- **EEG**: Generalized 3 Hz spike/polyspike wave complexes
- **Treatment**: Valproate
- Prognosis: Favorable response if myoclonic absence seizures are the only seizure type and are controlled

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Childhood onset

Self-limited

- Self-limited Epilepsy with Centrotemporal Spikes (SeLECTS)
- Self-limited epilepsy with autonomic seizures (SeLEAS)
- Childhood occipital visual epilepsy (COVE)
- Photosensitive occipital lobe epilepsy (POLE)

Genetic generalized epilepsy

- Childhood Absence epilepsy (CAE)
- Epilepsy with eyelid myoclonia (EEM)
- Epilepsy with myoclonic absence (EMA)

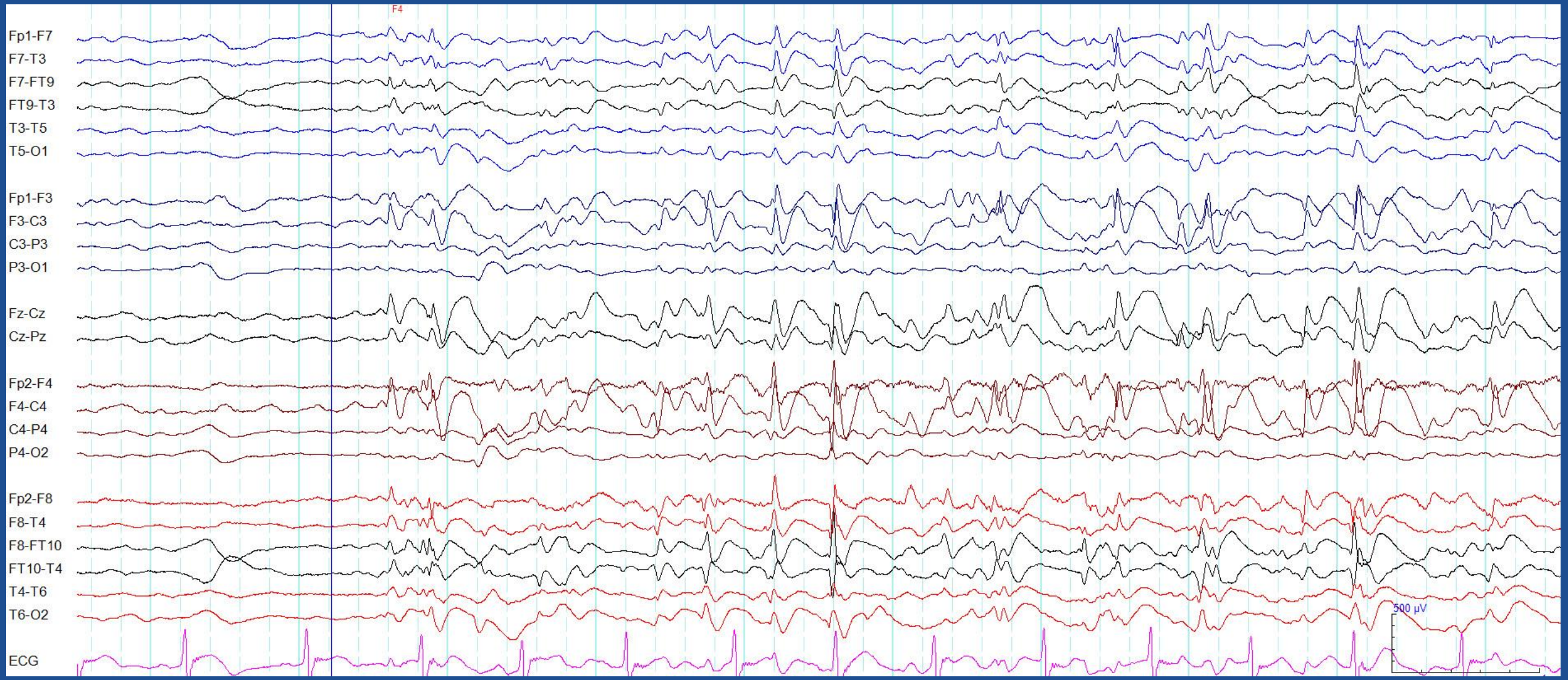
Developmental and epileptic encephalopathy

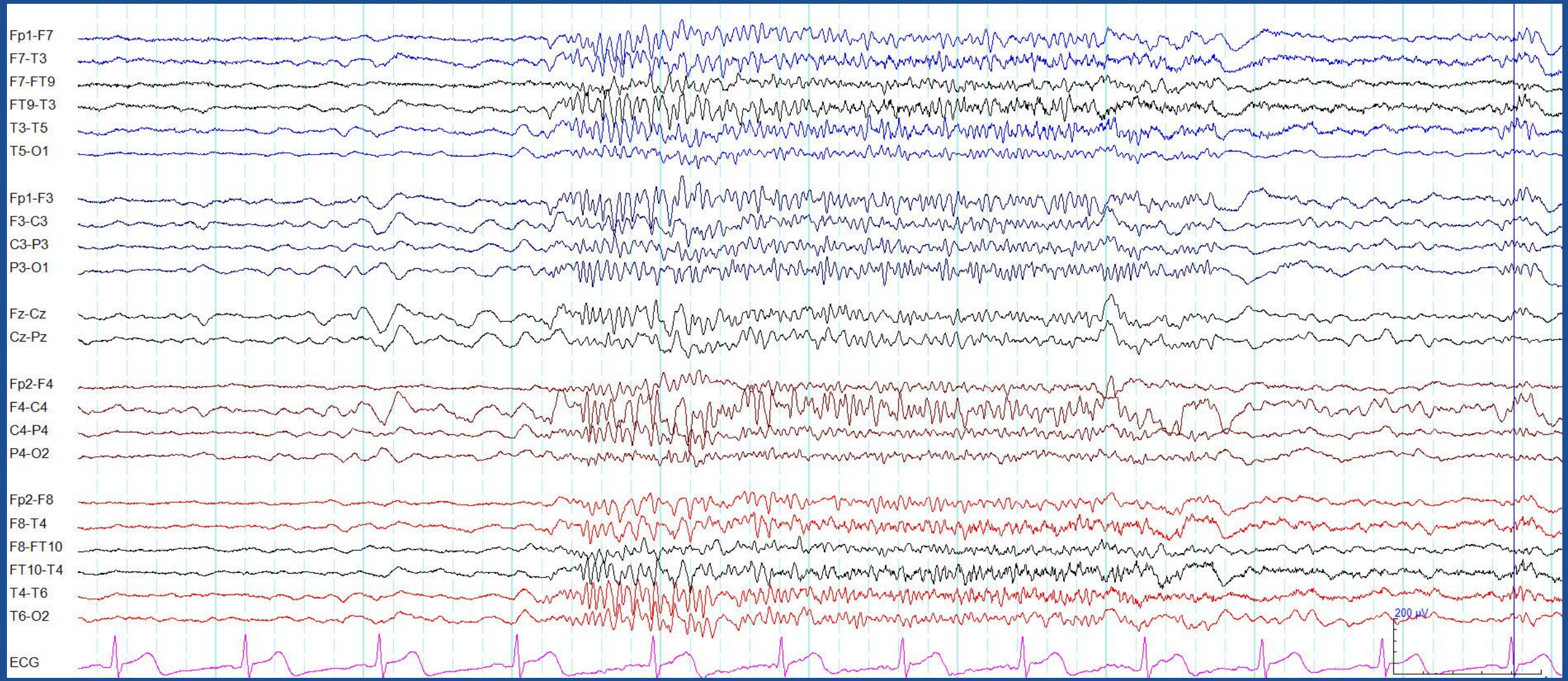
- Lennox–Gastaut syndrome(LGS)
- Epileptic encephalopathy with spike-and-wave activation in sleep
- Landau–Kleffner syndrome

Lennox-Gastaut Syndrome (LGS)

- Prevalence: 1–10% of childhood epilepsies
- **Onset**: 1–8 years (typically 3–5 years)
- **Etiologies**: Structural or genetic, May follow west syndrome
- **Seizure:** Multiple seizure types (tonic (mandatory), atonic, atypical absences)
- **EEG**: Generalized slow spike- and- wave complexes of <2.5 Hz, Generalized paroxysmal fast activity in sleep
- Cognitive and behavioral impairments
- **Treatment**: Often requires polytherapy
- Prognosis: Developmental outcome typically poor

- An 8-year-old boy with a history of infantile spasms diagnosed at 7 months of age presented with multiple seizure types, including tonic seizures during sleep, atypical absences, and drop attacks beginning around age 3.
- Developmental delay
- The patient is currently on polytherapy with poor seizure control.


F4
Fp1-F7
F7-T3
F7-FT9
FT9-T3
T3-T5
T5-O1
Fp1-F3
F3-C3
C3-P3
P3-O1
Fz-Cz
Cz-Pz
Fp2-F4
F4-C4
C4-P4
P4-O2
Fp2-F8
F8-T4
F8-FT10
FT10-T4
T4-T6
T6-O2
ECG
500 µV


Fp1-F7
F7-T3
F7-FT9
FT9-T3
T3-T5
T5-O1
Fp1-F3
F3-C3
C3-P3
P3-O1
Fz-Cz
Cz-Pz
Fp2-F4
F4-C4
C4-P4
P4-O2
Fp2-F8
F8-T4
F8-FT10
FT10-T4
T4-T6
T6-O2
ECG
200 µV

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Landau-Kleffner Syndrome (LKS)

- Prevalence: 1–10% of childhood epilepsies
- **Onset**: Ages 2-8 years, with a peak around ages 3-6 years
- **Etiologies**: *GRIN2A* (20%)
- **Seizure:** Focal motor, GC, atypical absence seizures
  - Seizures never occur in 20-30% of patients
- Language regression and Neuropsychiatric problem (ADHD, Emotional lability)
- **EEG**: Epileptiform discharge over posterior temporal regions
- **Treatment**:
  - Valproate, ethosuximide or clobazam
  - Prednisolone

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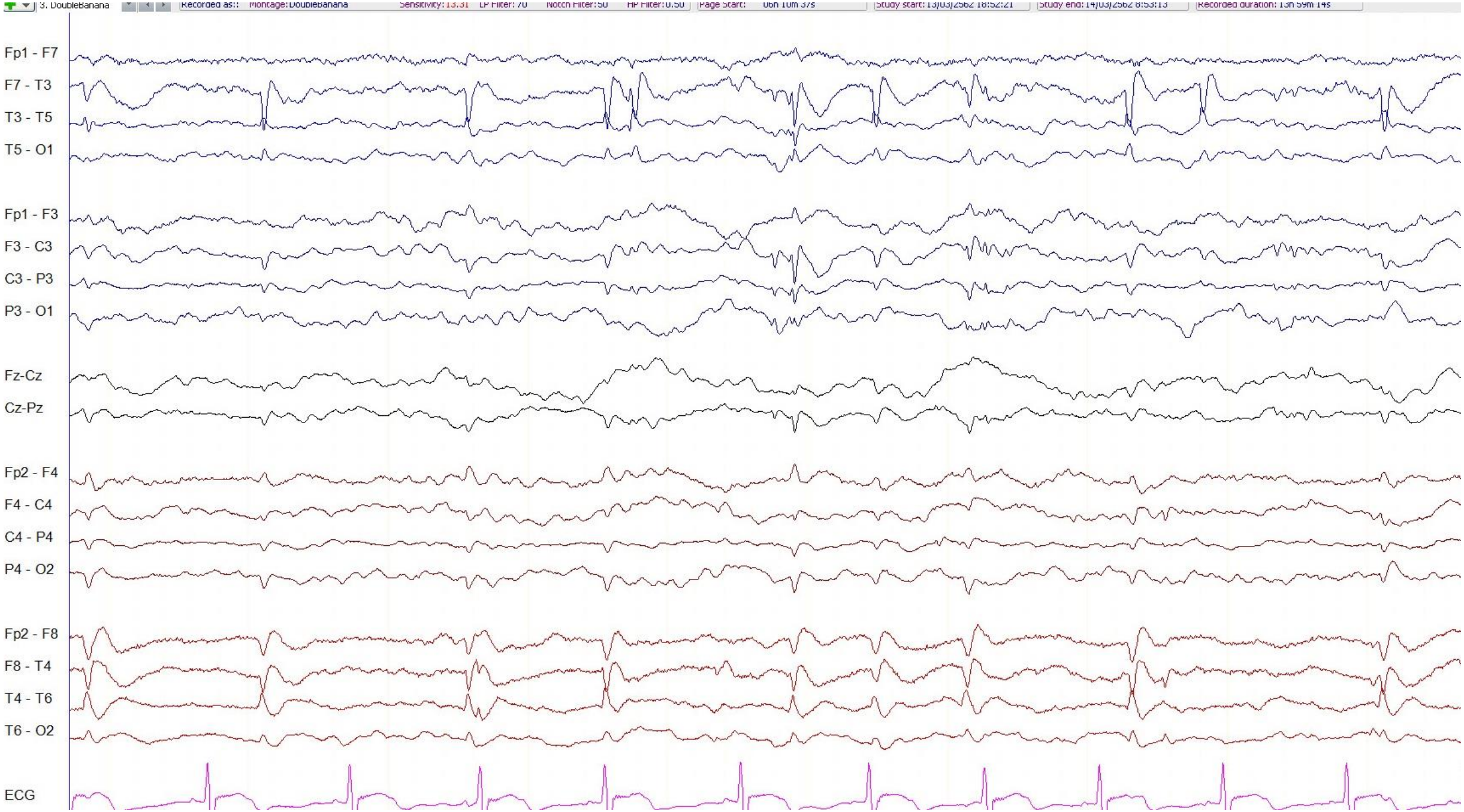
- 6-year-old boy developed anxiety, fear of causing harm, insomnia, social withdrawal, and bedwetting. He later showed increased anxiety, school refusal, and appetite loss without hallucinations for 3 weeks

Differential diagnosis

- PANs/PANDAS
- Autoimmune encephalitis
- DEE-SWAS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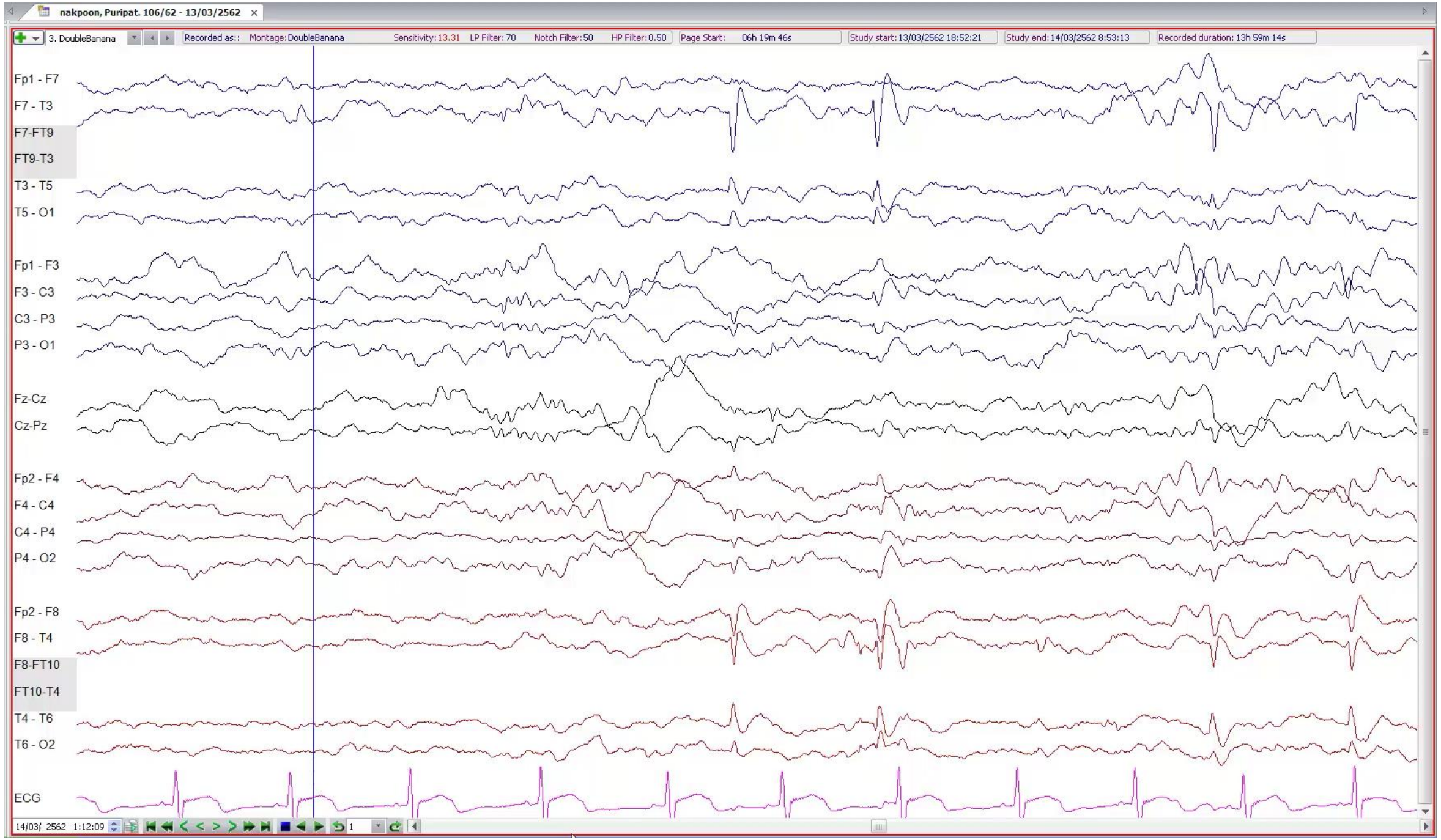
Recorded duration: 13h 59m 14s
Fp1 - F7
F7 - T3
T3 - T5
T5 - O1
Fp1 - F3
F3 - C3
C3 - P3
P3 - O1
Fz-Cz
Cz-Pz
Fp2 - F4
F4 - C4
C4 - P4
P4 - O2
Fp2 - F8
F8 - T4
T4 - T6
T6 - O2
ECG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Study end:14/03/2562 8:53:13
Recorded duration: 13h 59m 14s
Fp1 - F7
F7 - T3
F7-FT9
FT9-T3
T3 - T5
T5 - O1
Fp1 - F3
F3 - C3
C3 - P3
P3 - O1
Fz-Cz
Cz-Pz
Fp2 - F4
F4 - C4
C4 - P4
P4 - O2
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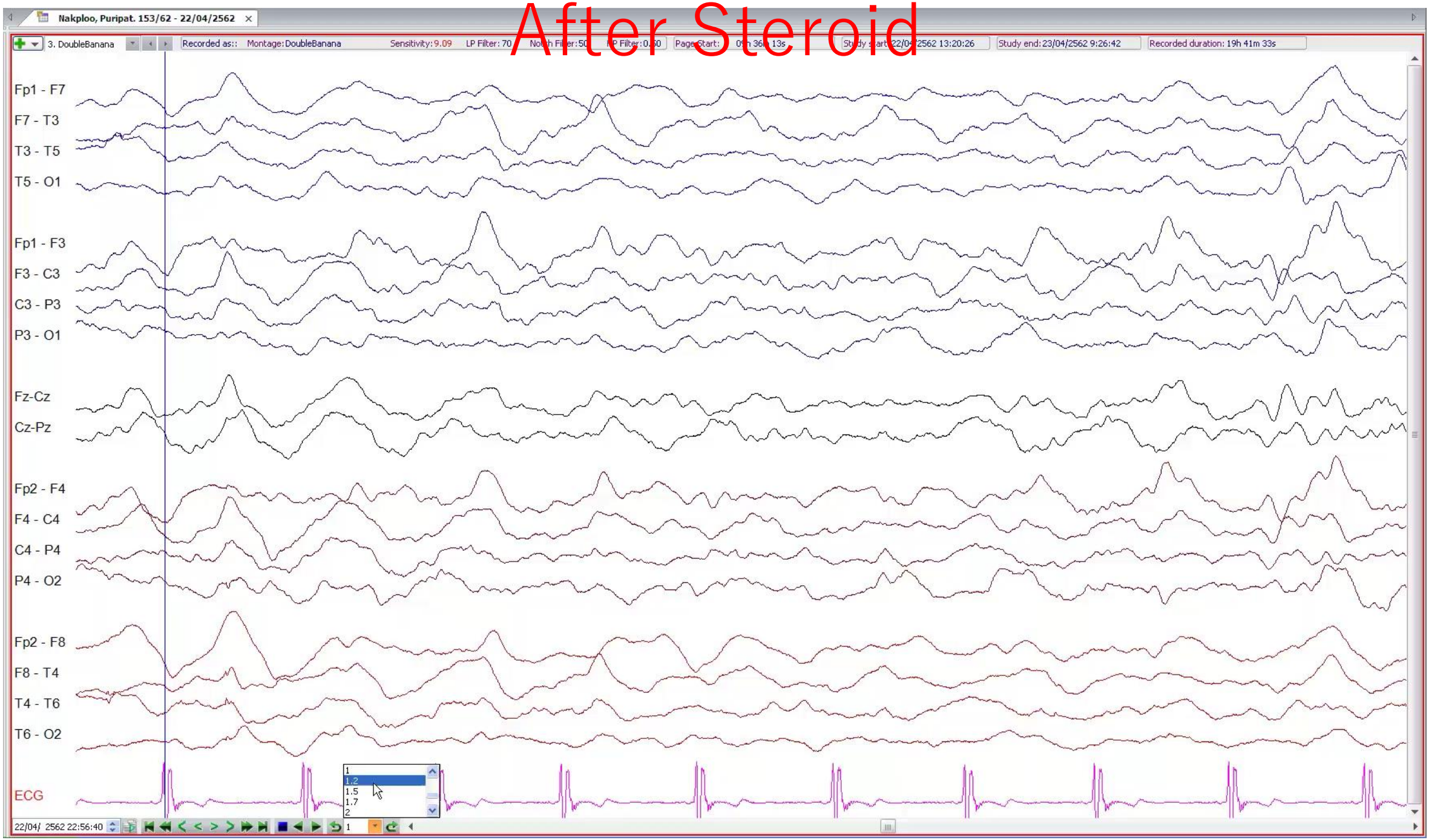
T6 - O2
ECG
14/03/ 2562 1:12:09


After Steroid
Nakploo, Puripat. 153/62 - 22/04/2562
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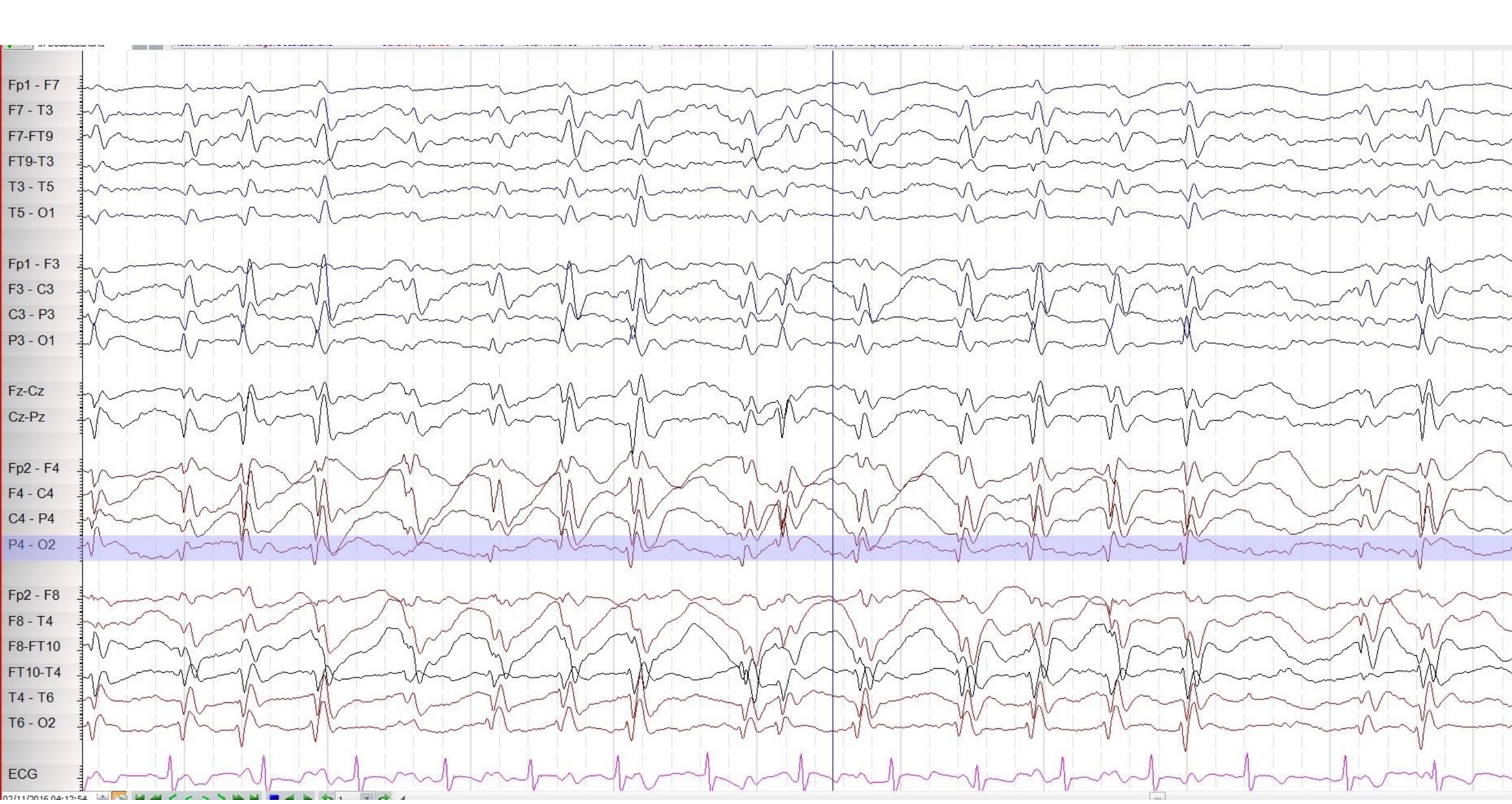
Fp1 - F7
F7 - T3
T3 - T5
T5 - O1
Fp1 - F3
F3 - C3
C3 - P3
P3 - O1
Fz-Cz
Cz-Pz
Fp2 - F4
F4 - C4
C4 - P4
P4 - O2
Fp2 - F8
F8 - T4
T4 - T6
T6 - O2
ECG
1
1.2
1.5
1.7
2
22/04/ 2562 22:56:40
1

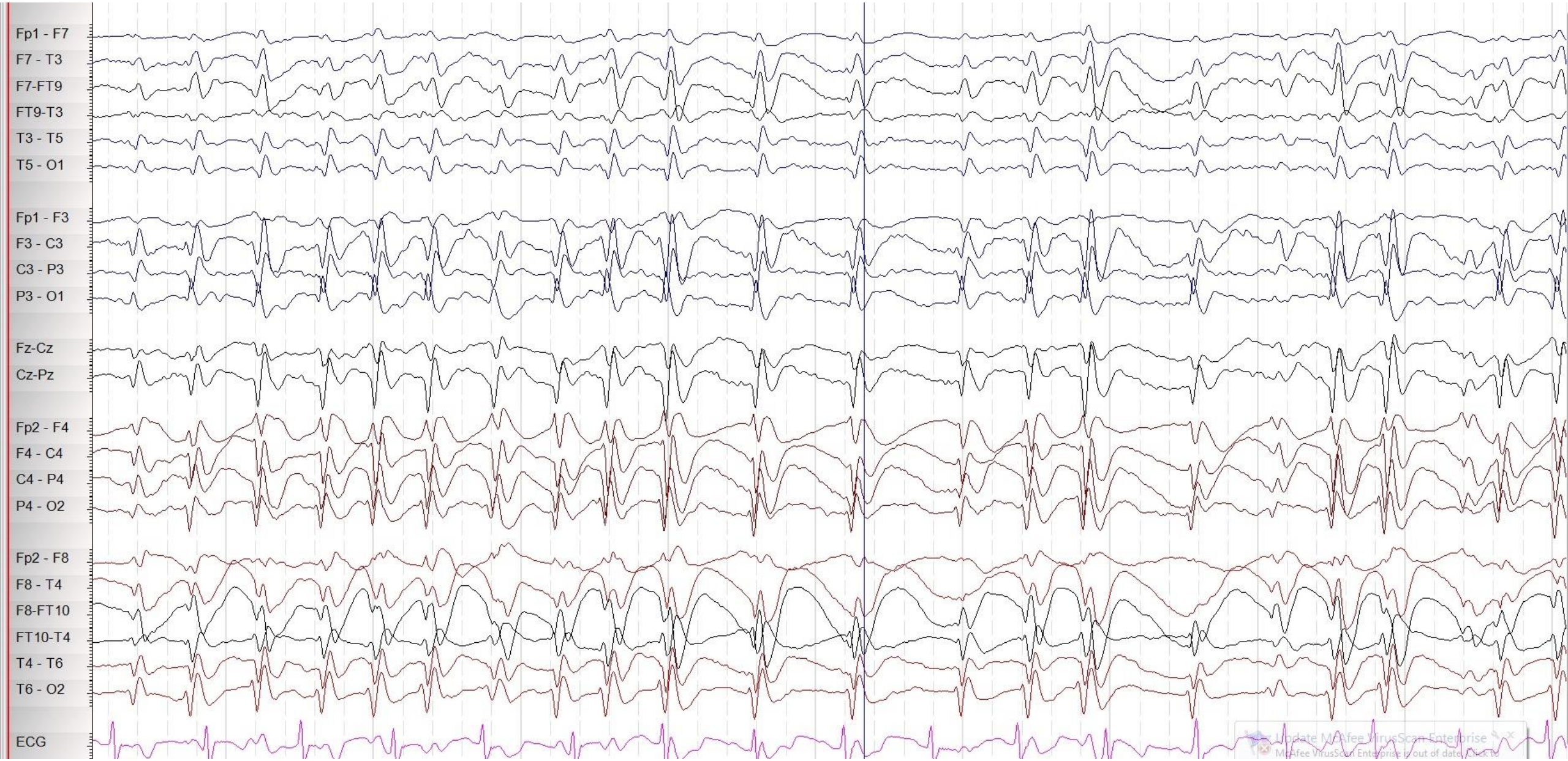
DEE-SWAS and EE-SWAS

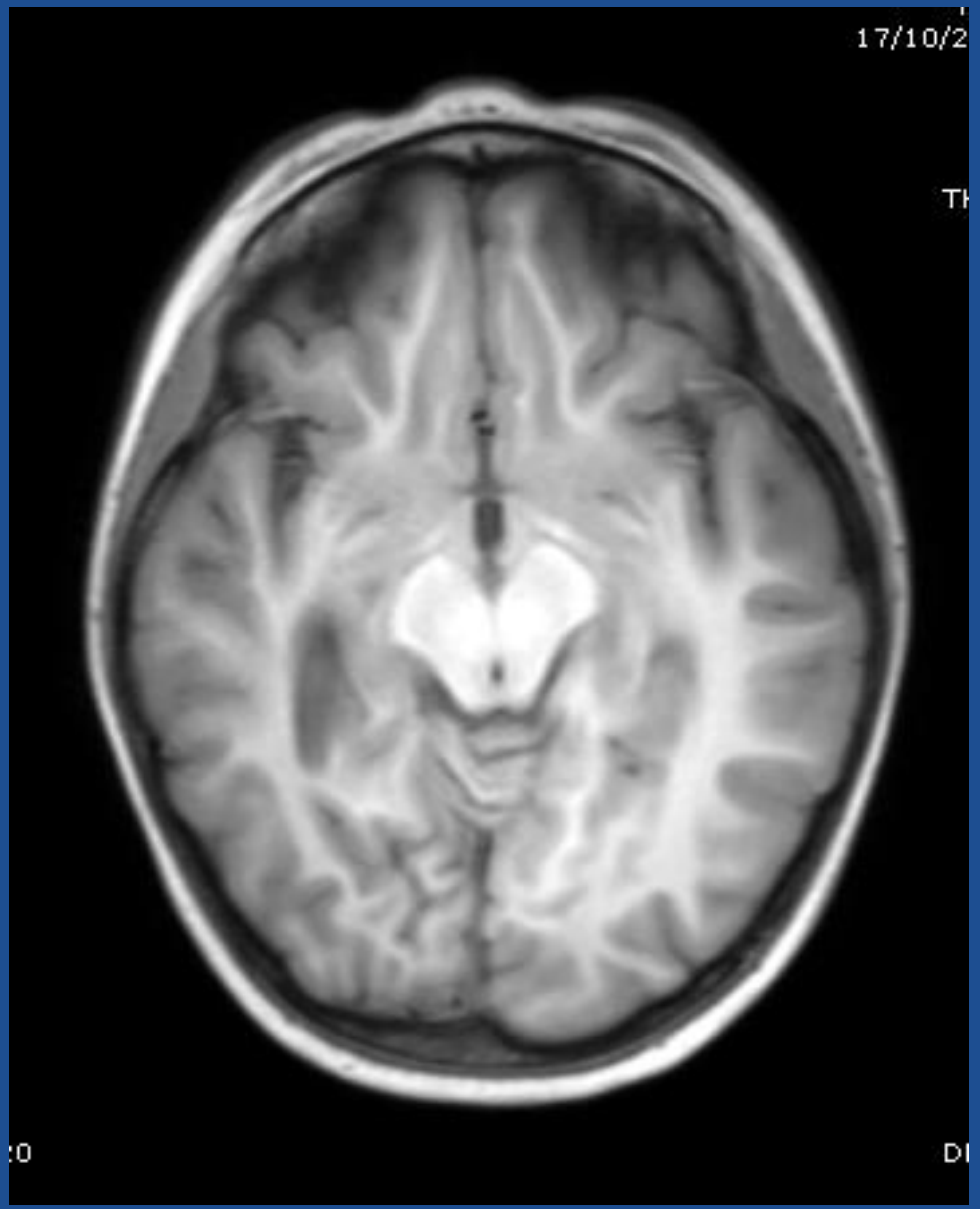
- combinations of
  - developmental regression (auditory agnosia, global regression of behavior and motor skills)
  - marked spike- and- wave activation in sleep
- 0.5%– 0.6% of all epilepsy in pediatric tertiary centers
- Seizure: Typically focal motor and focal to bilateral tonic–clonic seizures, atypical absence seizures, atonic seizures, and focal motor seizures with negative myoclonus
- EEG: Slow (1.5–2 Hz) spike and wave abnormalities in NREM, may occur more focally (typically frontally) or multifocally

DEE-SWAS and EE-SWAS

- MRI brain is recommended.
  - Abnormal (41-49%): PVL, cortical dysplasia, polymicrogyria
- Clinical seizures typically remit around puberty, even in patients with a structural lesion


Fp1 - F7
F7 - T3
F7-FT9
FT9-T3
T3 - T5
T5 - O1
Fp1 - F3
F3 - C3
C3 - P3
P3 - O1
Fz-Cz
Cz-Pz
Fp2 - F4
F4 - C4
C4 - P4
P4 - O2
Fp2 - F8
F8 - T4
F8-FT10
FT10-T4
T4 - T6
T6 - O2
ECG


Fp1 - F7
F7 - T3
F7-FT9
FT9-T3
T3 - T5
T5 - O1
Fp1 - F3
F3 - C3
C3 - P3
P3 - O1
Fz-Cz
Cz-Pz
Fp2 - F4
F4 - C4
C4 - P4
P4 - O2
Fp2 - F8
F8 - T4
F8-FT10
FT10-T4
T4 - T6
T6 - O2
ECG


17/10/2
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DI

Epileptic syndrome

Neonatal/infantile onset

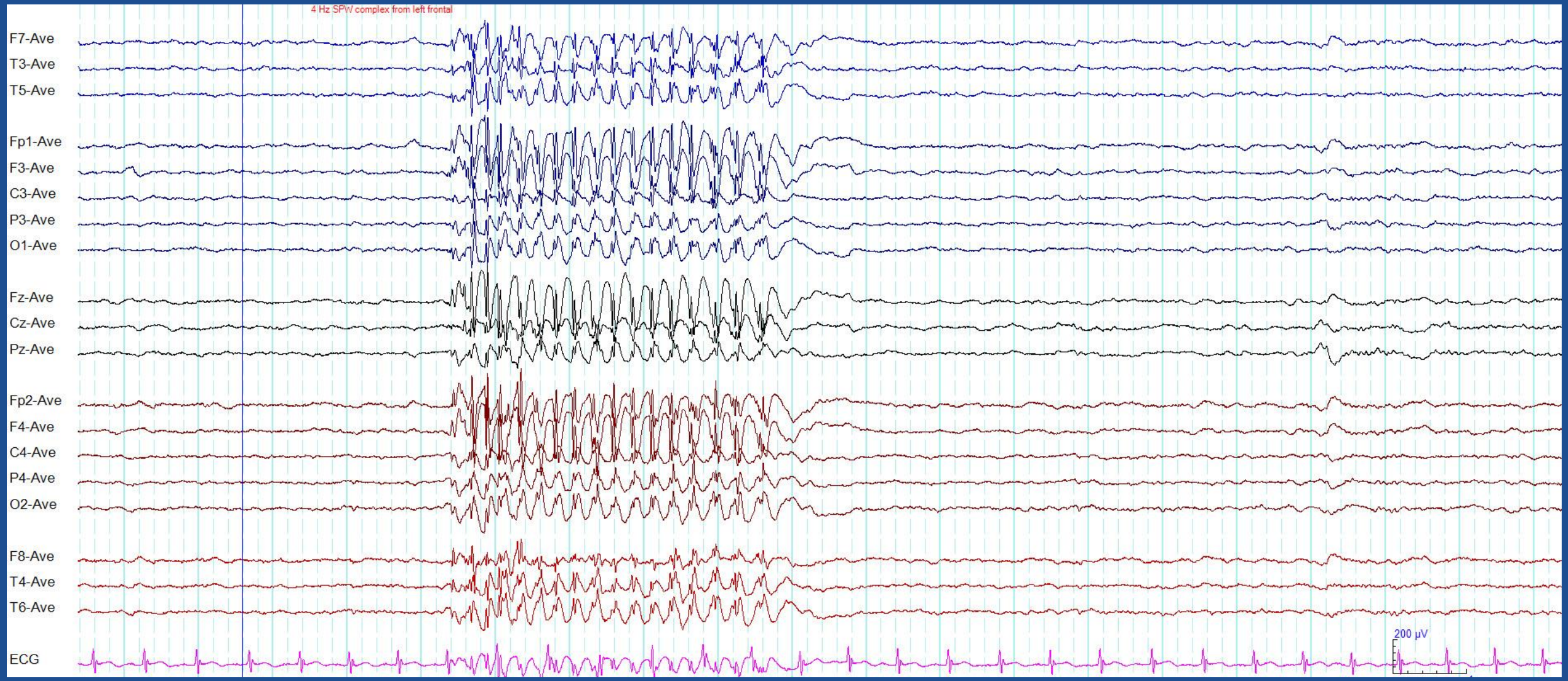
Childhood Onset

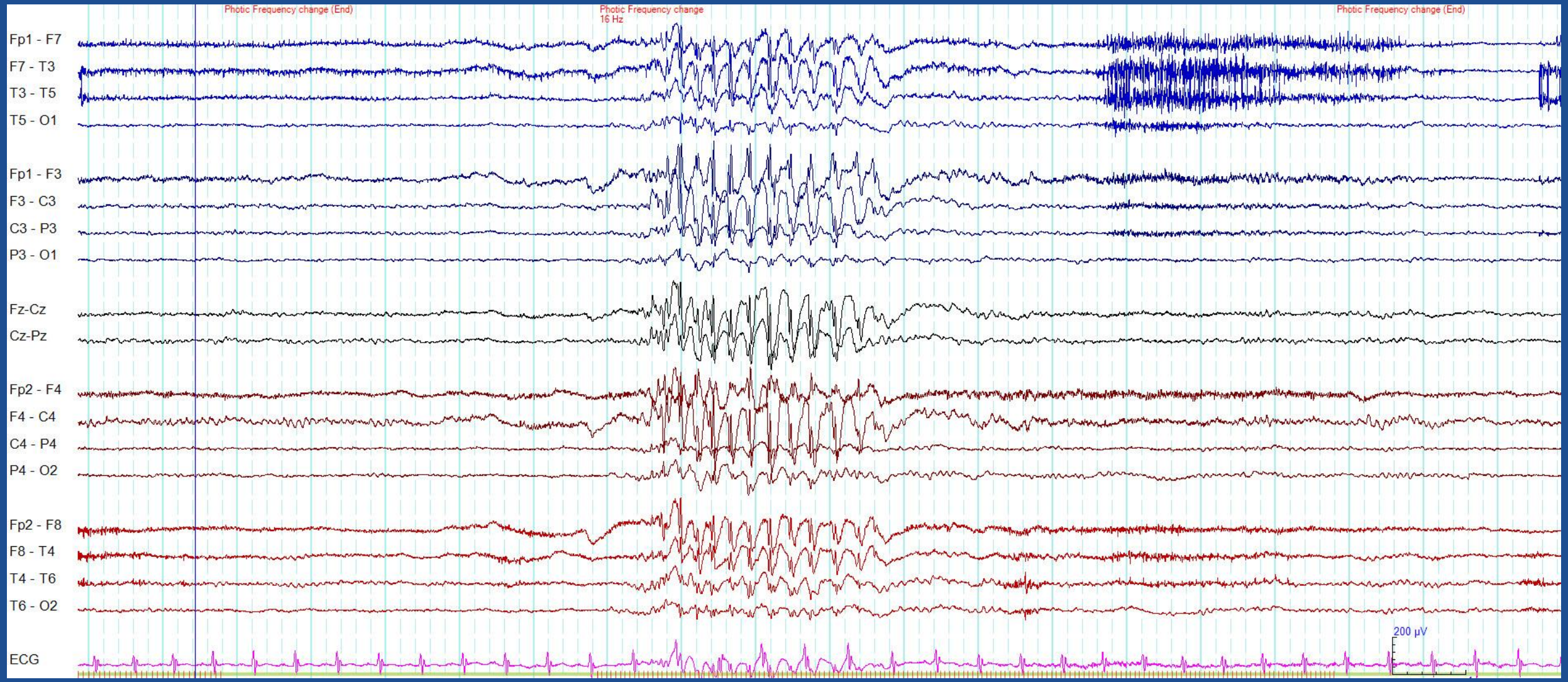
Adolescent/Adult Onset

- A 15-year-old girl with presented with generalized tonic-clonic seizure shortly after waking.

Differential diagnosis

- JME
- GTCa
- JAE


4 Hz SPW complex from left frontal
F7-Ave
T3-Ave
T5-Ave
Fp1-Ave
F3-Ave
C3-Ave
P3-Ave
O1-Ave
Fz-Ave
Cz-Ave
Pz-Ave
Fp2-Ave
F4-Ave
C4-Ave
P4-Ave
O2-Ave
F8-Ave
T4-Ave
T6-Ave
ECG
200 µV


Photic Frequency change (End)
Photic Frequency change
16 Hz
Photic Frequency change (End)
Fp1 - F7
F7 - T3
T3 - T5
T5 - O1
Fp1 - F3
F3 - C3
C3 - P3
P3 - O1
Fz-Cz
Cz-Pz
Fp2 - F4
F4 - C4
C4 - P4
P4 - O2
Fp2 - F8
F8 - T4
T4 - T6
T6 - O2
ECG
200 µV

Idiopathic Generalized Epilepsy Syndrome (IGE)

- Childhood onset
  - CAE
- Adolescent/Adult Onset
  - JME
  - GTCa
  - JAE

Idiopathic Generalized Epilepsy Syndrome (IGE)

- Etiology
  - Polygenic inheritance
  - Genetic susceptibility
  - few cases with monogenic causes reported
- Genetic testing
  - Not routinely indicated for diagnosis
  - Consider in cases with:
    - Intellectual disability
    - Drug-resistant epilepsy
    - Atypical presentation

Generalized Tonic-Clonic Seizures Alone

- Prevalence: Unknown
- **Onset**: 10–25 years (range = 5– 40 years)
- **Seizure: GTC**
- **EEG**: 3– 5.5-Hz generalized spike or polyspike-wave, photoparoxysmal response can be seen.
- **Treatment**: Board spectrum
- Prognosis: usually drug responsive, life-long therapy

Juvenile Myoclonic Epilepsy

- Prevalence: 1-3 per 10 000 persons
- **Onset**: 10–24 years (range = 8– 40 years)
- **Seizure: Myoclonic seizure, GTC, Absence seizure**
- **EEG**: 3– 5.5-Hz generalized spike or polyspike-wave, photoparoxysmal response can be seen.
- **Treatment**: Valproate, avoid sodium channel blocker
- Prognosis: usually drug responsive, life-long therapy

Juvenile Absence Seizures

- Prevalence: 2.4%–3.1% of new onset epilepsy in children and adolescent
- **Onset**: 9–13 years (range = 8– 20 years)
- **Seizure: Absence seizure, GTC**
- **EEG**: 3– 5.5-Hz generalized spike or polyspike-wave, OIRDA is not seen.
- **Treatment**: Valproate
- Prognosis: usually drug responsive, treatment may be required for life.

Idiopathic Generalized Epilepsy Syndrome (IGE)

- Etiology
  - Polygenic inheritance
  - Genetic susceptibility
  - few cases with monogenic causes reported
- Genetic testing
  - Not routinely indicated for diagnosis
  - Consider in cases with:
    - Intellectual disability
    - Drug-resistant epilepsy
    - Atypical presentation

Reference

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PEDIATRIC PRECISION MEDICINE CENTER


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Department of Pediatrics