

Clinical

Record ID

Local Identifier

(Sample identifier as allocated by the group contributing the sample.)

Date of last data collection

(Most recent date that information was collected that has been used to complete the form. Use 01 (Jan) as month or 01 as day if said information is missing.)

Sex

- Male
 Female
 Unknown
 Other

Person completing form

(Clinician or researcher completing the form.)

Clinician responsible for data

Hispanic/Latino status

- Hispanic or Latino origin
 Not of Hispanic or Latino origin
 Unknown
 Not reported

Ethnicity

- Native Hawaiian/other Pacific Islander
 Chinese
 Japanese
 Asian Other
 Black or African American
 Western European
 Eastern European
 French Canadian
 Ashkenazi Jewish
 Sephardic Jewish
 Caucasian other, please specify
 Other/mixed ethnicities, please specify
 Unknown

(Ethnicity category to be selected from list provided. The list of ethnicity categories contains the minimum acceptable categories as per the USA standards.)

Ethnicity comments

Year of birth

Existing exome data Yes
 No
 Unknown
 (applies to exome data sequenced after 1st Jan 2013 only)

Patient deceased No
 Unknown
 Yes (SUDEP)
 Yes (Other epilepsy related - status epilepticus, trauma)
 Yes (Death unrelated to epilepsy)
 Yes (Unknown causes)

Seizure Types

	Yes	No	Unknown
Febrile seizures Seizure of any type (or unknown type) provoked by a documented fever of >38°C/100.4°F	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Aura (simple partial) See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Focal dyscognitive (complex partial) See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Bilateral convulsive See ILAE definition, focal seizure evolving to a bilateral convulsion section	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Focal other seizure types, please specify Focal seizures that do not fit the criteria for an aura or focal dyscognitive seizure or are of unknown type. A focal clonic seizure with retained awareness can be included here.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Other seizure types, please specify Seizures of unknown type or rare cases of generalised seizures	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other seizure types comments

Febrile seizures

Classical febrile seizures

- Yes
 No
 Unknown

(Self-limited convulsive seizures with a documented fever of $>38\text{C}^{\circ}\text{F}/100.4\text{F}^{\circ}\text{F}$ occurring between the age of 6 months and 6 years with no known history of afebrile seizures.)

Age (years) at first occurrence (classic febrile)

(if available)

Age (years) at last occurrence (classic febrile)

(if available)

Other seizures provoked by fever

- Yes
 No
 Unknown

Age (years) at first occurrence of fever provoked seizures

(if available)

Age (years) at last occurrence of fever provoked seizures

(if available)

Age of onset

Age (years) at first occurrence (aura)

(if available)

Age (years) at first occurrence (dyscognitive)

(if available)

Age (years) at first occurrence (bilateral)

(if available)

Age (years) at first occurrence (other)

Age (years) at first seizure

(Minimum of all seizure onsets (computed))

Age (years) of onset correction

(Overrides the age of onset in case type of seizure at onset is not known)

Age of last occurrence

Age (years) at last occurrence (aura)

(if available)

Age (years) at last occurrence (dyscognitive)

(if available)

Age (years) at last occurrence (bilateral)

(if available)

Age (years) at last occurrence (other)

(if available)**Neurological examination**

Neurological examination

- Normal
 Abnormal please specify
 Not done
 Unknown

Neurological examination comments

Investigations

EEG finding 1

- Normal
 Generalized spike and wave, specify frequency
 Generalized polyspike and wave
 Generalized epileptiform unspecified
 Epileptiform unspecified
 Focal or multi-focal epileptiform, specify location
 Focal slowing
 Generalized slowing
 Photo-paroxysmal response
 Other, please specify
 Unknown
 Not done

GSW frequency

- > or = 3Hz
 < 3Hz
 Unknown

 Location of focal epileptiform

- Temporal
- Frontal
- Occipital
- Parietal
- Multi-focal
- Unspecified
- Unknown

(If localization is near the anatomical boundary of two lobes or could reflect one of two sites (e.g. F7, 'fronto-temporal') then both lobes should be selected. If there are two or more independent foci, then select 'multifocal' and the relevant lobes. Centrottemporal localization should be entered as 'temporal' and 'parietal'.)

Type of photoparoxysmal response

- Generalized
 - Occipital
 - Other focal
 - Non-epileptiform
 - Unknown
-

EEG finding 2

- None
 - Generalized spike and wave, specify frequency
 - Generalized polyspike and wave
 - Generalized epileptiform unspecified
 - Epileptiform unspecified
 - Focal or multi-focal epileptiform, specify location
 - Focal slowing
 - Generalized slowing
 - Photo-paroxysmal response
 - Other, please specify
 - Unknown
 - Not done
-

GSW frequency

- > or = 3Hz
 - < 3Hz
 - Unknown
-

Location of focal epileptiform

- Temporal
- Frontal
- Occipital
- Parietal
- Multi-focal
- Unspecified
- Unknown

(If localization is near the anatomical boundary of two lobes or could reflect one of two sites (e.g. F7, 'fronto-temporal') then both lobes should be selected. If there are two or more independent foci, then select 'multifocal' and the relevant lobes. Centrottemporal localization should be entered as 'temporal' and 'parietal'.)

Type of photoparoxysmal response

- Generalized
- Occipital
- Other focal
- Non-epileptiform
- Unknown

EEG finding 3

- None
- Generalized spike and wave, specify frequency
- Generalized polyspike and wave
- Generalized epileptiform unspecified
- Epileptiform unspecified
- Focal or multi-focal epileptiform, specify location
- Focal slowing
- Generalized slowing
- Photo-paroxysmal response
- Other, please specify
- Unknown
- Not done

GSW frequency

- > or = 3Hz
- < 3Hz
- Unknown

Location of focal epileptiform

- Temporal
- Frontal
- Occipital
- Parietal
- Multi-focal
- Unspecified
- Unknown

(If localization is near the anatomical boundary of two lobes or could reflect one of two sites (e.g. F7, 'fronto-temporal') then both lobes should be selected. If there are two or more independent foci, then select 'multifocal' and the relevant lobes. Centrottemporal localization should be entered as 'temporal' and 'parietal'.)

Type of photoparoxysmal response

- Generalized
- Occipital
- Other focal
- Non-epileptiform
- Unknown

Neuroimaging

Neuroimaging performed

- CT
- MRI
- Not done
- CT and MRI
- Unknown

Neuroimaging findings

- Normal
 Nonspecific abnormality, please specify
 Hippocampal sclerosis
 Malformation: Focal Cortical Dysplasia
 Malformation: Heterotopia
 Malformation: Peri-ventricular nodular heterotopia
 Malformation: Polymicrogyria
 Malformation: Lissencephaly
 Malformation: Double Cortex
 Malformation: Hypothalamic Hamartoma
 Malformation: Hemimegaencephaly
 Malformation: Schizencephaly
 Benign Tumor: DNET
 Benign Tumor: Ganglioglioma
 Benign Tumor: unknown
 Traumatic Brain Injury
 Vascular and/or ischemic abnormalities: ischemic stroke
 Vascular and/or ischemic abnormalities: hypoxic ischemic injury
 Vascular and/or ischemic abnormalities: hemorrhage
 Vascular and/or ischemic abnormalities: cerebral angioma
 Other, please specify
 Unknown

Neuroimaging findings comments

Comorbidities

	Yes	No	Unknown
Intellectual Disability	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Autism spectrum disorder	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Psychosis	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Drug resistant Failure of adequate trials of two tolerated and appropriately chosen and used AED schedules (whether as monotherapies or in combination) to achieve sustained seizure freedom (see Kwan, P. et al, Epilepsia 2010)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Degree of intellectual disability

- mild
 moderate
 severe
 profound
 cannot classify

Family History

Reported family history of consanguinity

- Yes
 No
 Unknown

Family History

- Yes
 No
 Unknown
(Family history of any seizures (including febrile) regardless of reported aetiology. Family history refers to any biological relative of the proband, including their offspring.)

First degree relative affected

- Yes
 No
 Unknown
(First-degree relative is defined as the proband's biological mother, father, brother, sister, son or daughter. The relative is regarded as 'affected' if they have any history of seizures (including febrile) regardless of reported aetiology.)

Details of family history of epilepsy

(Additional information about family history, other than that captured in the 'First degree relative affected?' section.)

Epilepsy Syndrome

Focal syndromes

- "Benign" Childhood Epilepsies: Childhood Epilepsy with Centrotemporal Spikes [ILAE Definition]
- "Benign" Childhood Epilepsies: Atypical Childhood Epilepsy with Centrotemporal Spikes [ILAE Definition]
- "Benign" Childhood Epilepsies: Benign Occipital Epilepsy (Panayiotopoulos) [ILAE Definition]
- "Benign" Childhood Epilepsies: Benign Occipital Epilepsy (Gastaut) [ILAE Definition]
- "Benign" Childhood Epilepsies: Idiopathic photosensitive occipital lobe epilepsy [ILAE Definition]
- Other Non-Structural Focal Epilepsies: FrontalNon-lesional epilepsy with clinical and/or EEG evidence of a frontal localization [ILAE Definition]
- Other Non-Structural Focal Epilepsies: FrontotemporalNon-lesional epilepsy with clinical and/or EEG evidence of a frontotemporal localization.]
- Other Non-Structural Focal Epilepsies: TemporalNon-lesional epilepsy with clinical and/or EEG evidence of a temporal localization [ILAE Definition] that does not meet the criteria for Childhood Epilepsy with Centrotemporal Spikes. Mesial temporal lobe epilepsy secondary to hippocampal sclerosis can be included here.
- Other Non-Structural Focal Epilepsies: OccipitalNon-lesional epilepsy with clinical and/or EEG evidence of an occipital localization [ILAE Definition] that does not meet the criteria for any of the benign occipital epilepsy syndromes.
- Other Non-Structural Focal Epilepsies: TemporoccipitalNon-lesional epilepsy with clinical and/or EEG evidence of a temporoccipital localization.
- Other Non-Structural Focal Epilepsies: ParietalNon-lesional epilepsy with clinical and/or EEG evidence of a parietal localization [ILAE Definition]
- Other Non-Structural Focal Epilepsies: MultifocalNon-lesional epilepsy with clinical and/or EEG evidence of more than one localization. Please provide additional details in the Epilepsy Syndrome Comments section.
- Other Non-Structural Focal Epilepsies: UnspecifiedNon-lesional epilepsy with clinical and/or EEG evidence of focal onset but the specific localization is undetermined.
- Structural Focal Epilepsies: Malformation: Focal cortical dysplasiaFocal epilepsy with definite Focal cortical dysplasia of cerebral cortex on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: LissencephalyFocal epilepsy with definite Lissencephaly on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: Subcortical band heterotopiaFocal epilepsy with definite Subcortical band heterotopia on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: Grey matter heterotopiaFocal epilepsy with definite grey matter heterotopia (e.g. PVNH) on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: PolymicrogyriaFocal epilepsy with definite polymicrogyria on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: Hypothalamic HamartomaFocal epilepsy with definite

- Hypothalamic Hamartoma on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: Hemimegalencephaly Focal epilepsy with definite Hemimegalencephaly on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Malformation: Schizencephaly Focal epilepsy with definite Schizencephaly on brain imaging [ILAE Definition]
- Structural Focal Epilepsies: Traumatic Brain Injury Focal epilepsy due to acquired brain injury [ILAE Definition]
- Structural Focal Epilepsies: Stroke Focal epilepsy due to hemorrhagic or ischemic stroke [ILAE Definition]
- Structural Focal Epilepsies: Hypoxic Ischemic Injury Focal epilepsy due to Hypoxic ischemic injury [ILAE Definition]
- Structural Focal Epilepsies: Benign Tumor Focal epilepsy due to benign tumor (ganglioglioma [ILAE Definition] or DNET [ILAE Definition])
- Structural Focal Epilepsies: Vascular malformation: cerebral angioma Focal epilepsy associated with cerebral angioma [ILAE Definition]
- Structural Focal Epilepsies: Mixed epilepsy lesions Focal epilepsy with multiple different epilepsy lesions
- Structural Focal Epilepsies: Other, please specify Use syndrome comments

Evidence for focal epilepsy diagnosis

- Seizure semiology only
- Seizure semiology and EEG
- Seizure semiology and imaging
- Seizure semiology, imaging and histopathology
- Seizure semiology, EEG and imaging
- Not localized (focal unspecified)
- EEG only
- Data not available

Epilepsy Syndrome Comments
