

# Clinical

Record ID

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Local Identifier

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

Date of last data collection

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(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

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(Clinician or researcher completing the form.)

Clinician responsible for data

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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

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Year of birth

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Patient deceased

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

Existing exome data

- Yes  
 No  
 Unknown  
 (Applies to exome data sequenced after 1st Jan 2013 only)

### Generalized Seizure Types See ILAE definition for seizures

**Additional comments about seizure semiology can be added through field comments if desired.**

	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"/>
Generalized Tonic-Clonic See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Myoclonic See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Absence See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Atypical Absence See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Tonic See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Atonic See ILAE definition	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Other seizure types, please specify Generalized seizures of unknown type or rare cases of focal attacks	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other seizure types comments

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### Febrile seizures

Classical febrile seizures

- Yes  
 No  
 Unknown  
 (Self-limited convulsive seizures with a documented fever of >38°C/100.4°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)

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**Age of onset Enter -9 if time of onset in unknown beyond a span of 12month**

Age (years) at first occurrence (Generalized tonic-clonic)	_____
	(if available)
Age (years) at first occurrence (Myoclonic)	_____
	(if available)
Age (years) at first occurrence (Absence)	_____
	(if available)
Age (years) at first occurrence (Atypical absence)	_____
	(if available)
Age (years) at first occurrence (Tonic)	_____
	(if available)
Age (years) at first occurrence (Atonic)	_____
	(if available)
Age (years) at first occurrence (Other)	_____
	(if available)
Age at first seizure (years, excluding classical febrile seizures)	_____
	(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. )

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**Age of last occurrence**

Age (years) at last occurrence (Generalized tonic-clonic)	_____
	(if available)
Age (years) at last occurrence (Myoclonic)	_____
	(if available)
Age (years) at last occurrence (Absence)	_____
	(if available)
Age (years) at last occurrence (Atypical absence)	_____
	(if available)
Age (years) at last occurrence (Tonic)	_____
	(if available)
Age (years) at last occurrence (Atonic)	_____
	(if available)
Age (years) at last occurrence (Other)	_____
	(if available)

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**Neurological examination**

Neurological examination

- Normal  
 Abnormal please specify  
 Not done  
 Unknown

Neurological examination comments

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**Investigations**

EEG finding 1

- Normal  
 Generalized spike and wave, specify frequency  
 Generalized polyspike and wave  
 Generalized paroxysmal fast activity (GPFA)  
 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  
 (Where multiple findings exist, please enter in chronological order. )

GSW frequency

- > or = 3Hz  
 < 3Hz  
 Unknown

Other epileptiform comments

\_\_\_\_\_

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.)

Type of photoparoxysmal response

- Generalized  
 Occipital  
 Other focal  
 Non-epileptiform  
 Unknown

EEG finding 2

- Normal
- Generalized spike and wave, specify frequency
- Generalized polyspike and wave
- Generalized paroxysmal fast activity (GPFA)
- Generalized epileptiform unspecified
- Epileptiform unspecified
- Focal or multi-focal epileptiform, specify location
- Focal slowing
- Generalized slowing
- Photo-paroxysmal response
- Other, please specify
- Unknown

GSW frequency

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

Other epileptiform comments

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.)

Type of photoparoxysmal response

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

EEG finding 3

- Normal
- Generalized spike and wave, specify frequency
- Generalized polyspike and wave
- Generalized paroxysmal fast activity (GPFA)
- Generalized epileptiform unspecified
- Epileptiform unspecified
- Focal or multi-focal epileptiform, specify location
- Focal slowing
- Generalized slowing
- Photo-paroxysmal response
- Other, please specify
- Unknown

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.)

Other epileptiform comments

Type of photoparoxysmal response

- Generalized  
 Occipital  
 Other focal  
 Non-epileptiform  
 Unknown

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## Neuroimaging

Neuroimaging performed

- CT  
 MRI  
 Not done  
 CT and MRI  
 Unknown

Neuroimaging findings

- Normal  
 Abnormal  
 Non-specific abnormality, please specify  
 Other, please specify  
 Unknown

Neuroimaging findings comments

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## 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)

Degree of intellectual disability

- mild
- moderate
- severe
- profound
- cannot classify

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## 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

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

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## Syndrome

Syndrome

- Childhood Absence Epilepsy [ILAE Definition] Childhood Absence Epilepsy with onset between 4 to 9 years
  - Epilepsy with Myoclonic Absences [ILAE Definition]
  - Childhood Absence Epilepsy/ Juvenile Absence Epilepsy overlap Patients that have a clinical picture of Childhood Absence Epilepsy or Juvenile Absence Epilepsy but with onset 9 to 11 years.
  - Juvenile Absence Epilepsy [ILAE Definition] Onset > 11y
  - Juvenile Myoclonic Epilepsy [ILAE Definition] Onset 8 to 20 years
  - Epilepsy with Generalized Tonic-Clonic Seizures Alone [ILAE Definition] Onset 5 to 20 years.
  - Other: Late Onset GGE Generalized epilepsy with onset in adulthood (>20y)
  - Other: Early Onset Absence Epilepsy Absence epilepsy with onset < 4y
  - Other: Epilepsy with Eyelid Myoclonia [ILAE Definition]
  - Other, please specify
  - Generalized unspecified
- (Syndrome definitions are based on current ILAE definitions.)

Epilepsy Syndrome comments

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