

Clinical

Collaborator Participant ID

(ID as given in the sample sheet to Broad)

Local identifier

(Sample identifier as allocated by the group contributing the sample and shipped to Broad institute.)

Broad Sample ID

Mixed case

- Yes
 No
(Tick 'yes' for mixed generalised and focal epilepsy cases only. Cases submitted as mixed must be supported by clear generalised and focal epileptiform activity on EEG. Tick 'no' for GGE cases.)

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

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

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"/>

Focal Seizure Types

	Yes	No	Unknown
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 comments

Febrile seizures

Classical febrile seizures

- Yes
 No
 Unknown

(Self-limited convulsive seizures with a documented fever of $>38^{\circ}\text{C}/100.4^{\circ}\text{F}$ 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)

Were any of these seizures complex febrile seizures?

- Yes
 No
 Unknown

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 Enter -9 if time of onset is unknown beyond a span of 12 month

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 (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)

(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.)

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 (aura)

(if available)

Age (years) at last occurrence (dyscognitive)

(if available)

Age (years) at last occurrence (bilateral)

(if available)

Age (years) at last occurrence (Atonic)

(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 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. Mixed cases must be supported by BOTH generalised AND focal epileptiform activity.)

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

Neuroimaging

Neuroimaging performed

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

Neuroimaging findings

- Normal
- Nonspecific abnormality
- 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.)

Syndrome

For mixed cases: select one generalized and one focal syndrome, or select unclassified mixed epilepsy syndrome and describe in comments section.

Syndrome
Required field

- 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
- Febrile Seizures only Self-limited 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.
- Febrile Seizures Plus (FS+)/Genetic Epilepsy with Febrile Seizures Plus (GEFS+) [ILAE Definition] (Syndrome definitions are based on current ILAE definitions.)

 Syndrome (focal)

Non-lesional epilepsy with clinical and/or EEG evidence of a frontal localization. For mixed epilepsy cases only. Purely focal syndromes should be entered on the focal epilepsy form.

- "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.
- Unclassified mixed epilepsy syndrome with generalized and focal seizures

 Epilepsy Syndrome comments
