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 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
☐ Hispanic
☐ French Canadian
☐ Ashkenazi Jewish
☐ Sephardic Jewish
☐ Caucasian other, please specify
☐ Other/mixed ethnicities, please specify
☐ Unknown

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)

Maternal DNA available

◯ Yes
◯ No
◯ Unknown

Paternal DNA available

◯ Yes
◯ No
◯ Unknown

Existing exome data

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

Birth details and antecedents

Gestational Age

◯ Known
◯ Unknown

Gestational age at birth (weeks) if known

_____

Head circumference at birth

◯ Known
◯ Unknown

Head circumference at birth (cm) if known

_____

Birth weight

◯ Known
◯ Small for gestational age
◯ Unknown

Birth Weight (grams) if known

_____

Head trauma with skull fracture, intracranial bleeding

◯ Yes
◯ No
◯ Unknown

CNS infection

◯ Yes
◯ No
◯ Unknown

Neonatal seizures

◯ Yes
◯ No
◯ Unknown

Normal neonatal period (other than seizures)

◯ Yes
◯ No
◯ Unknown

Neonatal period comments

_____

Other features

Head size

◯ Normal
◯ Large
◯ Small
◯ Unknown

Tone

◯ Hypotonic
◯ Hypertonic
◯ Normal
◯ Unknown

Dysmorphic

◯ Yes, please specify
◯ No
◯ Unknown

Dysmorphic features comments

_____

Movement disorder

◯ Yes, please specify
◯ No
◯ Unknown

Movement disorder comments

_____

Other abnormalities

◯ Yes, please specify
◯ No
◯ Unknown

Other abnormalities comments

_____

Previous genetic analysis

Conventional karyotype

◯ Normal
◯ Abnormal, please specify
◯ Unknown
◯ Finding of unknown significance, please specify
◯ Not done

Conventional karyotype comments

_____

Copy number analysis

◯ Normal
◯ Abnormal, please specify
◯ Unknown
◯ Finding of unknown significance, please specify
◯ Not done

Copy number analysis comments

_____

Gene panel performed

◯ Yes, please specify
◯ No
◯ Unknown

Gene panel details

_____
(Please provide the company and/or panel name. In-house panels can be included as 'in-house'.)

Gene panel results

◯ Normal
◯ Abnormal, please specify
◯ Unknown
◯ Finding of unknown significance, please specify

Gene panel results details

_____

Individual gene testing

◯ Normal
◯ Abnormal, please specify
◯ Unknown
◯ Finding of unknown significance, please specify
◯ Not done

Genetic testing comments

_____

Metabolic testing

◯ Normal
◯ Abnormal, please specify
◯ Unknown
◯ Finding of unknown significance, please specify
◯ Not done

Metabolic testing comments

_____

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	◯	◯	◯
Infantile/epileptic spasmsSee ILAE Definition	◯	◯	◯
TonicSee ILAE definition	◯	◯	◯
AtonicSee ILAE definition	◯	◯	◯
MyoclonicSee ILAE definition	◯	◯	◯
AbsenceSee ILAE definition	◯	◯	◯
Atypical AbsenceSee ILAE definition	◯	◯	◯
Generalized Tonic-ClonicSee ILAE definition	◯	◯	◯
HemiclonicSee ILAE definition, elementary motor section	◯	◯	◯
Bilateral clonicBilateral rhythmic jerking seizure without a tonic component.	◯	◯	◯
Focal seizures of any typeSeizure type to be selected for focal seizures of any type.	◯	◯	◯
Unclassified	◯	◯	◯

Status Epilepticus: convulsiveConvulsive seizure of sustained duration >5 minutes	◯	◯	◯
Status Epilepticus: Non-convulsiveNon-convulsive seizure (generalised or focal) of sustained duration >5 minutes	◯	◯	◯
Other seizure types, please specify	◯	◯	◯

Other seizure types comments

_____

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 in months at first occurrence (classic febrile)

_____
(if available)

Age in years at last occurrence (classic febrile)

_____
(if available)

Other seizures provoked by fever

◯ Yes
◯ No
◯ Unknown
(Any seizure provoked by fever that does not meet the criteria for a "Classical febrile seizure")

Age in months at first occurrence of fever provoked seizures

_____
(if available)

Age in years at last occurrence of fever provoked seizures

_____
(if available)

Age in months at first occurrence

Age in months at first occurrence (Absence)

_____

Age in months at first occurrence (Atonic)

_____

Age in months at first occurrence (Atypical Absence)

_____

Age in months at first occurrence (Bilateral clonic)

_____

Age in months at first occurrence (Focal)

_____

Age in months at first occurrence (Generalized tonic-clonic)

_____

Age in months at first occurrence (Hemiclonic) _____

Age in months at first occurrence (Infantile/epileptic spasms) _____

Age in months at first occurrence (Myoclonic) _____

Age in months at first occurrence (Other) _____

Age in months at first occurrence (Status Epilepticus: convulsive) _____

Age in months at first occurrence (Status Epilepticus: Non-convulsive) _____

Age in months at first occurrence (Tonic) _____

Age in months at first occurrence (Unclassified) _____

Age at first seizure (excluding classical febrile seizures _____
(Minumum of all seizure onsets (computed))

Age in months of onset correction _____
(Overrides the age of onset in case type of seizure at onset is not known)

Age in years at last occurrence

Age in years at last occurrence (Absence) _____

Age in years at last occurrence (Atonic) _____

Age in years at last occurrence (Atypical Absence) _____

Age in years at last occurrence (Bilateral clonic) _____

Age in years at last occurrence (Focal) _____

Age in years at last occurrence (Generalized tonic-clonic) _____

Age in years at last occurrence (Hemiclonic) _____

Age in years at last occurrence (Infantile/epileptic spasms) _____

Age in years at last occurrence (Myoclonic) _____

Age in years at last occurrence (Other) _____

Age in years at last occurrence (Status Epilepticus: convulsive) _____

Age in years at last occurrence (Status Epilepticus: Non-convulsive) _____

Age in years at last occurrence (Tonic) _____

Age in years at last occurrence (Unclassified) _____

Neurological examination

Neurological examination

- ◯ Normal
- ◯ Abnormal please specify
- ◯ Not done
- ◯ Unknown

Neurological examination comments _____

Investigations

EEG finding 1

- ◯ Normal
- ◯ Burst suppression
- ◯ Classic hypsarrhythmia
- ◯ Hypsarrhythmia variant
- ◯ Generalized spike and wave, specify frequency
- ◯ Generalized polyspike and wave
- ◯ Generalized paroxysmal fast activity (GPFA)
- ◯ Continuous Spike and Wave in slow-wave Sleep (CSWS)
- ◯ 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

Other epileptiform comments

_____

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.)

Type of photoparoxysmal response

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

EEG finding 2

◯ Normal
◯ Burst suppression
◯ Classic hypsarrhythmia
◯ Hypsarrhythmia variant
◯ Generalized spike and wave, specify frequency
◯ Generalized polyspike and wave
◯ Generalized paroxysmal fast activity (GPFA)
◯ Continuous Spike and Wave in slow-wave Sleep (CSWS)
◯ Generalized epileptiform unspecified
◯ Epileptiform unspecified
◯ Focal or multi-focal epileptiform, specify location
◯ Focal slowing
◯ Generalized slowing
◯ Photo-paroxysmal response
◯ Other, please specify
◯ Unknown

Other epileptiform comments

_____

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.)

Type of photoparoxysmal response

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

EEG finding 3

◯ Normal
◯ Burst suppression
◯ Classic hypsarrhythmia
◯ Hypsarrhythmia variant
◯ Generalized spike and wave, specify frequency
◯ Generalized polyspike and wave
◯ Generalized paroxysmal fast activity (GPFA)
◯ Continuous Spike and Wave in slow-wave Sleep (CSWS)
◯ Generalized epileptiform unspecified
◯ Epileptiform unspecified
◯ Focal or multi-focal epileptiform, specify location
◯ Focal slowing
◯ Generalized slowing
◯ Photo-paroxysmal response
◯ Other, please specify
◯ Unknown

Other epileptiform comments

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.)

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
- ◯ Malformations: Focal Cortical Dysplasia
- ◯ Malformations: Heterotopia
- ◯ Malformations: Peri-ventricular nodular heterotopia
- ◯ Malformations: Polymicrogyria
- ◯ Malformations: Pachygyria
- ◯ Malformations: Hemimegalencephaly
- ◯ Malformations: Schizencephaly
- ◯ Malformations: Lissencephaly
- ◯ Malformations: Double Cortex
- ◯ Malformations: Holoprosencephaly
- ◯ Malformations: Corpus callosum agenesis/dysplasia
- ◯ Malformations: Septo-optic dysplasia
- ◯ Malformations: other
- ◯ Vascular and/or ischemic abnormalities: hypoxic ischemic injury
- ◯ Vascular and/or ischemic abnormalities: Periventricular leukomalacia
- ◯ Vascular and/or ischemic abnormalities: hemorrhage
- ◯ Other: Hippocampal Sclerosis
- ◯ Other: Porencephaly
- ◯ Other: Hydrocephalus
- ◯ Other: Atrophy
- ◯ Other, please specify
- ◯
- ◯ Non-specific abnormality, please specify
- ◯ Unknown

Additional Neuroimaging abnormality 1

- ◯ None
- ◯ Malformations: Focal Cortical Dysplasia
- ◯ Malformations: Heterotopia
- ◯ Malformations: Peri-ventricular nodular heterotopia
- ◯ Malformations: Polymicrogyria
- ◯ Malformations: Pachygyria
- ◯ Malformations: Hemimegalencephaly
- ◯ Malformations: Schizencephaly
- ◯ Malformations: Lissencephaly
- ◯ Malformations: Double Cortex
- ◯ Malformations: Holoprosencephaly
- ◯ Malformations: Corpus callosum agenesis/dysplasia
- ◯ Malformations: Septo-optic dysplasia
- ◯ Malformations: other
- ◯ Vascular and/or ischemic abnormalities: hypoxic ischemic injury
- ◯ Vascular and/or ischemic abnormalities: Periventricular leukomalacia
- ◯ Vascular and/or ischemic abnormalities: hemorrhage
- ◯ Other: Hippocampal Sclerosis
- ◯ Other: porencephaly
- ◯ Other: hydrocephalus
- ◯ Other: atrophy
- ◯ Other, please specify
- ◯
- ◯ Non-specific abnormality, please specify
- ◯ Unknown

Additional Neuroimaging abnormality 2

- ◯ None
- ◯ Malformations: Focal Cortical Dysplasia
- ◯ Malformations: Heterotopia
- ◯ Malformations: Peri-ventricular nodular heterotopia
- ◯ Malformations: Polymicrogyria
- ◯ Malformations: Pachygyria
- ◯ Malformations: Hemimegalencephaly
- ◯ Malformations: Schizencephaly
- ◯ Malformations: Lissencephaly
- ◯ Malformations: Double Cortex
- ◯ Malformations: Holoprosencephaly
- ◯ Malformations: Corpus callosum agenesis/dysplasia
- ◯ Malformations: Septo-optic dysplasia
- ◯ Malformations: other
- ◯ Vascular and/or ischemic abnormalities: hypoxic ischemic injury
- ◯ Vascular and/or ischemic abnormalities: Periventricular leukomalacia
- ◯ Vascular and/or ischemic abnormalities: hemorrhage
- ◯ Other: Hippocampal Sclerosis
- ◯ Other: porencephaly
- ◯ Other: hydrocephalus
- ◯ Other: atrophy
- ◯ Other, please specify
- ◯
- ◯ Non-specific abnormality, please specify
- ◯ Unknown

Neuroimaging findings comments

_____

Comorbidities

	Yes	No	Unknown
Developmental delay prior to seizure onset	◯	◯	◯
Regression/plateau	◯	◯	◯
Intellectual Disability	◯	◯	◯
Autism spectrum disorder	◯	◯	◯
Psychosis	◯	◯	◯
Drug resistantFailure 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)	◯	◯	◯

Type of delay

- ◯ Motor
- ◯ Speech and Language
- ◯ Unknown
- ◯ Global

Age at regression in months if known

_____

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

Syndrome

- ☐ Neonatal onset: Ohtahara syndrome [ILAE Definition]
- ☐ Neonatal onset: Early myoclonic encephalopathy (EME) [ILAE Definition]
- ☐ Early onset epileptic encephalopathy (< 3 months) Epileptic encephalopathy with seizure onset of less than 3 months of age that does not meet the criteria for any other early onset epileptic encephalopathy.
- ☐ Infantile onset epileptic encephalopathy (not otherwise specified) Epileptic encephalopathy with seizure onset between 3 and 12 months of age that does not meet the criteria for any other infantile onset epileptic encephalopathy.
- ☐ Epilepsy of infancy with migrating focal seizures [ILAE Definition]
- ☐ West syndrome/infantile spasms [ILAE Definition]
- ☐ Late-onset epileptic spasms [ILAE Definition] Onset >1y
- ☐ Lennox-Gastaut syndrome [ILAE Definition]
- ☐ Epilepsy with myoclonic atonic seizures [ILAE Definition]
- ☐ Dravet syndrome [ILAE Definition]
- ☐ Landau-Kleffner syndrome (LKS) [ILAE Definition]
- ☐ Epileptic encephalopathy with continuous spike-and-wave during sleep (CSWS) [ILAE Definition]
- ☐ Febrile Infection Related Epilepsy Syndrome (FIRES) [ILAE Definition]
- ☐ Hemiconvulsion-Hemiplegia-Epilepsy Epilepsy with hemispheric atrophy secondary to a prolonged focal motor seizure in infancy, usually during a febrile illness. Hemiplegia is also present (see Tenney, J.R. et al, Neurology 2012).
- ☐ Nonsyndromic epileptic encephalopathy with focal seizures Epileptic encephalopathy with predominantly focal seizures that does not meet the criteria for any epileptic encephalopathy syndrome.
- ☐ Nonsyndromic epileptic encephalopathy with generalized seizures Epileptic encephalopathy with predominantly generalized seizures that does not meet the criteria for any epileptic encephalopathy syndrome.
- ☐ Nonsyndromic epileptic encephalopathy with mixed or unclassified seizures Epileptic encephalopathy with mixed or unclassified seizure types that does not meet the criteria for any epileptic encephalopathy syndrome.

Comments for multiple syndromes

_____

Epilepsy syndrome comments

_____

(In cases where an evolution has occurred, multiple syndromes should be selected and a comment made.)