**Epi25 GGE/Mixed REDCap Database**

*Glossary*

|  |  |
| --- | --- |
| Absence | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/absence-typical-overview.html> Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Age at first occurrence | Age of onset in years of that particular seizure type if known. If unknown, leave the field blank. This information is used to support the assignment of a specific epilepsy syndrome and will be reviewed as part of data quality control checking. |
| Age at first seizure (of any type, excluding classical febrile seizures) | Age of onset in years of any unprovoked seizure, including an unclassified seizure or seizure of unknown type but excluding classical febrile seizures. Value is computed as the minimum of all seizure onsets. Age at first seizure can be overridden manually in the “Age (years) of onset correction” field. This may be used if the seizure type at onset is unknown. |
| Age at last occurrence | Age at last occurrence in years of that particular seizure type if known. If unknown, leave the field blank. If seizure type is ongoing, enter age at last known occurrence. This information is collected to give an indication of epilepsy severity and drug responsiveness. This data may be reviewed as part of data quality control checking, especially in the context of the drug resistance field, or in the case of a genomic finding. |
| Atonic | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/atonic-overview.html>  Additional comments about seizure semiology can be added through REDCap if desired. |
| Atypical absence | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/absence-atypical-overview.html> Additional comments about seizure semiology can be added through REDCap if desired. |
| Aura (simple partial)\* | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/aura-overview.html> Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Autism Spectrum Disorder | To be selected ‘Y’ where a clear diagnosis of Autism Spectrum Disorder has been made. Otherwise, ‘Unknown’ can be selected. Additional comments about co-morbidities can be added through REDCap if required (see REDCap instructions). |
| Bilateral convulsive\* | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/motor-overview> in ‘Focal seizure evolving to a bilateral convulsion’ section. Additional comments about seizure semiology can be added through REDCap if desired. |
| Broad Sample ID | ID submitted to Broad Institute with samples. |
| Classical febrile seizures | 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. Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Clinician responsible for data | Clinician/Investigator who will be the contact person for the duration of the Epi25 study for any queries relating to the phenotypic data. |
| Complex febrile seizures | A febrile seizure with (a) focal onset, (b) prolonged duration (lasting ≥10 minutes), or (c) multiple seizures within the same febrile illness (Berg & Shinnar, 1996). |
| Details of family history of epilepsy | Free text field for comments about the family history of epilepsy/seizures. The level of detail provided is at the discretion of the person entering the data and can include the type of relationship of the affected individuals to the proband and any information about their type of epilepsy, seizure semiology, onset age, severity etc. This information will not be used for group analyses but may be reviewed in the case of a genomic finding. |
| 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* (>12 months) seizure freedom (*Epilepsia.* 2010 Jun;51(6):1069-77. doi: 10.1111/j.1528-1167.2009.02397.x. Epub 2009 Nov 3. Kwan P et al.). |
| EEG findings | Finding to be selected from list provided. Where ‘Generalized spike and wave, specify frequency’ is selected, ‘GSW frequency’ will need to be completed. Where ‘Photo-paroxysmal response’ is selected, ‘Type of photo-paroxysmal response’ will need to be completed. A ‘normal’ EEG result should only be entered in ‘EEG finding 1’ if the patient has never had any documented EEG abnormalities. If only one EEG abnormality has ever been documented, leave EEG finding 2 blank. The form allows for three EEG findings – please select the abnormalities that best represent and support the final syndrome selected. Where multiple findings exist, please enter in chronological order. Additional information about EEG, including age at EEG if deemed important, can be added through REDCap. |
| Epilepsy syndrome comments | Free text field for comments about the epilepsy syndrome if deemed important, for example a comment about unusual features. This field is required only if ‘Other, please specify’ is selected. This information will not be used for group analyses but may be reviewed as part of data quality control checking or in the case of a genomic finding. |
| Evidence for focal epilepsy diagnosis\* | Evidence for the localization used to assign the specific focal syndrome component of the mixed case. This information may be used as an indicator of the certainty of the diagnosis and could be reviewed as part of data quality control checking. |
| Febrile seizures | Seizure of any type (or unknown type) provoked by a documented fever of >38°C/100.4°F. Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Focal dyscognitive (complex partial)\* | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/dyscognitive-overview.html>  Additional comments about seizure semiology can be added through REDCap if desired. |
| Focal other \* | Seizure type category to be used for 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. |
| Generalized Tonic-Clonic | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/convulsive-overview.html>  Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Intellectual Disability | To be selected ‘Y’ where a clear diagnosis of intellectual disability has been made. Otherwise, ‘Unknown’ can be selected. Additional comments about co-morbidities can be added through REDCap if required (see REDCap instructions). |
| Local identifier | Existing sample identifier or code allocated by the group contributing the sample. |
| Mixed case | Tick ‘yes’ for mixed epilepsy cases with evidence for both generalized and focal seizures. Ideally, this should be both clinical and EEG evidence for the two types. Where the clinical evidence for both types is equivocal, but the EEG data clear-cut, these cases can be included. Tick ‘no’ to enter a purely GGE case. Please refer to [eligibility criteria](http://epi-25.org/epi25-eligibility-criteria) for mixed cases. |
| Myoclonic | See ILAE definition:  <https://www.epilepsydiagnosis.org/seizure/myoclonic-overview.html>  Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Neuroimaging findings | Select neuroimaging findings from drop-down menu if performed. ‘Normal indicates’ no abnormality. ‘Non-specific abnormality’ indicates a neuroimaging change that is not believed to be related to the patient’s epilepsy. ‘Other, please specify’ is for other findings that may be entered in the free text neuroimaging findings comments box. |
| Neuroimaging findings comments | Comments can include type of abnormality, location in the brain, age at scan or any other details deemed important. To be completed where ‘Abnormal’, ‘Nonspecific abnormality, please specify’ or ‘Other, please specify’ are selected. |
| Neuroimaging performed | Type of imaging technology used to be selected from the list. Otherwise, ‘Not done’ can be selected. Information about data acquisition, for example MRI tesla, can be added as a comment through REDCap if desired (see REDCap instructions). |
| Neurological examination comments | Please provide information about abnormal neurological findings. This information will not be used for group analyses but may be reviewed as part of data quality control checking or in the case of a genomic finding. |
| Other seizure types comments | Please provide information about the ‘other seizure type’, including details of seizure semiology and why the seizure type is unclassifiable if applicable. |
| Other seizure types, please specify | Seizure type category to be used for generalized seizures of unknown type or rare cases of focal attacks. |
| Other seizures provoked by fever | Any seizure provoked by a documented fever of >38°C/100.4°F that does not meet the criteria for a ‘Classical febrile seizure’. Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Patient deceased | To be selected from list. SUDEP (Sudden Unexpected Death in Epilepsy) is a sudden, unexpected, non-traumatic and non-drowning death in someone with epilepsy in whom post-mortem examination is unrevealing. |
| Psychosis | To be selected ‘Y’ where a clear history of psychosis is present. Otherwise, ‘Unknown’ can be selected. Additional comments about co-morbidities can be added through REDCap if required. |
| Syndrome | Syndrome definitions are based on current ILAE definitions.  *Childhood Absence Epilepsy:* <https://www.epilepsydiagnosis.org/syndrome/cae-overview.html> with onset 4 to 9 years.  *Epilepsy with Myoclonic Absences*: <https://www.epilepsydiagnosis.org/syndrome/epilepsy-myoclonic-absences-overview.html>  *Childhood Absence Epilepsy/ Juvenile Absence Epilepsy:*  Patients that have a clinical picture of Childhood Absence Epilepsy or Juvenile Absence Epilepsy but with onset 9 to 11 years.  *Juvenile Absence Epilepsy:* <https://www.epilepsydiagnosis.org/syndrome/jae-overview.html> with onset >11 years.  *Juvenile Myoclonic Epilepsy:*  <https://www.epilepsydiagnosis.org/syndrome/jme-overview.html> with onset 8 to 20 years.  *Epilepsy with Generalized Tonic-Clonic Seizures Alone:*  <https://www.epilepsydiagnosis.org/syndrome/egtcsa-overview.html> with onset 5 to 20 years.  *Other: Late Onset Genetic Generalized Epilepsy*  Generalized epilepsy with onset in adulthood (>20 years old)  *Other: Early Onset Absence Epilepsy*  <https://www.epilepsydiagnosis.org/syndrome/cae-overview.html> with onset under 4 years old.  *Other: Epilepsy with Eyelid Myoclonia*  <https://www.epilepsydiagnosis.org/syndrome/emwa-overview.html>  *Other: please specify*  Other generalized epilepsy syndrome not on syndrome list.  *Generalized unspecified*  Generalized epilepsy that does not meet all the criteria for any specific sub-syndrome.  *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+)*  <https://www.epilepsydiagnosis.org/syndrome/fbp-overview.html> |
| Syndrome (focal)\* | Focal syndromes for mixed cases where a clear generalized and focal syndrome exist.  Syndrome definitions are based on current ILAE definitions.  *“Benign” Childhood Epilepsies: Childhood Epilepsy with Centrotemporal Spikes:* <https://www.epilepsydiagnosis.org/syndrome/ects-overview.html>  *“Benign” Childhood Epilepsies: Atypical Childhood Epilepsy with Centrotemporal Spikes:* <https://www.epilepsydiagnosis.org/syndrome/atypical-ects-overview.html>  *“Benign” Childhood Epilepsies: Benign Occipital Epilepsy (Panayiotopoulos):* <https://www.epilepsydiagnosis.org/syndrome/panayiotopoulos-overview.html>  *“Benign” Childhood Epilepsies: Benign Occipital Epilepsy (Gastaut):* <https://www.epilepsydiagnosis.org/syndrome/late-childhood-occipital-overview.html>  *“Benign” Childhood Epilepsies: Idiopathic photosensitive occipital lobe epilepsy:* <https://www.epilepsydiagnosis.org/syndrome/idiophatic-pole-overview.html>  *Other Non-lesional Focal Epilepsies: Frontal:*  Non-lesional epilepsy with clinical and/or EEG evidence of a frontal localization (<https://www.epilepsydiagnosis.org/seizure/frontal-lobe-overview.html>).  *Other Non-lesional Focal Epilepsies: Frontotemporal:*  Non-lesional epilepsy with clinical and/or EEG evidence of a frontotemporal localization.  *Other Non-lesional Focal Epilepsies: Temporal:*  Non-lesional epilepsy with clinical and/or EEG evidence of a temporal localization (<https://www.epilepsydiagnosis.org/seizure/temporal-overview.html>) 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-lesional Focal Epilepsies: Occipital:*  Non-lesional epilepsy with clinical and/or EEG evidence of an occipital localization (<https://www.epilepsydiagnosis.org/seizure/occipital-overview.html>) that does not meet the criteria for any of the benign occipital epilepsy syndromes.  *Other Non-lesional Focal Epilepsies: Temporoccipital:*  Non-lesional epilepsy with clinical and/or EEG evidence of a temporoccipital localization.  *Other Non-lesional Focal Epilepsies: Parietal:*  Non lesional epilepsy with clinical and/or EEG evidence of a parietal localization (<https://www.epilepsydiagnosis.org/seizure/parietal-overview.html>)  *Other Non-lesional Focal Epilepsies: Multifocal:*  Non-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-lesional Focal Epilepsies: Unspecified*  Non-lesional epilepsy with clinical and/or EEG evidence of focal onset but the specific localization is undetermined. |
| Tonic | See ILAE definition: <https://www.epilepsydiagnosis.org/seizure/tonic-overview.html>  Additional comments about seizure semiology can be added through REDCap if desired (see REDCap instructions). |
| Unclassified mixed epilepsy syndrome with generalized and focal seizures\* | This is to be selected for mixed cases where there is not a clear-cut generalized and focal syndrome, but a mixed epilepsy case clear generalized and focal seizures supported by generalized and focal epileptiform activity on EEG. Please refer to [eligibility criteria](http://epi-25.org/epi25-eligibility-criteria) for mixed cases. |
| Year of birth | Year of birth. USA PHI regulations allow YOB only (http://cphs.berkeley.edu/hipaa/hipaa18.html) |

\*Fields that appear only when entering a mixed case.