EGI Clinical Data Collection Form Cover Page

Please find enclosed the EGI Clinical Data Form for my patient.

This form was completed by: ________________________________

On (date): ________________________________

For Office/EGI Use Only:

Study/EGI Identifier: ________________________________

Date Received: ________________________________

Received by: ________________________________

Columbia IRB-AAA05656
Patient Name: ____________________________________________

First    Middle    Last

Date of Birth: ____________________________  Current Age: ______________

MM/DD/YYYY

Gender:  □ Male    □ Female

PROBAND  □ Mother  □ Father  □ Other: ____________________________ (specify)

Select One

Hispanic or Latino:  □ Yes  □ No  Ashkenazi Jewish:  □ Yes  □ No

Race (select all that apply):

□ African American/Black   □ Native American/Alaskan Native
□ Asian   □ Native Hawaiian/Pacific Islander
□ Caucasian/White   □ Other

Physician who ordered Clinical/Research Sequencing: ____________________________

Name

_________________________    ____________________________    ____________________________

Phone    Fax    Email

Lab of Clinical/Research Sequencing: ____________________________

Lab Name

_________________________    ____________________________    ____________________________

Country    State/Province    Phone (if known)

Sequencing lab Patient ID number: ____________________________

EGI Referral Source:  □ CURE/EGI website  □ EGI Facebook/Twitter
□ EGI eNewsletter  □ Media source (news article, etc.)  □ Doctor Referral
□ Advocacy organization: ____________________________  □ Other: ____________________________

For Office/EGI Use Only:

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Received by: ____________________________  Columbia IRB-AAA05656
Family History

Family History of Epilepsy: □ Yes □ No □ Unknown

Number of 1st Degree Relatives Affected (specify numbers below):

_____ Mother  _____ Father

_____ Brother(s) _____ Sister(s)

Additional Relatives with Epilepsy: □ Yes □ No □ Unknown

If yes, details: ___________________________________________________________________________________

Family History of Congenital Abnormality, chromosomal abnormality, movement disorder, or intellectual disability: □ Yes □ No □ Unknown

If yes, details:

<table>
<thead>
<tr>
<th>Number of Relatives</th>
<th>Specify Relationship(s)</th>
<th>Describe Abnormality</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
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</tbody>
</table>

Family history of other known or suspected Genetic Condition:

□ Yes □ No □ Unknown

If yes, details:

<table>
<thead>
<tr>
<th>Number of Relatives</th>
<th>Specify Relationship(s)</th>
<th>Describe Abnormality</th>
</tr>
</thead>
<tbody>
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</tbody>
</table>

Family history of consanguinity*:

□ Yes □ No □ Unknown

*Are the parents related by blood?
**Birth and Neonatal History**

<table>
<thead>
<tr>
<th></th>
<th>____________ wks</th>
<th>Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gestational Age at Birth</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Birth Weight</td>
<td>______ lbs ______ oz</td>
<td>Unknown</td>
</tr>
<tr>
<td>Head Circumference @birth (if known)</td>
<td>____________ cm</td>
<td>Unknown</td>
</tr>
<tr>
<td>Neonatal Seizures</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Evidence of Neonatal Encephalopathy</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Pre- or perinatal complications:</td>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>

Describe pre- or perinatal complications here: ____________________________

**Epilepsy History**

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>Febrile Seizures</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Febrile status epilepticus</td>
<td></td>
<td></td>
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<tr>
<td>Intracranial surgery:</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
### CNS Infection:  
- [ ] Yes
- [ ] No
- [ ] Unknown

### Degenerative disorders:  
- [ ] Yes
- [ ] No
- [ ] Unknown

### Metabolic disorders (e.g. PKU, mitochondrial encephalopathy):  
- [ ] Yes
- [ ] No
- [ ] Unknown

### Head Trauma w/skull fracture, intracranial bleeding or loss of consciousness longer than 15 minutes:  
- [ ] Yes
- [ ] No
- [ ] Unknown

### Confirmed/Suspicion for autoimmune epilepsy  
- [ ] Yes
- [ ] No
- [ ] Unknown

### Age at first unprovoked seizure  
- [ ] Years
- [ ] Months

#### Seizure types (choose all that apply):
- [ ] Infantile/epileptic spasms
- [ ] Tonic
- [ ] Atonic
- [ ] Myoclonic
- [ ] Typical absence
- [ ] Atypical absence
- [ ] Generalized tonic clonic (GTC)
- [ ] Focal seizures
- [ ] Focal seizures with secondary generalization
- [ ] Unclassifiable convulsive seizure
- [ ] Status Epilepticus:
  - [ ] Convulsive
  - [ ] Non-convulsive
(Seizure Types, cont’d)

☐ Non-convulsive seizures
☐ Reflex seizures
  ☐ Photosensitive
  ☐ Other: ____________________________________________

☐ Other seizure type: ____________________________________________
☐ Unknown

EEG Results (choose all that apply over the patient’s course):

☐ Normal
☐ Classic hypsarrhythmia
☐ Hypsarrhythmia variant
☐ Generalized spike-wave
☐ Generalized paroxysmal fast activity (GPFA)
☐ Slow or disorganized for age
☐ Focal sharp waves/spikes
☐ Multi-focal sharp waves/spikes
☐ Focal slowing
☐ Focal attenuation
☐ ESES (electrical status epilepticus of sleep)
☐ Photoparoxysmal response
☐ Other: ____________________________________________
☐ Unknown

Neuro-imaging:  
  MRI  ☐ Yes  ☐ No
  CT  ☐ Yes  ☐ No

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Neuro-imaging results (Select all that apply):

- Normal

- Malformations:
  - Focal Cortical Dysplasia
  - Heterotopia
  - Peri-ventricular nodular heterotopia
  - Polymicrogyria
  - Pachygyria
  - Hemimegalencephaly
  - Schizencephaly
  - Lissencephaly
  - Double cortex
  - Holoprosencephaly
  - Corpus callosum agenesis/dysplasia
  - Septo-optic dysplasia
  - Hypothalamic hamartoma
  - Other hamartoma
  - Cerebellar malformation
  - Other: ________________________________

- Neurocutaneous syndrome:
  - Tuberous sclerosis complex
  - Sturge-Weber syndrome
  - Other: ________________________________

(Neuroimaging Malformations continued on next page)
(Neuroimaging Malformations, cont’d)

☐ Vascular and/or ischemic abnormalities:
  ○ Hypoxic ischemic injury
  ○ Periventricular leukomalacia
  ○ Intraventricular hemorrhage
  ○ Arterial infarction
  ○ Watershed infarction
  ○ Venous thrombosis or infarction
  ○ Intraparenchymal hemorrhage
  ○ Vascular malformation
  ○ Vasculitis
  ○ Porencephaly
  ○ Vascular subarachnoid hemorrhage
  ○ Other: ____________________________

☐ Trauma:
  ○ Subdural hematoma
  ○ Epidural hematoma
  ○ Cerebral contusion
  ○ Penetrating head trauma
  ○ Traumatic subarachnoid hemorrhage
  ○ Other: ____________________________

☐ Tumor:
  ○ Glioma
  ○ Other glial tumor
  ○ Meningioma
  ○ Dysembryoplastic neuroepithelial tumor
  ○ Other: ____________________________

(Neuroimaging Malformations continued on next page)
(Neuroimaging Malformations, cont’d)

☐ Other:
   ○ Hydrocephalus
   ○ Diffuse Atrophy
   ○ Focal Atrophy
   ○ MTS
   ○ Other: ________________________________

☐ Unknown

Development / Cognition
Type of Delay:
☐ None
☐ Gross motor
☐ Fine motor
☐ Language
☐ Personal-social
☐ Global
☐ Intellectual disability
   ○ Mild
   ○ Moderate
   ○ Severe
   ○ Profound
   ○ Cannot classify

☐ Learning difficulty
Delay prior to seizure onset: ☐ Yes ☐ No ☐ Unknown
Regression or plateau (specify which): ☐ Yes ☐ No ☐ Unknown
IQ or DQ (score, if available; specify which): ________________________ ☐ Unknown
Autism spectrum disorder: ☐ Yes ☐ No ☐ Unknown

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Date Received: __________________
Received by: ________________________________
Columbia IRB-AAAA5656
### Other Features

<table>
<thead>
<tr>
<th>Condition</th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>Microcephaly</td>
<td></td>
<td></td>
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<tr>
<td>If yes, list head circumference and age measured:</td>
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<tr>
<td>Macrocephaly</td>
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<tr>
<td>If yes, list head circumference and age measured:</td>
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<tr>
<td>Abnormal Tone / Hypotonia</td>
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<tr>
<td>Abnormal Tone / Spasticity</td>
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<tr>
<td>Weakness</td>
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<tr>
<td>Movement Disorder</td>
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<tr>
<td>Ataxia</td>
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<tr>
<td>Autonomic Abnormality</td>
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<tr>
<td>Psychiatric Disorder</td>
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<tr>
<td>Non-epileptic/psychogenic seizures</td>
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<tr>
<td>(EEG confirmed)</td>
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<tr>
<td>Vision Abnormality</td>
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<tr>
<td>Hearing Abnormality</td>
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<tr>
<td>Dysmorphic Features</td>
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<tr>
<td>Constitutional Abnormality</td>
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<td>Cutaneous Abnormality</td>
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<td>Cardiac Abnormality</td>
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<td>Renal Abnormality</td>
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<tr>
<td>Endocrine Abnormality</td>
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<tr>
<td>Cancer Diagnosis</td>
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<tr>
<td>Abnormality of other organs</td>
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</tbody>
</table>

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Received by: ________________________________  Columbia IRB-AAAO5656

Page 10 of 15
Describe any abnormalities here:____________________________________________________

____________________________________________________

Abnormal Genetic or Metabolic Testing
Abnormal genetic testing (e.g. karyotype, microarray):
☐ Yes     ☐ No     ☐ Unknown     ☐ No genetic testing done

Describe any abnormalities here:____________________________________________________

____________________________________________________

Abnormal metabolic testing:
☐ Yes     ☐ No     ☐ Unknown     ☐ No metabolic testing done

Describe any abnormalities here:____________________________________________________

____________________________________________________

For Office/EGI Use Only:
Study/EGI Identifier:_______________________________ Date Received:_____________________

Received by:__________________________________________________ Columbia IRB-AAAO5656
Epilepsy Classification, Syndrome, Etiology and Outcome:
Adequate information to classify epilepsy syndrome:  ☐ Yes  ☐ No
Select Syndrome (if applicable):
☐ Neonatal onset
  ☐ Self-limited (benign) familial neonatal epilepsy (BFNE)
  ☐ Self-limited (benign) neonatal seizures (BNS)
  ☐ Ohtahara syndrome
  ☐ Early myoclonic encephalopathy (EME)
  ☐ Developmental delay, epilepsy and neonatal diabetes (DEND)
  ☐ Hyperekplexia
☐ Usually onset less than age 2 years
  ☐ Febrile seizures (FS)
  ☐ Febrile seizures Plus (FS+)
  ☐ Self-limited (benign) infantile epilepsy
  ☐ Self-limited (benign) familial infantile epilepsy
  ☐ Self-limited (benign) familial neonatal-infantile seizures (BFNIS)
  ☐ Dravet syndrome
  ☐ West syndrome/infantile spasms
  ☐ Epilepsy of infancy with migrating focal seizures
  ☐ Myoclonic epilepsy in infancy (MEI)
  ☐ Early onset epileptic encephalopathy (< 3 months)
  ☐ Infantile onset epileptic encephalopathy (not fitting into specific syndrome above)
  ☐ Coppola Syndrome/migrating partial seizures of infancy
  ☐ Aicardi Syndrome
  ☐ Myoclonic-Atonic epilepsy (Doose Syndrome)
  ☐ Myoclonic-Absence epilepsy (Tassinari syndrome)

(Epilepsy Classification continued on next page)
(Epilepsy Classification, cont’d)

- Usually onset older than age 2 years
  - Febrile seizures (FS)
  - Febrile seizures (FS) Plus+
  - Self-limited (benign) epilepsy with centrotemporal spikes (BECTS)
  - Mesial temporal lobe epilepsy with hippocampal sclerosis (MTLE with HS)
  - Early onset childhood occipital epilepsy (Panayiotopoulos type)
  - Late onset childhood occipital epilepsy (Gastaut type)
  - Autosomal-dominant nocturnal frontal lobe epilepsy (ADNFLE)
  - Familial focal epilepsy with variable foci (FFEVF)
  - Childhood absence epilepsy (CAE)
  - Absence seizures with eyelid myoclonias (Jeavons syndrome)
  - Epilepsy with myoclonic absences
  - Lennox-Gastaut syndrome
  - Epilepsy with myoclonic atonic (previously astatic) seizures
  - Landau-Kleffner syndrome (LKS)
  - Epileptic encephalopathy with continuous spike-and-wave during sleep (CSWS)
  - Febrile Infection Related Epilepsy Syndrome (FIRES)
  - Rasmussen syndrome
  - Gelastic seizures with hypothalamic hamartoma
  - Hemiconvulsion-Hemiplegia-Epilepsy
  - Late-onset epilepsy spasms
  - Reflex epilepsies (e.g. photoconvulsive, musicogenic)

(Epilepsy Classification continued on next page)
(Epilepsy Classification, cont’d)

- Usually adolescent to adult onset
  - Juvenile Absence Epilepsy
  - Juvenile Myoclonic Epilepsy
  - Epilepsy with Generalized Tonic Clonic Seizures Alone (previously on Awakening)
  - Mesial temporal lobe epilepsy with hippocampal sclerosis (MTLE with HS)
  - Autosomal-dominant nocturnal frontal lobe epilepsy (ADNFLE)
  - Familial focal epilepsy with variable foci (childhood to adult) (FFEVF)
  - Autosomal dominant epilepsy with auditory features (ADEAF)
  - Familial temporal lobe epilepsy
  - Rasmussen syndrome – late onset
  - Gelastic seizures with hypothalamic hamartoma
  - Febrile Infection Related Epilepsy Syndrome (FIRES)
  - Late-onset epileptic spasms
  - Reflex epilepsies

If no syndrome present:

- Nonsyndromic epilepsy
  - With focal seizures
  - With generalized seizures
  - With mixed or unclassified seizures
Current seizure outcome:

- Drug-resistant (inadequate response to a reasonable trial of two or more primary anti-seizure treatments)
- Responder (>90% overall reduction in seizure frequency for >6 months on anti-seizure treatment)

Successful treatment: ______________________________

- Seizure-free without treatment
- Deceased: If so, epilepsy-related?  □ Yes  □ No  □ Unknown

Ever at least one year seizure free?  □ Yes  □ No  □ Unknown

Current number of:
- anti-seizure medications __________
- dietary therapies __________
- brain stimulation __________

Is patient or trio included in any other databases/studies? (e.g. EPGP, Epi4K, PERC, REN, CPEN, Seizure Tracker, etc.). If so, explain: ________________________________________________________________

___________________________________________________________________

Has the patient had an extreme response to any anti-seizure therapy (e.g. complete seizure cessation, life-threatening rash, etc.)? If so, explain: __________

___________________________________________________________________

___________________________________________________________________

___________________________________________________________________

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