Ambry Genetics®

Neurology Testing Family Studies Form

PLEASE SUBMIT THIS COMPLETED FORM AND ANY SUPPLEMENTAL DOCUMENTATION WITH THE SPECIMEN

FamilyStudies@ambrygen.com

FAMILY STUDY PARTICIPANT INFORMATION

Legal Name (Last, First, MI)	Date of Birth (MM/DD/YY)	Sex Assigned at Birth ☐F ☐M

Relationship to Proband ☐Mother ☐Father	Gender (optional) ☐Man ☐Woman ☐Nonbinary ☐Self-described	Genetic Ancestry: ☐Ashkenazi Jewish ☐Asian ☐Black/African American ☐French Canadian/Cajun ☐Hispanic/Latino ☐Mediterranean ☐Middle Eastern ☐Native American ☐Pacific Islander ☐Portuguese ☐White ☐Unknown ☐Other:

Send kit to family study participant ☐Yes ☐No	Address	City	State/Country	Zip

Specimen Type(s) ☐Blood (EDTA preferred) ☐Saliva ☐DNA ☐Other:	Collection Date (Required)	Indication Variant Study

PROBAND INFORMATION (Previously tested relative)

Legal Name (Last, First, MI)	Date of Birth (MM/DD/YY)	Ambry Accession number

FAMILY STUDIES TEST REQUEST	GENE	VARIANT
All VUS detected in proband (With the exception of VUS detected in autosomal recessive genes and gross deletion/duplications.)	See Proband Report	See Proband Report

ORDERING PROVIDER

Ordering Physician	Address	City	State /Country	Zip

Phone	Fax/Email	Original Ordering Provider: ☐Yes ☐No	Additional Authorized Recipient

CONTACT PERSON

Name (Last, First, MI)	Phone	Fax	Email

FAMILY STUDY PARTICIPANT CLINICAL HISTORY

PLEASE SUPPLY ANY AVAILABLE CLINIC NOTES (IF APPLICABLE)

☐Unaffected ☐Affected (If yes, please complete sections below) Diagnosis/Suspected diagnosis: _____

Neurodevelopment ☐N/A

☐Developmental Delay ☐Motor ☐Language ☐Global

Delay prior to seizure onset ☐Yes ☐No ☐N/A

☐Intellectual disability

☐Mild ☐Moderate ☐Severe ☐Profound

IQ score: _____ Head Circumference: _____

☐Regression or Plateau ☐Yes ☐No

☐Autism (Please describe behaviors):

Neurocutaneous Features ☐N/A

☐Café au lait ☐Telangiectasias ☐BCC ☐Lentigines ☐Angiofibromas

☐Fibromas ☐Shagreen patch ☐Hypomelanotic macules ☐Vitiligo

☐Other: _____

Other Features ☐N/A

☐MRI Results:

☐Microcephaly ☐Hypotonia ☐Spasticity ☐Movement disorder

☐Psychiatric disorder ☐Vision disorder ☐Dysmorphic features ☐Cardiac disorder

☐Renal Disorder ☐Endocrine disorder ☐Brain or spine tumor(s)

☐Peripheral nervous system tumor(s) ☐Vascular/ischemic abnormality ☐Head trauma

Epilepsy ☐N/A

☐Seizures: ☐Yes ☐No Age at first unprovoked seizure: _____

Seizures are ☐Refractory ☐Well-controlled

Check all that apply: ☐Infantile/epileptic spasms ☐Tonic ☐Atonic ☐Myoclonic

☐Typical absence ☐Generalized tonic clonic ☐Focal seizures ☐Status epilepticus

☐Convulsive ☐Non-convulsive ☐Neonatal seizures ☐Febrile seizures

☐Unclassified ☐Other: _____

☐EEG Results:

☐Normal ☐Classic hypsarrhythmia ☐Hypsarrhythmia variant

☐Generalized spike wave ☐Generalized paroxysmal fast activity (GPFSA)

☐Slow or disorganized for age ☐Focal or multi-focal sharp waves ☐Unknown

☐Other: _____

Comments:

IMPORTANT INFORMATION

- Please provide documentation on diagnosis, clinic symptoms and family history if available, as this will help yield the most accurate interpretation.
- Concurrent parental testing is the most efficient method of obtaining informative segregation data. However, variant testing can still proceed if only one parent is available.
- Family Studies is a free, research-based program and can take 2-3 months to report upon receipt of the last family member's sample and required documents. When a family studies report is issued, it is found under the proband's order and includes the results of all individuals that underwent testing. Family variant studies do not always lead to immediate reclassification.

Ordering Physician Signature: **Date:**

7 Argonaut, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

MKT-SPEC-TRF-30057-EN v2 04.15.25