Ambry Genetics

Neurology Parental Variant Study Requisition

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

COLLECTION DATE (REQUIRED)

If date of collection is not provided, three calendar days before specimen receipt will be used (for specimens stored longer than 30 days, the day of archive retrieval will be used as the date of service)

FAMILY STUDY PARTICIPANT INFORMATION

Legal Name (Last, First, MI)	DOB (MM/DD/YY)	Sex Assigned at Birth: ☐F ☐M	Gender (optional) ☐Man ☐Woman ☐Nonbinary ☐Self-described	Relationship to Proband ☐Mother ☐Father

Genetic Ancestry: ☐Ashkenazi Jewish ☐Asian ☐Black/African American ☐French Canadian/Cajun ☐Hispanic/Latino ☐Mediterranean ☐Middle Eastern ☐Native American ☐Pacific Islander ☐Portuguese ☐White ☐Unknown ☐Other:

Indication: Variant Study

SPECIMEN INFORMATION (Please see ambrygen.com/specimen-requirements for details)

☐Personal history of allogenic bone marrow or peripheral stem cell transplant

Specimen ID	Medical Record #

PROBAND INFORMATION (Previously tested relative)

Legal Name (Last, First, MI)	DOB (MM/DD/YY)	Ambry Accession number

FAMILY STUDIES TEST REQUEST	GENE	ALTERATION
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

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.
- The current turnaround time for results is 2-3 months. Please contact the Family Studies Program if results are needed sooner and we will try our best to accommodate.

Ordering Physician Signature: **Date:**

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