Ambry Genetics®
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Neurology Family Studies Form

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

FAMILY STUDY PARTICIPANT INFORMATION

Name (Last, First, MI)	DOB (MM/DD/YY)	Relationship to Proband	Biological Sex
		☐ Mother ☐ Father	☐ F ☐ M

Ethnicity: ☐ African American ☐ Asian ☐ Caucasian ☐ Hispanic ☐ Jewish (Ashkenazi) ☐ Portuguese ☐ Other:

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

PROBAND INFORMATION (Previously tested relative)

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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Family Study Participant Consent Form

Test Purpose

The purpose of the testing being performed is to assist the analysis of you and/or your family member's (proband's) result. The result involves a variant of unknown significance (VUS), which is an alteration(s) with limited and/or conflicting evidence regarding association with disease. Medical management is based on personal and family clinical histories, not VUS carrier status. Unless the variant is reclassified to a clinically actionable alteration, a report will only be generated for the proband, and it may be possible to infer information about a family member's result(s) based on the proband's report. If a family member is unclear about their results from the study, their healthcare provider or genetic counselor can contact the Ambry Family Studies Program for further discussion.

Test Method

The blood, body fluid, or tissue specimen submitted is required for isolation and purification of DNA and/or RNA for molecular genetic testing. The test will cover the specific test(s) requested on the Ambry Genetics Family Study requisition form.

Ambry's Rights

Ambry reserves the right to 1) refuse testing if one of the conditions in the Patient Consent form is not met, or 2) cancel testing if the proband's result no longer requires further clarification.

Research & Recontact Consent

Ambry Genetics is committed to improving genetic testing for all patients and contributing to scientific research. For more information on research at Ambry Genetics, please visit ambrygen.com/patient-resources. NOTE: If left blank, consent is interpreted as "NO".

☐ I agree to use of my de-identified biospecimen for research to improve genetic testing for all patients and contribute to scientific research.

☐ I am a New York state resident and I give Ambry Genetics permission to store my sample for up to 1 year after testing completion.

☐ In addition to agreeing above, I agree to be contacted by Ambry Genetics regarding research opportunities.

_____ _____

Family Study Participant Signature Date

_____

Family Study Participant Name (please print)

Previously Tested Relative (Proband) as indicated on the Family Studies Requisition

_____

Proband Name (Write "Self" if you are the proband)

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

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Please Keep This Copy For Your Records

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