

Application for Complimentary Family Studies

XLMR

Family studies on variant(s) of unknown significance (VUS) may help elucidate if the genetic change detected in your patient is an inherited or de novo event and/or whether it segregates with disease. Complimentary family studies are considered on a case by case basis and is authorized at the discretion of the medical director. Review of the proband's family history and clinical information is required.

In order to be considered for complimentary family study, please complete the following information and fax to Ambry Genetics with the required documents at 949-900-5501 (**ATTN: XLMR**) or email an attachment to **XLMR@ambrygen.com**. Due to the high volume of requests please allow **7-10 days** for a response from one of our genetic counselors.

Proband's Information:

Proband's Name:	Accession #:
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Physician/GC Information:

Name:
Phone Number:
Fax Number:
Email Address:

1. Which VUS(s) are you interested in testing?
2. Which of the proband's relatives are available for testing? *In general, the most informative relatives are the proband's mother and maternal male relatives (affected or unaffected).*

Name	Gender	Relationship to Proband	Healthy or Affected

3. Please submit the following documents:
 - a. Completed proband information form (see attached)
 - b. Detailed pedigree
 - c. Supplemental clinical information, such as a consultation letter from a geneticist

Exclusions for complimentary family studies:

1. The VUS is found at a high frequency in the normal population (based on data from the SNP database) or at a very high frequency in affected cohorts (Ambry cohort or published cohorts).
2. Mutation of the gene involved is not known to have phenotypic consequences consistent with the patient's clinical symptoms based on published literature.
3. Previous family studies have been performed on variant and/or it has been reclassified.

Please be aware that results for family studies have a longer turnaround time (**4-6 months**) since priority is given to paid clinical specimens. If you have any questions please contact Ambry Genetics and ask to speak with one of our XLMR specialists at 949-900-5500 or email **XLMR@ambrygen.com**.

Probands Information Form

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Clinical information can help us provide genotype phenotype correlation in your patient's report and optimizes our molecular analysis.

Proband's Information:

Patient name:
Date of Birth:
Accession Number:
Gender: <input type="checkbox"/> Male <input type="checkbox"/> Female
Ethnicity:

1. Family history (or attached pedigree): _____
2. Intellectual delay/mental retardation:
 - A) ID/MR
 Mild Moderate Severe Profound
 - B) Overall IQ: _____
 - C) Verbal Aptitude
 Normal mild deficiency monderate deficiency non-verbal
 - D) Autism:
 No autistic behaviors
 Autistic behavior (please describe): _____

3. Dysmorphic features (please describe): _____

4. Congenital anomalies (please describe): _____
5. Hx of Seizures: No Yes diagnosed epilepsy
6. Previous studies:
 - A) MRI/CT studies (findings): _____
 - B) Chromosome analysis: _____
 - C) Microarray analysis: _____
 - D) Other molecular studies: _____
8. Growth indices:
 - A) Head circumference: _____ %
 - B) Weight: _____ %
 - C) Height: _____ %