Ambry Genetics®

Cardiovascular Application for Family Studies

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 the disorder. Family studies are considered on a case-by-case basis and are authorized at the discretion of the medical director. Review of the proband's family history and clinical information is required.

Conditions exhibiting autosomal dominant and X- linked inheritance may be considered for this program.* In order to be considered for a family study, please complete the following information and fax to Ambry Genetics at +1 949-271-5621 (ATTN: Family Studies) or email an attachment to familystudies@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 name: _____ Accession number: _____

Physician/GC: _____

Phone number: _____ Fax number: _____ Email: _____

1. Which VUS(s) are you interested in testing?

2. Which of the proband's relatives are available for testing (if known)? *Please only list informative relatives.*

NAME (leave blank if unknown)	RESIDES OUTSIDE US*	GENDER	RELATIONSHIP TO PROBAND	HEALTHY	AFFECTED	DISEASE/SYMPTOMS (if affected)	DX AGE (if affected)
	☐			☐	☐		
	☐			☐	☐		
	☐			☐	☐		
	☐			☐	☐		

** Please indicate if any of the proband's relatives live outside the United States, as special considerations may apply.*

Please note clinical documentation may be requested on informative relatives to aid variant interpretation and reclassification efforts.

Exclusions for family studies: 1. The VUS is found at a high frequency in the general population (as reported by databases such as the SNP database, 1000 Genomes Project, Exome Sequencing Project, etc.) or at a very high frequency in patient 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.

Please be aware that results for cardiovascular family studies have a 2-3 month turnaround. If you have any questions or concerns, please contact Ambry Genetics and ask to speak with one of our family studies specialists at +1 949-457-4773 or email familystudies@ambrygen.com.

*In certain cases, autosomal recessive disorders may be considered for family studies if an appropriate number of familial specimens are available to obtain adequate segregation data. Please consult one of our genetic counselors for special consideration.

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Proband Information Form

Clinical information can help us provide genotype-phenotype correlation in your patient's report and optimizes our molecular analysis. **Please attach pedigree and appropriate medical records that may aid variant assessment, or fill out the information below.**

PROBAND INFORMATION

Name (Last, First, MI)

Accession number	DOB (MM/DD/YY)	Gender ☐F ☐M	Ethnicity: ☐African American ☐Asian ☐Caucasian ☐Hispanic ☐Jewish (Ashkenazi) ☐Portuguese ☐Other:

CLINICAL HISTORY

PLEASE SUPPLY CLINIC NOTES AND PEDIGREE

☐ No personal history of cardiovascular disease

Sudden cardiac arrest ☐ Y ☐ N If yes, # episodes:_____ Age first incident: _____

Episodes:_____ Age first incident:_____

Syncope: ☐ Y ☐ N If yes, # episodes:_____ Age first incident: _____

History of cardiomyopathy ☐ Y ☐ N Age at dx:_____

☐ HCM ☐ DCM ☐ ARVD ☐ LVNC ☐ RCM

☐ Other cardiomyopathy types:_____

History of arrhythmia ☐ Y ☐ N Age at dx: _____

☐ Long QT ☐ Short QT ☐ Brugada ☐ CPVT ☐ ARVD

☐ Other arrhythmia types: _____

Other features/syndromes

☐ Clinical diagnosis of Marfan syndrome

☐ Aortic aneurysm/dilation, age at dx:_____

☐ Other aneurysm, location:_____ Age at dx:_____

☐ Aortic/vascular dissection, location:_____Age at dx:_____

☐ Arterial tortuosity

☐ BAV ☐ MVP

☐ Congenital heart defect type:_____

☐ Ectopia lentis ☐ Myopia

☐ Marfanoid habitus

☐ Pectus deformity type:_____

☐ Scoliosis ☐ Joint hypermobility ☐ Joint contractures

☐ Pneumothorax

☐ Craniosynostosis

☐ Facial clefting, type : _____

☐ Xanthoma(s)

☐ Epistaxis (nosebleeds) ☐ Telangiectasia

☐ AVM location: _____

☐ Amyloidosis, age at dx: _____

☐ Neuromuscular disease, specify: _____

☐ Hearing loss, describe: _____

☐ Genetic syndrome, specify: _____

☐ Other, specify:_____

CLINICAL TESTING AND PROCEDURES

LDL-C: _____ Total cholesterol:_____ Age at testing:_____

Procedures (*e.g.:* EKG, ECHO, etc.) Age: _____ Result (*e.g.:* LVIDd, PWd, Qtc, etc): _____ Type: _____

Age: _____ Result (*e.g.:* LVIDd, PWd, Qtc, etc): _____ Type: _____

Cardiovascular device implant (eg: pacemaker, ICD, LVAD, etc.): Age at implantation: _____Type: _____

Additional history: