GeneDx Variant Testing Programs – General Overview

GeneDx classifies sequencing variants in accordance with the 2015 ACMG standards and guidelines for the interpretation of sequence variants [Richards et al. (2015) Genetics in Medicine: Official Journal of the American College Of Medical Genetics: (PMID: 25741868).]. A classification of variant of uncertain significance (VUS) is assigned if there is insufficient information about the variant to lead to a benign, likely benign, pathogenic, or likely pathogenic interpretation using the standards set forth in the guidelines. Because the clinical significance of a VUS is unknown, these results generally cannot be used for risk assessment or medical management purposes.

In some situations, testing family members for the presence or absence of a VUS may provide information that could contribute to the eventual reclassification of a VUS to a clinically meaningful result. The GeneDx Variant Testing Programs offer select informative families the opportunity for relatives to undergo testing for the familial VUS**. Relatives accepted into a GeneDx Variant Testing Program are offered targeted testing for the familial VUS at no additional charge, provided they submit relevant clinical and/or family history information**. GeneDx will make the final determinations for VTP in its sole discretion. Note that classifying a genetic variant as anything other than a VUS requires data from multiple lines of evidence and there is no guarantee that participation in a Variant Testing Program will lead to reclassification of a VUS.

GeneDx has several Variant Testing Programs, each established for different disease states and clinical situations. Note that each Variant Testing Program runs independently and has specific requirements based on the unique nature of the clinical specialty. Please select a link below for more information about individual GeneDx Variant Testing Programs.

Program*	Send Pedigree and Clinical Information to**:	Notes
Cardiogenetics	Fax: 301-519-2892 Email: genedx@genedx.com	May require documentation of EKG, ECG or other studies for at-risk family members
Clinical Genomics (Exome and Genome Sequencing)	Fax: 301-519-2892 Email: genedx@genedx.com	
GenomeDx (Microarray)	Fax: 301-519-2892 Email: genedx@genedx.com	Parents may automatically be accepted into this VTP; if so, this will be stated on the proband's test report
Hereditary Cancer	Fax: 201-421-2314 Email: genedx@genedx.com	
Metabolic/Mitochondrial Disorders	Fax: 301-519-2892 Email: genedx@genedx.com	

Neurology	Fax: Email:	301-519-2892 genedx@genedx.com	Parents may automatically be accepted into this VTP; if so, this will be stated on the proband's test report
Ophthalmology	Fax: Email:	301-519-2892 genedx@genedx.com	May require documentation of specific evaluations or studies (such as ERG) in at risk family members
Rare Disorders	Fax: Email:	301-519-2892 genedx@genedx.com	
*Please refer to the program specific VTP links for more details about each VTP program and the application process ** Include the proband's name, GeneDx accession # (specimen ID#), and the name of the VTP program you are applying to on your fax/email			

For more information about variant classification see:

Data Analysis and Variant Classification Process

New ACMG Guidelines

GeneDx Webinar Series