

Cytogenetics and Molecular Genetics Parental and Familial Variant Testing Policy

Policy on Parental Testing: WSLH Cytogenetics and Molecular Genetics will recommend parental testing in the event analysis of the biological parents will aid in the interpretation of a variant of uncertain significance or when the assessment of phase is critical to a determination of pathogenicity. Studies requested for these purposes will be done as an additional test, free of charge, and may use an alternative method at the director's discretion using the residual proband specimen as an assay positive control if needed. Requests for parental specimens will be made as a recommendation in the proband's final report. Parental results will be reported as an amendment to the proband's original record and additionally may be reported independently when confirming microarray findings.

Test Name	SLH Test Code	CPT Code(s)	Cost
Microarray analysis: Parental study	890PAR	N/A	No Additional Cost
FISH analysis: Parental confirmation of array findings	871	N/A	No Additional Cost
Sanger sequencing: Parental confirmation of exome findings	895M52	N/A	No Additional Cost
Sanger sequencing: First degree relative (FDR) confirmation of exome findings	895M52	N/A	No Additional Cost

Policy on Carrier Testing of a Familial Variant: WSLH Cytogenetics and Molecular Genetics will offer carrier testing of a familial (private) variant as a fee for service test when a pathogenic or likely pathogenic variant has been detected in a proband using a clinically validated test. The fee for the assay will depend on the method selected for the confirmatory test (see below). Documentation of the specific familial variant(s) is required; if the original testing in the family was performed by another laboratory (not WSLH), we require a copy of the original test report. A sample from the proband (or another known positive family member) is required for FISH analysis studies to use as an assay positive control. Please call the laboratory at 608-262-0402 to discuss familial variant testing prior to submitting a sample.

Test Name	SLH Test Code	CPT Code(s)	Cost
Microarray Analysis: Family study	890FAM	81229	\$420
FISH Analysis: Familial study	871	88271, 88273	\$446
Targeted Variant Analysis via Sanger DNA Sequencing	895M53	81403	\$150

Additional information including accepted specimen types and turn-around-times can be found on our [online reference manual](#).

