



TARGETED FAMILIAL VARIANTS (Test #100, 200, 300, 1400)

We offer single (Test Code 100), double (Test Code 200), or triple (Test Code 300) exon sequencing for any of the genes on our test menu. Targeted testing (PCR, MLPA, and/or FISH) of relatives of patients who had copy number testing (del/dup studies via aCGH or chromosomal microarray studies (CMA)) at PreventionGenetics are available via Test Code 1400.

INDICATIONS

Candidates for this test are:

- Family members who may have familial pathogenic variants.
- Patients wishing to confirm research results.
- Setting phase of variants.
- Clarifying interpretation of variants.

PRICING

Test Code 100	Targeted Family Mutations-1 variant	\$ 250
Test Code 200	Targeted Family Mutations-2 variants	\$ 370
Test Code 300	Targeted Family Mutations-3 variants	\$ 440
Test Code 1400	Targeted Family Deletion/Duplication	\$ 340

Free testing for variants of unknown significance may be offered in up to two family members of probands who were tested at PreventionGenetics. Free testing for deletion/duplications of unknown significance discovered by aCGH or CMA will be available for up to two family members of the proband only if we are able to test the del/dup by PCR, MLPA, or FISH.

ORDERING

Please complete fully our [Targeted Variant Test Requisition Form](#).

TURNAROUND TIME

Maximum of 30 days, although many tests are completed in 2-3 weeks. STAT testing is available with a 10 calendar TAT but incurs a 25% surcharge.

POSITIVE CONTROLS

For sequencing tests, if the proband was tested at an outside laboratory, we recommend that you submit a sample from that individual to serve as a positive control. The positive control sample will be tested free of charge, and no report will be provided.

Please label the sample and Test Requisition Form: “POSITIVE CONTROL ONLY-No Report”. We will not issue a report for samples submitted as positive controls. Please include the outside laboratory report.

If a positive control is not provided, negative results will carry a limitation stating that PreventionGenetics did not have the opportunity to verify that we can detect the variant in this family.

DELETIONS/DUPLICATIONS via aCGH

Targeted testing (Test Code 1400) for gross deletions/duplications discovered by aCGH will be available for family members of the proband only if we are able to test the del/dup by PCR or MLPA. If this is not possible, then the full gene deletion/duplication test (Test Code 600) may be necessary. Please [contact us](#) if you have questions about ordering family studies for deletions/duplications.

PRENATAL TESTING

Please see our [Prenatal Policy](#) for details of prenatal testing. If Test Code 990 is ordered (Targeted Prenatal Variants), parents are offered testing at no charge.

SPECIMEN REQUIREMENTS

2-5 ml EDTA of whole blood is preferred, but many other DNA sources are acceptable. See the specimen requirements page of the [Targeted Variant Test Requisition Form](#).

CPT CODES

See the detailed test description for the gene of interest for specific applicable [CPT codes](#). Detailed test descriptions may be found using our [test search page](#).

CONTACTS

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