Overview

Useful For

Diagnostic or predictive testing for specific conditions when 1 or more variants have been identified in a family member

Carrier screening for individuals at risk for having a variant that was previously identified in a family member

Genetics Test Information

Documentation of the specific familial variants is **required** and must be provided with the specimen in order to perform this test. Consultation with the laboratory is required prior to ordering this test.

Note: analysis of the area surrounding the familial variant may be required in the performance of this assay, which could result in identification of additional variants. Contact the laboratory at 800-533-1710 with any questions regarding assay performance.

The preferred specimen for this test is whole blood. Other specimens may be acceptable for certain genes as follows.

The following genes are available for testing on the blood spot specimen type:

ABCD1, ACADM, ACADS, ACADVL, ARSA, ARSB, CFTR, CPT2, CPOX, FECH, FTCD, GAA, GALT, GALC, GBA, GLA, GNS, HMBS, IDS, IDUA, MMACHC, MMADHC, NAGLU, PPOX, SGSH, SLC25A20, SMN1, SMPD1,* and *SUMF1*

***Note: CFTR deletion/duplication analysis is not offered on dried blood spot specimens.**

The following genes are available for testing on fibroblasts and skin biopsy specimen types:

ABCD1, ACADM, ACADS, ACADVL, APOA1, APOA2, ARSA, ARSB, ATP7B, BTD, CDKN1C, CPOX, CPT2, CTRC, FECH, FGA, FTCD, GAA, GALC, GALT, GBA, GLA, GNPTAB, GNS, GRN, GSN, HEXA, HMBS, IDS, IDUA, LYZ, MAPT, MMACHC, MMADHC, NAGLU, PKHD1, PPOX, PRSS1, RET, SCG5, SGSH, SLC25A20, SMN1, SMPD1, SPINK1, SUMF1, TTR, and *UBE3A*

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

The following genes are available for testing on prenatal specimen types:

ABCD1, AGXT, ARSA, ARSB, BTD, CDKN1C, CFTR, CPT2, GLA, GALC, GALT, GBA, GNPTAB, GNS, GRHPR, HEXA, IDS, IDUA, MLYCD, MMACHC, MMADHC, NAGLU, NPC1, NPC2, PKHD1, SGSH, SLC25A20, SMN1, SMPD1, SUMF1, and *UBE3A.* Contact the laboratory to inquire about genes not included on this list.

Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
MATCC	Maternal Cell Contamination, B	Yes	No
CULFB	Fibroblast Culture for Genetic Test	Yes	No

Test ID	Reporting Name	Available Separately	Always Performed
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
_G001	Gene GRHPR	No, (Bill Only)	No
_G002	Gene PPOX	No, (Bill Only)	No
_G003	Gene CFTR_SEQ	No, (Bill Only)	No
_G004	Gene CFTR_MLPA	No, (Bill Only)	No
_G005	Gene MLH1	No, (Bill Only)	No
_G006	Gene MSH2	No, (Bill Only)	No
_G007	Gene MSH6	No, (Bill Only)	No
_G008	Gene MECP2_SEQ	No, (Bill Only)	No
_G009	Gene MLH3	No, (Bill Only)	No
_G010	Gene CHEK2	No, (Bill Only)	No
_G011	Gene IDUA	No, (Bill Only)	No
_G012	Gene AXIN2	No, (Bill Only)	No
_G013	Gene BMPR1A	No, (Bill Only)	No
_G014	Gene PTEN	No, (Bill Only)	No
_G015	Gene SMAD4	No, (Bill Only)	No
_G016	Gene STK11	No, (Bill Only)	No
_G017	Gene TP53	No, (Bill Only)	No
_G018	Gene IDS	No, (Bill Only)	No
_G019	Gene FLCN	No, (Bill Only)	No
_G020	Gene SPINK1	No, (Bill Only)	No
_G021	Gene PRSS1	No, (Bill Only)	No
_G022	Gene CTRC	No, (Bill Only)	No
_G025	Gene ABCD1	No, (Bill Only)	No
_G026	Gene CDH1	No, (Bill Only)	No
_G027	Gene NAGLU	No, (Bill Only)	No
_G028	Gene SGSH	No, (Bill Only)	No
_G029	Gene ARSB	No, (Bill Only)	No
_G030	Gene GNPTAB	No, (Bill Only)	No
_G031	Gene SEPT9	No, (Bill Only)	No
_G032	Gene ACADVL	No, (Bill Only)	No
_G033	Gene ACADM	No, (Bill Only)	No
_G034	Gene ACADS	No, (Bill Only)	No
_G035	Gene FECH	No, (Bill Only)	No
_G036	Gene MAPT	No, (Bill Only)	No
_G037	Gene PKHD1	No, (Bill Only)	No
_G038	Gene GRN	No, (Bill Only)	No

Test ID	Reporting Name	Available Separately	Always Performed
_G039	Gene FTCD	No, (Bill Only)	No
_G040	Gene CDKN1C	No, (Bill Only)	No
_G041	Gene CPOX	No, (Bill Only)	No
_G042	Gene ATP7B	No, (Bill Only)	No
_G043	Gene GAA	No, (Bill Only)	No
_G044	Gene HMBS	No, (Bill Only)	No
_G045	Gene GALT	No, (Bill Only)	No
_G046	Gene GLA	No, (Bill Only)	No
_G047	Gene BTD	No, (Bill Only)	No
_G048	Gene HEXA	No, (Bill Only)	No
_G049	Gene AGXT	No, (Bill Only)	No
_G050	Gene APC	No, (Bill Only)	No
_G051	Gene MLYCD	No, (Bill Only)	No
_G052	Gene MMACHC	No, (Bill Only)	No
_G053	Gene GBA	No, (Bill Only)	No
_G054	Gene SMPD1	No, (Bill Only)	No
_G055	Gene CPT2	No, (Bill Only)	No
_G056	Gene TTR	No, (Bill Only)	No
_G057	Gene UBE3A	No, (Bill Only)	No
_G058	Gene GALC	No, (Bill Only)	No
_G059	Gene GSN	No, (Bill Only)	No
_G060	Gene LYZ	No, (Bill Only)	No
_G061	Gene FGA	No, (Bill Only)	No
_G062	Gene APOA1	No, (Bill Only)	No
_G063	Gene APOA2	No, (Bill Only)	No
_G064	Gene MMADHC	No, (Bill Only)	No
_G065	Gene SLC25A20	No, (Bill Only)	No
_G066	Gene ARSA	No, (Bill Only)	No
_G067	Gene NPC1/2_SEQ and NPC1/2_MLPA	No, (Bill Only)	No
_G068	Gene PMS2_LR and PMS2_SEQ	No, (Bill Only)	No
_G069	Gene PMS2_MLPA	No, (Bill Only)	No
_G070	Gene RAI1	No, (Bill Only)	No
_G071	Gene MUTYH	No, (Bill Only)	No
_G072	Gene HGSNAT	No, (Bill Only)	No
_G073	Gene GNS and GRHPR_MLPA	No, (Bill Only)	No
_G074	Gene PSAP	No, (Bill Only)	No

Test ID	Reporting Name	Available Separately	Always Performed
_G075	Single-gene Large Del/Dup	No, (Bill Only)	No
_G076	Gene MECP2_MLPA	No, (Bill Only)	No
_G077	Gene RET	No, (Bill Only)	No
_G078	Gene SUMF1	No, (Bill Only)	No
_G079	Gene CASR_Seq	No, (Bill Only)	No
_G080	Gene VHL_SEQ	No, (Bill Only)	No
_G081	VHL_MLPA	No, (Bill Only)	No
_G082	Gene SHDP_MLPA	No, (Bill Only)	No
_G083	Gene SDHB, SDHC, and SDHD_MLPA	No, (Bill Only)	No
_G084	Gene SDHB, SDHC, SDHD_Seq	No, (Bill Only)	No
_G085	Gene BRCA1	No, (Bill Only)	No
_G086	Gene BRCA2	No, (Bill Only)	No
_G087	Gene DMD_MLPA	No, (Bill Only)	No
_G088	Gene PMP22_MLPA	No, (Bill Only)	No
_G089	Gene MPZ_MLPA	No, (Bill Only)	No
_G102	Gene SERPINA1	No, (Bill Only)	No
_G112	Gene SDHAF2	No, (Bill Only)	No
_G113	Gene TMEM127	No, (Bill Only)	No
_G114	Gene MAX	No, (Bill Only)	No
_G115	Gene SMN1	No, (Bill Only)	No
_G125	Gene PMP22_SEQ	No, (Bill Only)	No
_G127	Gene GJB2_SEQ	No, (Bill Only)	No
_G128	Gene HBA1/HBA2_SEQ	No, (Bill Only)	No
_G129	Gene HBB_SEQ	No, (Bill Only)	No
_G130	Known Familial Variant,Other	No, (Bill Only)	No
G168	Gene CSTB	No, (Bill Only)	No
G169	Gene CACNA1A	No, (Bill Only)	No

Testing Algorithm

For prenatal specimens only: If amniotic fluid (nonconfluent cultured cells) is received, amniotic fluid culture will be added and charged separately. If chorionic villus specimen (nonconfluent cultured cells) is received, fibroblast culture will be added and charged separately. For any prenatal specimen that is received, maternal cell contamination studies will be added.

Testing for variants detected by whole exome sequencing (WES) or large panels: Any familial mutation targeted testing (FMTT) orders for a variant that was detected by WES or next-generation sequencing (NGS) large panel assays requires a proband sample that has been previously tested at Mayo Clinic Laboratories. Contact the

laboratory to determine whether adequate DNA is available in the laboratory or if a new proband sample is required.

The following algorithms are available in Special Instructions:

-Fabry Disease Diagnostic Testing Algorithm

-Full Gene Analysis/Multi-Gene Panels versus Familial Mutation Targeted Testing

-Lynch Syndrome Testing Algorithm

-Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

Special Instructions

- Informed Consent for Genetic Testing
- Fabry Disease Diagnostic Testing Algorithm
- Familial Mutation Testing: Required Patient Information
- Full Gene Analysis/Multi-Gene Panels versus Familial Mutation Targeted Testing
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Lynch Syndrome Testing Algorithm
- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm
- Informed Consent for Genetic Testing (Spanish)

Method Name

Polymerase Chain Reaction (PCR) followed by DNA Sequencing Analysis, Gene Dosage Analysis by Array Comparative Genomic Hybridization (aCGH), and/or Gene Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA)

NY State Available

Yes

Specimen

Specimen Type

Varies

Advisory Information

This testcan only be performed if a variant has previously been identified in a family member of this individual.

Additional Testing Requirements

All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Shipping Instructions

Specimen preferred to arrive within 96 hours of collection.

Prenatal specimens can be sent Monday through Thursday and **must be received by 5 p.m. CST on Friday** in order to be processed appropriately.

Necessary Information

Testing may be delayed if the required documentation is not received (ie, Familial Mutation Testing: Required Patient

Information [T721]) in Special Instructions.

Specimen Required

Refer to Genetics Information for a complete list of genes tested by specimen type.

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts

Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 Full T-75 or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper, or Card-Blood Spot Collection Card

Specimen Volume: 2 to 5 Blood spots on collection card

Collection Instructions:

1. An alternative blood collection option for a patient >1 year of age is finger stick.

2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.

3. Do not expose specimen to heat or direct sunlight.

4. Do not stack wet specimens.

5. Keep specimen dry

Additional Information:

1. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777) in Special Instructions.

2. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800) in Special Instructions.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Tissue

Container/Tube: Tissue block

Collection Instructions: Submit a formalin-fixed, paraffin-embedded tissue block.

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Prenatal Specimens

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

Acceptable

Specimen Type: Confluent cultured cells

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Familial Mutation Testing: Required Patient Information (T721) in Special Instructions

3. If not ordering electronically, complete, print, and send a Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Specimen Minimum Volume

Amniotic Fluid: 10 mL
Blood: 1 mL
Chorionic Villi: 5 mg
Blood Spots: 2

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical and Interpretive

Clinical Information

This test is available to test for the presence of 1 or more variants previously identified in a family member. Targeted testing is used for diagnostic or predictive testing in cases in which variants have been previously identified in an affected family member. Targeted testing is available for the genes listed in the table below or variants in genes detected previously by large panels or whole exome sequencing (WES) at Mayo Clinic Laboratories.

Genes Available for Testing*			
ABCD1	*ACADM*	*ACADS*	*ACADVL*
AGXT	*APC*	*APOA1*	*APOA2*
ARSA	*ARSB*	*ATP7B*	*AXIN2*
BMPR1A	*BRCA1*	*BRCA2*	*BTD*
CASR	*CDH1*	*CDKN1C*	*CFTR*
CHEK2	*CPOX*	*CPT2*	*CTRC*
DMD	*FECH*	*FGA*	*FLCN*
FTCD	*G6PD*	*GAA*	*GALC*
GALT	*GBA*	*GJB2*	*GLA*
GNPTAB	*GNS*	*GRHPR*	*GRN*
GSN	*HEXA*	*HBA1/HBA2*	*HBB*
HGSNAT	*HMBS*	*IDS*	*IDUA*
LYZ	*MAPT*	*MAX*	*MECP2*
MLH1	*MLH3*	*MLYCD*	*MMACHC*
MMADHC	*MSH2*	*MSH6*	*MUTYH*
NAGLU	*NPC1*	*NPC2*	*PKHD1*
PMS2	*PMP22*	*PPOX*	*PRSS1*
PSAP	*PTEN*	*RAI1*	*RET*
SERPINA1	*SCG5*	*SDHAF2*	*SDHB*
SDHC	*SDHD*	*SEPT9*	*SGSH*
SLC25A20	*SMAD4*	*SMN1*	*SMPD1*
SPINK1	*STK11*	*SUMF1*	*TACSTD1/EPCAM*
THEM127	*TP53*	*TTR*	*UBE3A*
VHL			

*FMTT is available for family members of a patient who had testing performed by the Genomics Laboratory at Mayo Clinic Laboratories. For these individuals, this test can be used to detect variants in the genes listed in the table above, in addition to any gene detected via large panels or WES. Contact the laboratory to determine whether adequate DNA is available in the laboratory or if a new proband sample is required.

Refer to the following resources for information regarding the listed gene targets. GeneReviews-NCBI Bookshelf, available at www.ncbi.nlm.nih.gov/books/NBK1116/ or OMIM, available at www.omim.org/.

Reference Values

An interpretive report will be provided.

Interpretation

All detected alterations are evaluated according to American College of Medical Genetics and Genomics (ACMG) recommendations.(1) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions

Clinical Correlations:

The identification of a disease-causing variant in an affected family member is necessary before predictive testing for other family members can be performed. If a familial variant has not been previously identified, call 800-533-1710 to discuss testing options.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Technical Limitations:

Rare allelic variants (polymorphisms) exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Analysis is performed for the familial variants provided only. This assay does not rule out the presence of other variants within this gene or within other genes that may be associated with hereditary cancer syndromes.

Note: Analysis of the area surrounding the familial variant may be required in the performance of this assay, which could result in identification of additional variants.Contact the laboratory with any questions regarding assay performance.

In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing**.**

Clinical Reference

1. Richards S, Aziz N, Bale S, et al: Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med 2015 May;17(5):405-424

Performance

Method Description

DNA sequencing and/or dosage analysis by array comparative genomic hybridization or multiplex ligation-dependent probe amplification is utilized to test for the presence of the specific mutations previously identified in a family member.(Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Monday through Friday; Varies

Analytic Time

10 days

Maximum Laboratory Time

16 days

Specimen Retention Time

Whole Blood: 2 weeks (if available), Extracted DNA: 3 months

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
FMTT	Familial Mutation, Targeted Testing	51966-0

Result ID	Test Result Name	Result LOINC Value
36528	Result Summary	50397-9
36529	Result	82939-0
36530	Interpretation	69047-9
36531	Additional Information	48767-8
36532	Specimen	31208-2
36533	Source	31208-2
36534	Method	49549-9
36535	Released By	18771-6