

Overview

Useful For

Diagnostic or predictive testing for specific conditions when a DNA variant of interest has been previously identified in a family member and follow-up testing for this specific variant in other family members is desired

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

Segregation analysis for a familial DNA variant

Confirmation of germline status for variants detected via somatic testing

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 |
| 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 | 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 | 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 |
| _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 | No, (Bill Only) | No |

| | | | |
|-------|--|-----------------|----|
| | NPC1/2_MLPA | | |
| _G068 | Gene PMS2 | 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 |
| _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 |
| _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 |
| _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 | No, (Bill Only) | No |
| _G126 | Gene G6PD | No, (Bill Only) | No |
| _G127 | Gene GJB2 | No, (Bill Only) | No |
| _G128 | Gene HBA1/HBA2 | No, (Bill Only) | No |
| _G129 | Gene HBB | 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 |
| _STR1 | Comp Analysis using STR (Bill only) | No, (Bill only) | No |
| _STR2 | Add'l comp analysis w/STR (Bill Only) | No, (Bill only) | No |
| G230 | Gene AR | No, (Bill Only) | No |
| G231 | Gene FXN | No, (Bill Only) | No |
| G232 | Gene PALB2 | No, (Bill Only) | No |

Genetics Test Information

Familial variant targeted testing is available for most genes that are currently part of another genetic test offered by Mayo Clinic Laboratories. Additional genes may also be available and require consultation with the laboratory prior to

ordering.

Genes or gene regions generally not offered under this test ID currently include, but are not limited to, the following:

- Genes with limitations related to patents
- Genes with limitations related to homology
- Mitochondrial DNA genes for heteroplasmy
- Globin genes for prenatal testing
- CFHR1, CFHR2, CFHR3, CFHR4, CFHR5*
- CYP21A2*
- NOTCH2* exons 1-4
- PKD1* single- or multi-exon deletions/duplications involving exons 1-33
- PKLR*
- TNXB* single- or multi-exon deletions/duplications involving exons 32-44
- TTC25 (ODAD4)* exons 9-12
- TTN (NM_001256850.1)* exons 154-156
- UGT1A1*

Additional genes/variants may be unavailable per laboratory discretion.

Targeted testing is available regardless as to whether the family/individual had previous testing through Mayo Clinic Laboratories or another laboratory. See Additional Testing Requirements if the familial variant was previously identified at an outside laboratory. 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 on prenatal specimens.

The preferred specimen for this test is whole blood. Other specimens may be acceptable depending on the gene and methodology required. Contact the laboratory if you have questions regarding a specific specimen type. In general, deletion/duplication analysis requires a higher volume and concentration of DNA, therefore, whole blood is the preferred specimen type.

In some cases, testing for a known variant may require submission of additional proband sample or may not be available for technical or legal reasons. Testing may be declined at the discretion of the laboratory.

Testing Algorithm

Pricing for this test is based on the genes selected. The assigned CPT codes correspond to the reflexed bill only G code.

Samples received by the laboratory undergo review for appropriateness of testing and clinical utility. For prenatal samples, variant curation will be performed prior to initiating testing. For all other samples, if the proband was not tested at Mayo Clinic Laboratories, a full variant curation will not typically be completed before testing is performed. Variants may occasionally be classified differently by independent laboratories.

For prenatal specimens only:

- If amniotic fluid (nonconfluent cultured cells) is received, amniotic fluid culture/genetic test will be added at an additional charge.
- If chorionic villus specimen (nonconfluent cultured cells) is received, fibroblast culture for genetic test will be added at

an additional charge.

For any prenatal specimen that is received, maternal cell contamination studies will be added.

If skin biopsy is received, fibroblast culture and cryopreservation will be performed at an additional charge. If viable cells are not obtained, the client will be notified.

For more information see [Full Gene Analysis/Multi-Gene Panels versus Familial Mutation Targeted Testing](#).

Special Instructions

- [Informed Consent for Genetic Testing](#)
- [Familial Variant 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](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Blood Spot Collection Instructions](#)

Method Name

Polymerase Chain Reaction (PCR) followed by DNA Sequencing Analysis, Gene Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA), and/or Quantitative Polymerase Chain Reaction (qPCR)

NY State Available

Yes

Specimen

Specimen Type

Varies

Ordering Guidance

This test can only be performed if a variant was previously detected in a family member of this individual. For additional information regarding requests for germline confirmation of somatic results or clinical confirmation of research results, call 800-533-1710.

For answers to frequently asked questions and more information, see [Familial studies](#) on MayoClinicLabs.com.

Additional Testing Requirements

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

If the familial variant was previously identified at an outside laboratory, sending a proband sample (ie, blood or DNA from a family member with a positive genetic test result) to be used as a positive control is strongly recommended to ensure that the specific familial variant can be detected by our laboratory. Proband samples should be sent as a separate

FMTT order, under the proband's identifiers (ie, do not send the patient and proband samples under the same order number). If a positive control is not provided, the possibility of a false-negative result should be considered.

Shipping Instructions

Specimen preferred to arrive within 96 hours of collection.

Necessary Information

The identification of a specific variant in an affected family member is required before this test can be performed for additional family members. If a familial variant has not been previously identified, call 800-533-1710 to discuss testing options.

Familial Variant Testing: Required Patient Information (T721) with documentation of the specific familial variant is required. Testing will be held until information is received. If information is not received within 14 days of sample receipt, testing may be canceled.

Specimen Required

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

Submit only 1 of the following specimen types:

Preferred:

Specimen Type: Whole blood

Container/Tube: Lavender top (EDTA)

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send whole blood specimen in original tube. **Do not aliquot.**

3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 14 days

Additional Information: For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.

Specimen Type: Blood spot

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

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Whatman FTA Classic paper, PerkinElmer 226 filter paper, or 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 older than 1 year is a fingerstick. For detailed instructions, see How to Collect Dried Blood Spot Samples.

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

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

4. Do not stack wet specimens.

5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.

2. For collection instructions, see [Blood Spot Collection Instructions](#)

3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)

4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

Specimen Type: Cultured fibroblasts

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

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

Additional Information: Indicate the tests to be performed on the fibroblast culture cells. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

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

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.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Extracted DNA

Container/Tube: 2 mL screw top tube

Specimen Volume: 100 mcL (microliters)

Collection Instructions:

1. The preferred volume is 100 mcL at a concentration of 250 ng/mcL.

2. Include concentration and volume on tube.

Specimen Stability Information: Frozen (preferred)/Ambient/Refrigerated

Prenatal Specimens

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional information:

1. [A separate culture charge will be assessed under](#) CULAF / Culture for Genetic Testing, Amniotic Fluid.

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

Specimen Type: Cultured amniocytes

Container/Tube: T-25 flask

Specimen Volume: 2 Full flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

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

Specimen Type: Chorionic villi

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

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

Additional Information:

1. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing.

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

Specimen Type: Cultured chorionic villi

Container/Tube: T-25 flasks

Specimen Volume: 2 Full flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

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

Forms

1. [**Familial Mutation Testing: Required Patient Information \(T721\)**](#) is required

2. **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:

-[Informed Consent for Genetic Testing \(T576\)](#)

-[Informed Consent for Genetic Testing-Spanish \(T826\)](#)

3. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:

-[Hematopathology/Cytogenetics Test Request \(T726\)](#)

-[Cardiovascular Test Request \(T724\)](#)

-[Coagulation Test Request \(T753\)](#)

Specimen Minimum Volume

Blood: 1 mL; Amniotic fluid: 10 mL; Chorionic villi: 10 mg

All other specimen types: See Specimen Required

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 & Interpretive**Clinical Information**

This test is available for the analysis of up to 4 genetic variants (single nucleotide variant, small insertion/deletion, or exon level deletion/duplication). Targeted testing is used for diagnostic or predictive testing in family members of an affected individual with a previously detected variant, carrier screening, segregation analysis, confirmation of research results, or testing for germline status of a variant detected by somatic or tumor testing. This test is available for any of the genes on Mayo Clinic Laboratories' (MCL) test menu. In addition, genes not on the MCL test menu may be able to be tested. Call the laboratory at 800-533-1710 with specific inquiries.

Interpretation

Evaluation and categorization of variants are performed using American College of Medical Genetics and Genomics 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-associated variant in an affected family member is necessary before predictive testing for other family members can be performed. Call 800-533-1710 to discuss testing options if a familial variant has not been previously identified.

Familial test results may be included on the test report when relevant to the interpretation of variants in a proband.

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 may be present and 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 the familial condition. **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.

Reclassification of Variants Policy:

Currently, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages healthcare providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

Clinical Reference

1. Richards S, Aziz N, Bale S, et al. ACMG Laboratory Quality Assurance Committee: 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;17(5):405-424

Performance**Method Description**

DNA sequencing and/or dosage analysis by quantitative polymerase chain reaction, array comparative genomic hybridization, or multiplex ligation-dependent probe amplification are utilized to test for the presence of the specific variants previously identified in a family member or via alternate testing (research, somatic). (Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

15 to 22 days

Specimen Retention Time

Whole Blood: 2 weeks (if available); Extracted DNA: 3 months; Blood Spot: 1 year; Cultured fibroblasts, skin biopsy, cord blood, amniotic fluid, cultured amniocytes, chorionic villi, cultured chorionic villi: 1 month

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & 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 account representative. 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. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

The applicable CPT code will be applied on a case-by-case basis.

| Gene | CPT Code (as appropriate) |
|---------------------------|------------------------------|
| Any gene not listed below | 81403 |
| APC | 81202 |
| AR | 81174 |
| BRCA1 | 81215 |
| BRCA2 | 81217 |
| CACNA1A | 81186 |
| CFTR | 81221 |
| CSTB | 81190 |
| FXN | 81289 |
| G6PD | 81248 |
| GJB2 | 81253 |
| HBA1 | 81258 |
| HBA2 | 81258 |
| HBB | 81362 |
| MECP2 | 81303 |
| MLH1 | 81293 |
| MSH2 | 81296 |
| MSH6 | 81299 |
| PALB2 | 81308 |
| PMP22 | 81326 |
| PMS2 | 81318 |
| PTEN | 81322 |
| SMN1 | 81337 |
| TP53 | 81353 |

81265-Maternal cell contamination (if appropriate)

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (if appropriate)

LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|---------|------------------------------------|--------------------|
| FMTT | Familial Variant, 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 | 85069-3 |
| 36535 | Released By | 18771-6 |