
Reporting Title: Familial Variant, Targeted Testing

Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

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 Requirements:

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 Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
36528	Result Summary	Alphanumeric		50397-9
36529	Result	Alphanumeric		82939-0
36530	Interpretation	Alphanumeric		69047-9
36531	Additional Information	Alphanumeric		48767-8
36532	Specimen	Alphanumeric		31208-2
36533	Source	Alphanumeric		31208-2
36534	Method	Alphanumeric		85069-3
36535	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

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)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
MATC C	Maternal Cell Contamination, B	1	81265	No	Yes
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	No	Yes
_G001	Gene GRHPR	1	81403	No	No, (Bill Only)
_G002	Gene PPOX	1	81403	No	No, (Bill Only)
_G003	Gene CFTR	1	81221	No	No, (Bill Only)
_G005	Gene MLH1	1	81293	No	No, (Bill Only)
_G006	Gene MSH2	1	81296	No	No, (Bill Only)
_G007	Gene MSH6	1	81299	No	No, (Bill Only)
_G008	Gene MECP2	1	81303	No	No, (Bill Only)
_G009	Gene MLH3	1	81403	No	No, (Bill Only)
_G010	Gene CHEK2	1	81403	No	No, (Bill Only)
_G011	Gene IDUA	1	81403	No	No, (Bill Only)
_G012	Gene AXIN2	1	81403	No	No, (Bill Only)
_G013	Gene BMPR1A	1	81403	No	No, (Bill Only)

_G014	Gene PTEN	1	81322	No	No, (Bill Only)
_G015	Gene SMAD4	1	81403	No	No, (Bill Only)
_G016	Gene STK11	1	81403	No	No, (Bill Only)
_G017	Gene TP53	1	81353	No	No, (Bill Only)
_G018	Gene IDS	1	81403	No	No, (Bill Only)
_G019	Gene FLCN	1	81403	No	No, (Bill Only)
_G020	Gene SPINK1	1	81403	No	No, (Bill Only)
_G021	Gene PRSS1	1	81403	No	No, (Bill Only)
_G022	Gene CTRC	1	81403	No	No, (Bill Only)
_G025	Gene ABCD1	1	81403	No	No, (Bill Only)
_G026	Gene CDH1	1	81403	No	No, (Bill Only)
_G027	Gene NAGLU	1	81403	No	No, (Bill Only)
_G028	Gene SGSH	1	81403	No	No, (Bill Only)
_G029	Gene ARSB	1	81403	No	No, (Bill Only)
_G030	Gene GNPTAB	1	81403	No	No, (Bill Only)
_G031	Gene SEPT9	1	81403	No	No, (Bill Only)
_G032	Gene ACADVL	1	81403	No	No, (Bill Only)
_G033	Gene ACADM	1	81403	No	No, (Bill Only)
_G034	Gene ACADS	1	81403	No	No, (Bill Only)
_G035	Gene FECH	1	81403	No	No, (Bill Only)
_G036	Gene MAPT	1	81403	No	No, (Bill Only)
_G037	Gene PKHD1	1	81403	No	No, (Bill Only)
_G038	Gene GRN	1	81403	No	No, (Bill Only)
_G039	Gene FTCD	1	81403	No	No, (Bill Only)
_G040	Gene CDKN1C	1	81403	No	No, (Bill Only)
_G041	Gene CPOX	1	81403	No	No, (Bill Only)
_G042	Gene ATP7B	1	81403	No	No, (Bill Only)
_G043	Gene GAA	1	81403	No	No, (Bill Only)
_G044	Gene HMBS	1	81403	No	No, (Bill Only)
_G045	Gene GALT	1	81403	No	No, (Bill Only)
_G046	Gene GLA	1	81403	No	No, (Bill Only)
_G047	Gene BTD	1	81403	No	No, (Bill Only)
_G048	Gene HEXA	1	81403	No	No, (Bill Only)
_G049	Gene AGXT	1	81403	No	No, (Bill Only)
_G050	Gene APC	1	81202	No	No, (Bill Only)
_G051	Gene MLYCD	1	81403	No	No, (Bill Only)
_G052	Gene MMACHC	1	81403	No	No, (Bill Only)
_G053	Gene GBA	1	81403	No	No, (Bill Only)
_G054	Gene SMPD1	1	81403	No	No, (Bill Only)
_G055	Gene CPT2	1	81403	No	No, (Bill Only)
_G056	Gene TTR	1	81403	No	No, (Bill Only)
_G057	Gene UBE3A	1	81403	No	No, (Bill Only)
_G058	Gene GALC	1	81403	No	No, (Bill Only)

_G059	Gene GSN	1	81403	No	No, (Bill Only)
_G060	Gene LYZ	1	81403	No	No, (Bill Only)
_G061	Gene FGA	1	81403	No	No, (Bill Only)
_G062	Gene APOA1	1	81403	No	No, (Bill Only)
_G063	Gene APOA2	1	81403	No	No, (Bill Only)
_G064	Gene MMADHC	1	81403	No	No, (Bill Only)
_G065	Gene SLC25A20	1	81403	No	No, (Bill Only)
_G066	Gene ARSA	1	81403	No	No, (Bill Only)
_G067	Gene NPC1/2_SEQ and NPC1/2_MLPA	1	81403	No	No, (Bill Only)
_G068	Gene PMS2	1	81318	No	No, (Bill Only)
_G070	Gene RAI1	1	81403	No	No, (Bill Only)
_G071	Gene MUTYH	1	81403	No	No, (Bill Only)
_G072	Gene HGSNAT	1	81403	No	No, (Bill Only)
_G073	Gene GNS and GRHPR_MLPA	1	81403	No	No, (Bill Only)
_G074	Gene PSAP	1	81403	No	No, (Bill Only)
_G077	Gene RET	1	81403	No	No, (Bill Only)
_G078	Gene SUMF1	1	81403	No	No, (Bill Only)
_G079	Gene CASR_Seq	1	81403	No	No, (Bill Only)
_G080	Gene VHL_SEQ	1	81403	No	No, (Bill Only)
_G084	Gene SDHB, SDHC, SDHD_Seq	1	81403	No	No, (Bill Only)
_G085	Gene BRCA1	1	81215	No	No, (Bill Only)
_G086	Gene BRCA2	1	81217	No	No, (Bill Only)
_G087	Gene DMD_MLPA	1	81403	No	No, (Bill Only)
_G089	Gene MPZ_MLPA	1	81403	No	No, (Bill Only)
_G102	Gene SERPINA1			No	No, (Bill Only)
_G112	Gene SDHAF2	1	81403	No	No, (Bill Only)
_G113	Gene TMEM127	1	81403	No	No, (Bill Only)
_G114	Gene MAX	1	81403	No	No, (Bill Only)
_G115	Gene SMN1	1	81337	No	No, (Bill Only)
_G125	Gene PMP22	1	81326	No	No, (Bill Only)
_G126	Gene G6PD	1	81248	No	No, (Bill Only)
_G127	Gene GJB2	1	81253	No	No, (Bill Only)
_G128	Gene HBA1/HBA2	1	81258	No	No, (Bill Only)
_G129	Gene HBB	1	81362	No	No, (Bill Only)
_G130	Known Familial Variant,Other			No	No, (Bill Only)
G168	Gene CSTB	1	81190	No	No, (Bill Only)
G169	Gene CACNA1A	1	81186	No	No, (Bill Only)
_STR1	Comp Analysis using STR (Bill only)	1	81265	No	No, (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)	1	81266	No	No, (Bill only)
G230	Gene AR	1	81174	No	No, (Bill Only)
G231	Gene FXN	1	81289	No	No, (Bill Only)
G232	Gene PALB2	1	81308	No	No, (Bill Only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2
MATCC	53291	Released By	Alphanumeric		18771-6
MATCC	55150	Method	Alphanumeric		85069-3

Reference Values: