

Reporting Title: Carrier Screen, Focused Panel
Performing Location: Rochester

Ordering Guidance:
This test is specifically for carrier screening purposes and is not intended for diagnostic purposes.

If the reproductive partner is also having this test performed, call the lab for a revised risk assessment.

Targeted testing for familial variants (also called site-specific or known mutation testing) is available for all genes on this panel under FMTT / Familial Variant, Targeted Testing, Varies. Call 800-533-1710 to obtain more information about this testing option.

Shipping Instructions:
Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:
Specimen Type: Whole blood
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.
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 whole blood specimen in original tube. **Do not aliquot.**
Additional Information: To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

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:
-[Informed Consent for Genetic Testing](#) (T576)
-[Informed Consent for Genetic Testing-Spanish](#) (T826)
2. [Molecular Genetics: Congenital Inherited Diseases Patient Information](#) (T521)

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
608337	Result Summary	Alphanumeric		50397-9

608338	Result	Alphanumeric		82939-0
608339	Additional Results	Alphanumeric		82939-0
608340	Offspring Risk	Alphanumeric		In Process
608341	Clinical Summary	Alphanumeric		55752-0
608342	Additional Information	Alphanumeric		48767-8
608343	Other Identified Alleles	Alphanumeric		In Process
608344	Method	Alphanumeric		85069-3
608345	Disclaimer	Alphanumeric		62364-5
608346	Specimen	Alphanumeric		31208-2
608347	Source	Alphanumeric		31208-2
608348	Released By	Alphanumeric		18771-6

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

Supplemental Report:
Supplemental

CPT Code Information:

- 81220
- 81329
- 81479
- 81257
- 81361
- 81222
- 81479 (if appropriate for government payers)

Reference Values:

An interpretive report will be provided.