
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 Mutation, 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)

3. Targeted Variants Detected by Focused Carrier Screening Tests

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		In Process
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:

No

CPT Code Information:

81220
81329
81479
81257
81361
81222

Reference Values:

An interpretive report will be provided.