

Reporting Title: CF and SMA Carrier Screen Panel

Performing Location: Rochester

Ordering Guidance:

This test is specifically for carrier screening purposes and is not intended for diagnostic purposes. For diagnostic testing, order CFMP / Cystic Fibrosis, CFTR Gene, Variant Panel, Varies.

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.

Necessary Information:

If there is a family history of cystic fibrosis (CF) or spinal muscular atrophy (SMA), the known genetic variant in the family should be supplied for best interpretation of results.

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.

Specimen Minimum Volume:

1 mL

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®
608350	Result Summary	Alphanumeric		50397-9
608351	Result	Alphanumeric		82939-0
608352	Interpretation	Alphanumeric		69047-9
608353	Additional Information	Alphanumeric		48767-8
608354	Method	Alphanumeric		85069-3
608355	Specimen	Alphanumeric		31208-2
608356	Source	Alphanumeric		31208-2
608357	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81220
81329
81222
81479 (if appropriate for government payers)



Reference Values:

An interpretive report will be provided.