

**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.

- 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		

Test Definition: CFSMN

Cystic Fibrosis and Spinal Muscular Atrophy  
Carrier Screen Panel, Varies

	Refrigerated		
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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.