

Reporting Title: Cystic Fibrosis (CF) Mutation Panel
Performing Location: Rochester

Ordering Guidance:
If testing is negative, and a diagnosis of cystic fibrosis is still suspected, consider CFTRZ / *CFTR* Gene, Full Gene Analysis, Varies.

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, the known 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		
	Refrigerated		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
606027	Result Summary	Alphanumeric		50397-9
606028	Result	Alphanumeric		82939-0
606029	Interpretation	Alphanumeric		69047-9
606030	Additional Information	Alphanumeric		48767-8
606031	Method	Alphanumeric		85069-3
606032	Specimen	Alphanumeric		31208-2
606033	Source	Alphanumeric		31208-2
606034	Released By	Alphanumeric		18771-6

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

Supplemental Report:

No

CPT Code Information:

- 81220
- 81222
- 81479 (if appropriate for government payers)

Reference Values:

An interpretive report will be provided.