

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 CFTRN / Cystic Fibrosis Transmembrane Conductance Regulator, *CFTR*, 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.

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
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: None
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:
1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. 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) | | |
| | Refrigerated | | |
| | Frozen | | |

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

Reference Values:

An interpretive report will be provided.