
Reporting Title: CFTR Gene, Full Gene Analysis**Performing Location: Rochester****Ordering Guidance:**

This test is not the preferred test for cystic fibrosis carrier screening. See CFMP / Cystic Fibrosis, CFTR Gene, Variant Panel, Varies.

Targeted testing for familial variants (also called site-specific or known variants testing) is available for variants identified in the CFTR gene. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about testing option, call 800-533-1710.

Additional Testing Requirements:

All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen. This must be a different order number than the prenatal specimen.

For cord blood specimens: Maternal cell contamination (MCC) studies are available. Order MATCC on both the cord blood and maternal specimens under separate order numbers. Cord blood testing will proceed without MCC studies, but results may be compromised if MCC is present.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

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.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblasts

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional information:

1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks is required to culture amniotic fluid before genetic testing can occur.

2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

Additional Information:

1. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 2 to 3 weeks is required to culture chorionic villi before genetic testing can occur.

2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Confluent cultured cells

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information: All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

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	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No
CULAF	CG767	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619775	Test Description	Alphanumeric		62364-5
619776	Specimen	Alphanumeric		31208-2
619777	Source	Alphanumeric		31208-2
619778	Result Summary	Alphanumeric		50397-9
619779	Result	Alphanumeric		82939-0
619780	Interpretation	Alphanumeric		69047-9
619781	Additional Results	Alphanumeric		82939-0
619782	Resources	Alphanumeric		99622-3
619783	Additional Information	Alphanumeric		48767-8
619784	Method	Alphanumeric		85069-3
619785	Genes Analyzed	Alphanumeric		82939-0
619786	Disclaimer	Alphanumeric		62364-5
619787	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81223

88233- Tissue culture, skin, solid tissue biopsy (if appropriate)

88240- Cryopreservation (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes
MATCC	Maternal Cell Contamination, B			No	Yes
_STR1	Comp Analysis using STR (Bill only)			No	No (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)			No	No (Bill only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2
MATCC	55150	Method	Alphanumeric		85069-3
MATCC	53291	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided