

GeneSeq PLUS, CFTR

Order Name: **GeneSeq P/CFTR**
Test Number: 5194965
Revision Date: 03/21/2023

TEST NAME	METHODOLOGY	LOINC CODE
GeneSeq PLUS, CFTR	Polymerase Chain Reaction	

SPECIMEN REQUIREMENTS

Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	8.5 mL (3 mL)	Whole Blood	ACD Solution A or B (Yellow Top)	Room Temperature
Alternate 1	8.5 mL (3 mL)	Whole Blood	EDTA (Lavender Top)	Room Temperature
Alternate 2	1	Saliva	Oragene Dx saliva kit	Room Temperature
Alternate 3	1	Buccal swab	PurFlock buccal swab kit	Room Temperature

Instructions

Specimen Type: Whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
Specimen Volume: 8.5 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
Minimum Volume: 3 mL whole blood or PurFlock buccal swab kit or Oragene Dx saliva kit
Collection: Standard phlebotomy. Follow PurFlock buccal swab kit or Oragene Dx 500 saliva kit collection instructions. Do not eat, drink, smoke, or chew gum 30 min prior to collection.
Specimen Storage: Maintain specimen at room temperature or refrigerate at 4C Do not freeze.
Special Instructions: Variants of uncertain significance (VUS) will be reported unless VUS opt out is indicated on the requisition. If requesting full gene sequencing for multiple genes, order **GeneSeq PLUS [5194962]**. To test fetal specimens, including cordblood, order test code **GeneSeq PLUS, Fetal Analysis [5194963]** Test orders must include an attestation that the provider has the patient's informed consent for genetic testing. See sample physician office consent form: Consent for Genetic Testing.

GENERAL INFORMATION

Expected TAT	14 - 21 days In some cases, additional time may be required for confirmatory or reflex tests.
Clinical Use	This test is used for carrier and diagnostic testing for cystic fibrosis. Next-generation sequencing: Identifies genetic variants, including small nucleotide variants (SNVs), insertions, deletions and copy number variants (CNVs). Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants or repeat expansions. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples or erroneous representation of family relationships.
Performing Labcorp Test Code	482449
Notes	Labcorp Test Code: 482449
Lab Section	Reference Lab