Inheritest(R) CF/SMA Panel

Order Name: **CF/SMA WB Panel**
Test Number: 5194938
Revision Date: 03/21/2023

TEST NAME	METHODOLOGY	LOINC CODE
Inheritest(R) CF/SMA Panel	See Test Notes	

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
Mininum 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: In cases in which there is a known variant documented in the family, the physician may prefer to order Targeted Variant Analysis, test code 482552. Test orders must include an attestation that the provider has the patient's informed consent for genetic

GENERAL INFORMATION

Expected TAT

14 - 21 days In some cases, additional time may be required for confirmatory or reflex tests.

Notes

Clinical Questionnaire for Inheritest® Carrier Screen and GeneSeq® PLUS
Methodology
Cystic fibrosis: Next-generation sequencing to identify genetic variants, including small nucleotide variants (SNVs), insertions, deletions and copy number variants (CNVs). Spinal muscular atrophy (SMA): Copy number assessment of SMN1 exon 7 by quantitative polymerase chain reaction (qPCR). For carrier screening, when two copies of SMN1 are detected, allelic discrimination qPCR targeting c.*3+80T>G in SMN1 is performed. The presence or absence of c.*3+80T>G correlates with an increased or decreased risk, respectively, of being a silent carrier (2+0).

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