Spinal Muscular Atrophy (SMA) Whole Blood

Order Name: **SMA WB**
Test Number: 5194936
Revision Date: 03/21/2023

TEST NAME	METHODOLOGY	LOINC CODE
Spinal Muscular Atrophy (SMA) Whole Blood	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**	**Buccal swab**	**PurFlock buccal swab kit**	**Room Temperature**
Alternate 3	**1**	**Saliva**	**Oragene Dx saliva 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: To test fetal specimens, including cordblood, order test code 481651, Spinal Muscular Atrophy (SMA), Fetal Analysis Test orders must include an attestation that the provider has the patient's informed consent for genetic testing.
Cause for Rejection: Frozen or hemolyzed specimen; quantity not sufficient for analysis; improper container or blood specimens more than four days post draw.

GENERAL INFORMATION

Expected TAT

8 - 14 days In some cases, additional time may be required for confirmatory or reflex tests.

Notes

Labcorp Test Code: 481630
Methodology
Copy number assessment of SMN1 exon 7 by quantitative polymerase chain reaction (qPCR); reflex testing to SMN2 copy number analysis is performed for individuals with 0 copies of SMN1. 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).

Service Provided By

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