


Spinal Muscular Atrophy (SMA), Fetal

Order Name: **SMA Fetal**
Test Number: **5194937**
Revision Date: **03/21/2023**

TEST NAME	METHODOLOGY	LOINC CODE
Spinal Muscular Atrophy (SMA), Fetal	See Test Notes	n/a

SPECIMEN REQUIREMENTS				
Specimen	Specimen Volume (min)	Specimen Type	Specimen Container	Transport Environment
Preferred	4 mL (3 mL)	Amniotic Fluid	Sterile Screwtop Container	Room Temperature
Alternate 1	See Instructions (See Instructions)	See Instructions	See Instructions	Room Temperature
Instructions	<p>Specimen Type: Cultured amniotic fluid or cultured chorionic (CVS) cells, or cordblood, is required for testing. Direct amniotic fluid or CVS specimen may be submitted; additional culture fee may be applied.</p> <p>Specimen Volume: One confluent T-25 flask or 4 mL cordblood. If amniotic fluid or CVS are cultured at another facility, please maintain back-up cultures.</p> <p>Minimum Volume: One confluent T-25 flask or 3 mL cordblood</p> <p>Collection: Standard sterile techniques. Transfer aseptically to sterile tubes. Amniotic fluid: Discard first 2 mL of fluid aspirated to avoid maternal cell contamination.</p> <p>Specimen Storage: Maintain specimen at room temperature. Do not freeze.</p> <p>Special Instructions: Test orders must include an attestation that the provider has the patient's informed consent for genetic testing.</p>			

GENERAL INFORMATION	
Expected TAT	14 - 21 days In some cases, additional time may be required for confirmatory or reflex tests.
Notes	<p>Labcorp Test Code: 481651</p> <p>Methodology</p> <p>Spinal muscular atrophy (SMA): Copy number assessment of SMN1 exon 7 by quantitative polymerase chain reaction (qPCR). Reflex testing to SMN2 copy number analysis is performed for affected fetus with 0 copies of SMN1. Maternal cell contamination analysis (MCC): Analysis of short tandem repeat markers by multiplex fluorescent polymerase chain reaction (PCR) and capillary electrophoresis.</p>
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