

**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	<b>4 mL (3 mL)</b>	<b>Amniotic Fluid</b>	<b>Sterile Screwtop Container</b>	<b>Room Temperature</b>
Alternate 1	<b>See Instructions (See Instructions)</b>	<b>See Instructions</b>	<b>See Instructions</b>	<b>Room Temperature</b>
Instructions	<p><b>Specimen Type:</b> 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><b>Specimen Volume:</b> 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><b>Minimum Volume:</b> One confluent T-25 flask or 3 mL cordblood</p> <p><b>Collection:</b> Standard sterile techniques. Transfer aseptically to sterile tubes. Amniotic fluid: Discard first 2 mL of fluid aspirated to avoid maternal cell contamination.</p> <p><b>Specimen Storage:</b> Maintain specimen at room temperature. Do not freeze.</p> <p><b>Special Instructions:</b> 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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