

Spinal Muscular Atrophy (SMA) – Diagnostic testing

ORDER NAME

Spinal Muscular Atrophy (SMA) Dx Test – SMN1 & SMN2 Copy Number (EPIC Order Code=LAB14955)

- If EPIC is not an option, complete and submit a paper requisition to Genomicslab@upmc.edu
Paper requisition can be found on the INFONET. Search for “genetics requisition form.”

BACKGROUND

Spinal muscular atrophy (SMA) is an autosomal recessive disorder characterized by muscle weakness and atrophy due to progressive degeneration and loss of the anterior horn cells in the spinal cord and brain stem. The disease is classified into subtypes based on age of onset and maximal function.

The spinal muscular atrophy diagnostic test determines if an individual or fetus is affected with spinal muscular atrophy.

This diagnostic test can be conducted postnatally or prenatally, utilizing amniocytes or chorionic samples. If the clinical indication is diagnosis of a pregnancy, and screening tests of the parents were performed outside of the UCGL laboratory, collection of maternal and paternal samples is required to confirm familial variants. Maternal Cell Contamination (MCC) studies are also required.

INDICATIONS FOR TESTING

- Abnormal fetal ultrasound suspicious of a diagnosis of SMA
- Confirm a suspected diagnosis of SMA

RESULT

95-98% of cases are caused by a homozygous deletion of the SMN1 gene while 2-5% have a sequence variant in the one remaining copy of SMN1. SMN2 copy number is also performed. SMN2 copy number inversely correlates with disease severity.

METHOD

Quantitative droplet digital PCR and allele-specific fluorescent hydrolysis probes.

LIMITATIONS

Carriers with two copies of SMN1 on one chromosome and zero copies on the second chromosome or carriers with pathogenic sequence variants will not be identified by this screening assay.

SPECIMEN REQUIREMENTS

- Postnatal: Whole blood – EDTA tube required, 3-5 ml
- Prenatal: Amniocytes or chorionic samples
 - Maternal Cell Contamination studies: Whole blood – EDTA tube required, 3-5 ml

TURNAROUND TIME

14 days

CPT CODE

81329