Spinal Muscular Atrophy (SMA) – Carrier screening

ORDER NAME
Spinal Muscular Atrophy Carrier Screen (EPIC Order Code=LAB12496)

- 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.

Spinal Muscular Atrophy Carrier Screen tests for the copy number of the SMN1 gene.

INDICATIONS FOR TESTING
SMA carrier screening is recommended by the American College of Medical Genetics and Genomics and the American College of Obstetricians and Gynecologists and is routinely offered to women who are currently pregnant. Other indications include:

- Preconception carrier screening /sperm and egg donor carrier screening
- Carrier identification for reproductive partner of known SMA carrier
- Carrier identification in individuals with a positive family history
- Abnormal fetal ultrasound

RESULT
Unaffected carriers have a heterozygous deletion in the SMN1 gene.

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
- Whole blood – EDTA tube required, 3-5 ml
- Previously extracted DNA (concentration >25 ng/ul, volume >20 ul, minimum of 1 ug total DNA, 260/280>1.7)
- Saliva provided in Oragene (OGD-500) collection kits

TURNAROUND TIME
14 days

CPT CODE
81329