TEST CHANGE

Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal

2013444, SMA DD FE

Specimen Requirements:	
Patient Preparation:	
Collect:	Cultured amniocytes or Cultured CVS AND Maternal Whole Blood Specimen: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation:	Cultured Amniocytes or Cultured CVS: Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80 percent confluence (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission. Maternal Whole Blood Specimen: Transport 2 mL whole blood. (Min: 1 mL)
Transport Temperature:	Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells. Maternal Whole Blood Specimen: Room temperature.
Unacceptable Conditions:	
Remarks:	Please contact an ARUP genetic counselor at 800-242-2787 x2141 prior to sample submission. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.
Stability:	Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable Maternal Whole Blood Specimen: Room Temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable
Methodology:	Multiplex Ligation-dependent Probe Amplification
Performed:	Varies
Reported:	Within 10 days
Note:	~~Note: Maternal specimens are recommended as controls for proper test interpretation. Submit maternal blood specimen for Maternal Cell Contamination in addition to fetal specimen.~~

CPT Codes:	81329; 81265 Fetal Cell Contamination (FCC)
New York DOH Approval Status:	This test is New York DOH approved.

Interpretive Data:

Background information for Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal Characteristics: Spinal muscular atrophy (SMA) is the most common lethal genetic disease in children. It is characterized by progressive muscle atrophy and weakness, poor weight gain, restrictive lung disease, scoliosis, and joint contractures due to degeneration of lower motor neurons and brain stem nuclei. Onset ranges from before birth to young adulthood and severity is highly variable. Individuals with SMA have no functional copies of the *SMN1* gene either due to homozygous loss of *SMN1* from deletion or gene conversion (95 percent) or loss of one *SMN1* gene and a pathogenic sequence variant in the other (5 percent). The *SMN2* gene produces a small amount of functional survival motor neuron protein compared to *SMN1*. An increased number of SMN2 gene copies may reduce disease severity but phenotype cannot be predicted with certainty.
Inheritance: Autosomal recessive.
Cause: Pathogenic variants in the *SMN1* gene.
Variants Tested: For copy number: *SMN1* (NM_000344.3) exon 7 c.840C and exon 8 c.*239G, and *SMN2* (NM_017411.3) exon 7 c.840T.
Clinical sensitivity: 95-98 percent.
Methodology: Multiplex probe ligation-dependent amplification (MLPA).
Analytical sensitivity and specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Single base pair substitutions, small deletions/duplications, regulatory region and deep intronic variants will not be detected. This test is unable to determine chromosomal phase of *SMN1* or *SMN2* copies.

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Reference Interval:

By report

HOTLINE NOTE: There is a component change associated with this test. One or more components have been added or removed. Refer to the Hotline Test Mix for interface build information.