Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: ARUP, ARUP

Patient: Pos, SMADDFE

DOB	
Sex:	Female
Patient Identifiers:	46723
Visit Number (FIN):	47059
Collection Date:	2/22/2023 08:47

Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal

ARUP test code 2013444

Maternal Contamination Study Fetal Spec	Fetal Cells Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	Whole Blood
SMA Copy Number, Specimen	Cultured Amnio
SMA Copy Number, SMN1 Copies	**0 copies** *
SMA Copy Number, SMN2 Copies	2 copies
SMA Copy Number, Interp	See Note Indication for testing: Prenatal diagnosis. Result: SMN1 gene copies: 0 copies SMN2 gene copies: 2 copies Interpretation: No copies of the SMN1 gene were detected by multiplex ligation-dependent probe amplification (MLPA) in this prenatal sample; therefore, this fetus is predicted to be affected with spinal muscular atrophy (SMA). 2 copies of the SMN2 gene was/were detected by MLPA. Although SMN2 copy number is inversely correlated with disease severity, it cannot be used to predict phenotype with certainty. Clinical findings and disease severity are variable. Recommendations: Genetic consultation is indicated, including a discussion of medical screening and management. Adult family members should be offered SMA carrier screening. BACKGROUND INFORMATION: 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

H=High, L=Low, *=Abnormal, C=Critical

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.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	23-053-101334	2/22/2023 8:47:00 AM	2/22/2023 8:47:40 AM	2/22/2023 11:13:00 AM
Maternal Contam Study, Maternal Spec	23-053-101334	2/22/2023 8:47:00 AM	2/22/2023 8:47:40 AM	2/22/2023 11:13:00 AM
SMA Copy Number, Specimen	23-053-101334	2/22/2023 8:47:00 AM	2/22/2023 8:47:40 AM	2/22/2023 11:13:00 AM
SMA Copy Number, SMN1 Copies	23-053-101334	2/22/2023 8:47:00 AM	2/22/2023 8:47:40 AM	2/22/2023 11:13:00 AM
SMA Copy Number, SMN2 Copies	23-053-101334	2/22/2023 8:47:00 AM	2/22/2023 8:47:40 AM	2/22/2023 11:13:00 AM
SMA Copy Number, Interp	23-053-101334	2/22/2023 8:47:00 AM	2/22/2023 8:47:40 AM	2/22/2023 11:13:00 AM

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