ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: 30 years Female

Specimen Collected: 09-Feb-26 14:07

SMA Copy Number, Fetal **|Received: 09-Feb-26 14:09** **Report/Verified: 09-Feb-26 14:27**

Procedure	Result	Units	Reference Interval
Maternal Contamination Study Fetal Spec	Fetal Cells [f1]		
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 [f2 i1]		

Result Footnote

f1: Maternal Contamination Study Fetal Spec

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

f2: SMA Copy Number, Interp

Indication for testing: Prenatal diagnosis.

Result:
SMN1 gene copies: 0
SMN2 gene copies: 2 copies

Interpretation: According to information provided to ARUP, the mother and father of this fetus are both carriers of spinal muscular atrophy (SMA), each harboring one copy of the SMN1 gene. 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). Two 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.

This result has been reviewed and approved by [redacted]

Test Information

i1: SMA Copy Number, Interp

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 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

Test Information

i1: SMA Copy Number, Interp

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 ligation-dependent probe 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. SMN2 copy numbers greater than 3 may not be reliable distinguished. 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 U.S. 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.