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PATIENT REPORT

Patient Age/Sex: Male

Specimen Collected: 04-Jun-24 09:08

Familial Targeted Sequencing, Fetal | **Received: 04-Jun-24 09:08** **Report/Verified: 04-Jun-24 09:21**

Procedure	Result	Units	Reference Interval
Maternal Contamination Study Fetal Spec	Fetal Cells		
Maternal Contam Study,Maternal Spec	Whole Blood		
FAM NGS FE Specimen	Cultured Amnio		
FAM FE Interp	Negative [i1]		

Test Information

i1: FAM FE Interp

BACKGROUND INFORMATION: Familial Targeted Sequencing, Fetal

METHODOLOGY: Probe hybridization-based capture of all coding exons and exon-intron junctions of the targeted gene(s) region(s), followed by massively parallel sequencing. Variants in genes, other than the gene(s) region(s) specifically requested, were not evaluated. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. Specificity is greater than 99.9 percent for all variant classes.

LIMITATIONS: A negative result does not exclude all genetic diagnoses in this fetus. This test only evaluates the specified familial variant(s) of interest and other pathogenic or likely pathogenic variants by massively parallel sequencing related to the condition of interest within the targeted gene(s) region(s). Refer to Targeted Sequencing Gene List for complete list of genes available for this test and any gene-specific technical limitations. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Regulatory region variants, deep intronic variants, and large deletions/duplications will not be identified. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) mutations, aneuploidies, or repeat expansions. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

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

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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.