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PATIENT REPORT

Patient Age/Sex: Female

Specimen Collected: 04-Jun-24 08:42

DNA Extract and Hold	**|Received: 04-Jun-24 08:42**		**Report/Verified: 04-Jun-24 08:49**
Procedure	**Result**	**Units**	**Reference Interval**
DNA Extract and Hold	Complete [i1]		

Whole Genome Sequencing	**|Received: 04-Jun-24 08:42**		**Report/Verified: 04-Jun-24 08:49**
Procedure	**Result**	**Units**	**Reference Interval**
WGS NGS Int	See Note [i2]		

Test Information

i1: DNA Extract and Hold

INTERPRETIVE INFORMATION: DNA Extract and Hold

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.

i2: WGS NGS Int

BACKGROUND INFORMATION: Whole Genome Sequencing

CHARACTERISTICS: The purpose of whole genome sequencing is to determine the patient's diagnosis when a genetic condition is suspected. The analyzed genome includes exons from all known human nuclear genes and all intronic variants suspected of influencing splicing. Parental samples are recommended for interpretation of results.

CLINICAL SENSITIVITY: Varies based on clinical symptoms, family history, inheritance pattern, and previous clinical evaluations.

METHODOLOGY: Genomic DNA is extracted from whole blood, prepared into libraries, then sequenced by massively parallel sequencing (MPS; also known as next generation sequencing [NGS]). Variant calling is performed using a custom bioinformatics pipeline that includes phenotype-based scores. Human genome build 19 (Hg 19) is used for data analysis.

ANALYTICAL SENSITIVITY: The analytical sensitivity of this test is 98.6 percent for single nucleotide variants (SNVs). Analytical sensitivity is 97.4 percent for insertions/duplications /deletions ranging in size from 1-15 bp, and 92.0 percent for those 16-50 bp in size.

LIMITATIONS OF ANALYSIS: A negative result does not exclude all genetic diagnoses. The human genome is not able to be completely analyzed as some genes have not been identified while others, due to technical limitations, cannot be sequenced or interpreted. Variants in intergenic or deep intronic regions will only be evaluated if an effect on gene expression is predicted via annotation software. Mode of inheritance, reduced penetrance, and genetic heterogeneity can reduce clinical sensitivity. Large deletions/duplications/insertions are not assessed by massively

Test Information

i2: WGS NGS Int

parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. Mitochondrial DNA (mtDNA) is not analyzed. This assay is not designed to detect low-level somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Please see Additional Technical Information located at http://ltd.aruplab.com/Tests/Pub/3016493 for more information.

LIMITATIONS FOR REPORTING AND INTERPRETATION: Only variants in genes suspected to be causative of the patient's symptoms are reported, with the exception of secondary pathogenic findings, if elected. Incorrect reporting of biological relationships among family members may affect result interpretation. Mode of inheritance, reduced penetrance, and genetic heterogeneity can reduce clinical sensitivity. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.

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