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PATIENT REPORT

Patient Age/Sex: Male

Specimen Collected: 04-Jun-24 08:42

DNA Extract and Hold	**Received: 04-Jun-24 08:42**		**Report/Verified: 04-Jun-24 08:50**
Procedure	**Result**	**Units**	**Reference Interval**
DNA Extract and Hold	Complete [i1]		

WGS Sequencing, Familial Control	**Received: 04-Jun-24 08:42**		**Report/Verified: 04-Jun-24 08:50**
Procedure	**Result**	**Units**	**Reference Interval**
WGS FRPT 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 FRPT Int

BACKGROUND INFORMATION: Whole Genome Sequencing, Familial Control

CHARACTERISTICS: The analyzed genome includes all exons from all known human nuclear genes and all intronic variants suspected of influencing splicing. These regions are sequenced to identify the cause(s) of a disorder in a family member. The American College of Medical Genetics (ACMG) recommends analysis of certain genes for secondary findings in all individuals undergoing genome sequencing. Please refer to ACMG Secondary Findings Gene List (http://ltd.aruplab.com/Tests/Pub/3016497) for an up-to-date list of genes analyzed. Note that this gene list is updated periodically and is only accurate for this sample at the time of reporting. Please contact an ARUP genetic counselor (800-242-2787 ext. 2141) for clarification regarding genes analyzed.

INHERITANCE: Varies depending on the specific gene and variant

CLINICAL SENSITIVITY: Varies by gene

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.

LIMITATIONS OF ANALYSIS: Deletions/duplications/insertions of any size may not be detected by massively 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. This assay is not designed to detect low-level somatic variants associated with disease.

Test Information

i2: WGS FRPT Int

Interpretation of this test result may be impacted if this individual has had an allogeneic stem cell transplantation. Mode of inheritance, reduced penetrance, and genetic heterogeneity could reduce the clinical sensitivity.

LIMITATIONS OF REPORTING: Secondary pathogenic findings, including variants identified in genes on the ACMG-recommended panel or other medically actionable variants at ARUP's discretion, are reported. Variants of unknown significance will not be reported. Single pathogenic variants in autosomal recessive genes will not be reported.

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