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PATIENT REPORT

Patient Age/Sex: Female

Specimen Collected: 25-May-23 14:03

Exome Seq, Familial Control |Received: 25-May-23 14:04 Report/Verified: 26-May-23 07:00

Procedure	Result	Units	Reference Interval
EXOME FRPT Int	Uncertain [i1]		

Test Information

i1: EXOME FRPT Int

BACKGROUND INFORMATION: Exome Sequencing, Familial Control

CHARACTERISTICS: The analyzed exome includes all exons from all known human nuclear genes and accounts for approximately 1-2 percent of the human genome. 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/3016589) 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. Mode of inheritance, reduced penetrance, and genetic heterogeneity can reduce the clinical sensitivity.

METHODOLOGY: Targeted capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to confirm reported variants. Human genome build 19 (Hg 19) was used for data analysis.

LIMITATIONS OF ANALYSIS: Not all pathogenic variants occur in the coding regions of genes. Some genes, or parts of genes, may not be adequately sequenced to allow for confident analysis. The following types of variants may not be detectable: those located in genes with corresponding pseudogenes, those in repetitive or high GC rich regions, large deletions / duplications / rearrangements, and mosaic variants. Rare variants in probe hybridization sites may compromise analytical 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.

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

Test Information

i1: EXOME FRPT Int

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab