Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: TEST,

Patient: NEG EXAMPLE, WGS FRPT

DOB
Sex: Male
Patient Identifiers: 49775
Visit Number (FIN): 50130
Collection Date: 6/12/2023 07:53

Whole Genome Sequencing, Familial Control

ARUP test code 3016497

WGS FRPT Int	Negative

RESULT
No secondary pathogenic variants were detected.

INTERPRETATION
The American College of Medical Genetics and Genomics (ACMG) recommends analysis of specific genes in all individuals undergoing genome sequencing even though these variants may not be related to the key clinical findings (Miller, 2022). Although no known secondary pathogenic variants were identified in the v3.1 list of ACMG-recommended genes in this individual, this result does not exclude the possibility this individual may carry a pathogenic variant in one of these genes, or in another gene that is not included on this list. Note that single pathogenic variants in autosomal recessive ACMG genes are not reported. The genes on the ACMG-recommended list for reporting are evaluated to the extent that standard genome sequencing will allow, and the clinical significance of the variants detected are evaluated using evidence from current literature and variant databases.

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. If there is clinical suspicion or family history of a genetic condition associated with one of the ACMG-recommended genes, additional targeted testing should be considered as genome sequencing will not identify all pathogenic variants in these genes.

REFERENCE
Miller DT, et al. ACMG SF v3.1 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2022;24(7):1407-1414. PMID: 35802134.

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

H=High, L=Low, *=Abnormal, C=Critical

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. 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.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
WGS FRPT Int	23-163-100760	6/12/2023 7:53:00 AM	6/12/2023 7:53:11 AM	6/12/2023 7:57:00 AM

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical