

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

Physician: TEST,

**Patient: NEGATIVE, EXOME FRPT**

**DOB**

**Sex:** Male

**Patient Identifiers:** 51721

**Visit Number (FIN):** 52108

**Collection Date:** 8/21/2023 07:39

## Exome Sequencing, Familial Control

ARUP test code 3016589

### EXOME 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 exome 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 exome 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 exome 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: 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://ftd.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

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: NEGATIVE, EXOME FRPT  
ARUP Accession: 23-233-100748  
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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 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
EXOME FRPT Int	23-233-100748	8/21/2023 7:39:00 AM	8/21/2023 7:39:52 AM	8/21/2023 7:43:00 AM

END OF CHART

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