Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
Sex:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Rapid Genome Sequencing, Familial Comparator

ARUP test code 3019953

RWGS FM Interp: Negative

TEST PERFORMED
Rapid Genome Sequencing, Familial Comparator

RESULT
No secondary findings variants were detected.

INTERPRETATION
The American College of Medical Genetics and Genomics (ACMG) recommends analysis of specific, medically actionable secondary findings (SF) genes in all consented individuals undergoing genome sequencing even though these variants may not be related to the indication for testing (1). Although no secondary findings variants involving genes from the ACMG SF v3.3 list (2) were identified in this individual, this result does not exclude the possibility this individual may carry a pathogenic variant involving one of these genes, or another gene that is not included on this list. If there is clinical suspicion or family history of a genetic condition associated with one of the ACMG SF genes, additional targeted testing should be considered as genome sequencing will not identify all pathogenic variants involving these genes. Note that single pathogenic variants in autosomal recessive ACMG SF genes are not reported. Please refer to the background information below for the methodology and limitations of this test.

RECOMMENDATIONS
- Medical management should rely on clinical findings and family history.

References
1: Miller DT, Lee K, Gordon AS, et al. Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2021 update: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021. PMID: 34012069.
2: Lee K, Abul-Husn NS, Amendola LM et al. ACMG SF v3.3 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 2025. PMID:40568962.

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Rapid Genome Sequencing,

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

Familial Comparator

CHARACTERISTICS: Rapid whole genome sequencing (RWGS) of familial comparator(s) is used to help determine the cause(s) of a disorder in the family proband. RWGS utilizes next generation sequencing (NGS) to interrogate more than 92 percent of the genome (excluding telomeric and centromeric regions), including the mitochondrial genome.

The American College of Medical Genetics and Genomics (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/3019953) for a list of genes analyzed. Note that this gene list is updated periodically and is only accurate for this sample at the time of reporting.

INHERITANCE: Varies by gene and/or variant.

CLINICAL SENSITIVITY: Varies by gene.

METHODOLOGY: Genomic DNA is extracted from whole blood or saliva, prepared into libraries, then sequenced by NGS. Variant calling is performed using the Illumina DRAGEN Bio-IT Platform incorporated with a custom bioinformatics pipeline. Human genome build 19 (Hg 19) is used for data analysis. The analytical procedure identifies single nucleotide variants (SNVs), small insertions/deletions, and copy number variants (CNVs) known, or suspected to be, disease-causing.

LIMITATIONS OF ANALYSIS: Due to technical limitations, some regions of the genome cannot be sequenced or interpreted. SNVs in intergenic or deep intronic regions will only be evaluated if an effect on gene expression is predicted based on annotation software. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations related to pseudogenes or repetitive or homologous regions. This assay is not designed to detect somatic variants, mosaic variants, trinucleotide repeats, uniparental disomy, absence of heterozygosity (AOH), or mitochondrial CNVs. This assay is not designed to detect CNVs greater than 50 bp but less than 1 kb in size. See Additional Technical Information located at http://ltd.aruplab.com/Tests/Pub/3019951 for more information on whole genome sequencing.

REPORTING CONSIDERATIONS: Secondary findings, including disease-associated variants identified in genes on the ACMG-recommended list, or other medically actionable variants at ARUP's discretion, are reported when elected. Interpretation of test results may be impacted if any of the tested individuals have undergone allogeneic stem cell transplantation.

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
RWGS FM Interp	25-307-106640	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

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

END OF CHART

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