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

Physician: ARUP, ARUP

Patient: FAM NGS, NEGATIVE EXAMPLE

DOB	
Sex:	Female
Patient Identifiers:	44376
Visit Number (FIN):	44703
Collection Date:	11/16/2022 13:44

Familial Targeted Sequencing

ARUP test code 3005867

FAM Interp

Negative

RESULT
Negative for the requested familial variant in the BRCA1 gene.

INTERPRETATION
The familial BRCA1 variant, c.1960A>T; p.Lys654Ter, was not detected by massively parallel sequencing of the BRCA1 gene. This variant was previously reported to be associated with hereditary breast and ovarian cancer (HBOC) syndrome in the family; therefore, this individual is predicted not to have the predisposition to hereditary cancers associated with this variant.

RECOMMENDATIONS
Genetic consultation is recommended.

COMMENTS
Reference Sequence: BRCA1 (NM_007294.4)

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Familial Targeted Sequencing

METHODOLOGY: Probe hybridization-based capture of all coding exons and exon-intron junctions of the targeted gene(s) region(s), followed by massively parallel sequencing. Variants in genes, other than the gene(s) region(s) specifically requested, were not evaluated. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected but the analytical sensitivity may be reduced. Specificity is greater than 99.9 percent for all variant classes.

LIMITATIONS: A negative result does not exclude all genetic diagnoses in this individual. This test only evaluates the specified familial variant(s) of interest and other pathogenic or likely pathogenic variants by massively parallel sequencing related to the condition of interest within the targeted gene(s) region(s). Refer to Targeted Sequencing Gene List for complete list of genes available for this test and any gene-specific technical limitations. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing.

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

Regulatory region variants, deep intronic variants, and large deletions/duplications will not be identified. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) mutations, aneuploidies, or repeat expansions. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

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
FAM Interp	22-320-112568	11/16/2022 1:44:00 PM	11/16/2022 1:44:21 PM	11/16/2022 1:46:00 PM

END OF CHART

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