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Tracy I. George, MD, Chief Medical Officer

Patient Report

**Patient Age/Gender:** 0 hours Female

**Specimen Collected: 18-Sep-20 12:06**

**BRCA1 and BRCA2 NGS, DelDup** **|Received: 18-Sep-20 12:07** **Report/Verified: 18-Sep-20 12:11**

| | Result | Units | Reference Interval |
|---|---|---|---|
| BRCA Specimen | Whole Blood | | |
| BRCA Interp | Negative f1 i1 | | |

## Result Footnote

f1: BRCA Interp

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

INDICATION FOR TESTING
Family history of breast cancer.

RESULT
No pathogenic variants were detected in the BRCA1 or BRCA2 genes.

INTERPRETATION
According to information provided to ARUP, this individual has a family history of breast cancer but no personal cancer diagnosis. No pathogenic variants were identified by massively parallel sequencing of the coding regions and exon-intron boundaries of the BRCA1 or BRCA2 genes. No large exonic deletions or duplications were identified in the BRCA1 or BRCA2 genes. This result decreases the likelihood of, but does not exclude a diagnosis of hereditary breast and ovarian cancer (HBOC) syndrome. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical screening and management of this individual should rely on clinical findings and family history. Genetic consultation is recommended. For optimal interpretation of this negative result, determination of a causative familial variant in an affected family member is necessary. Further testing may be warranted if there is a familial variant that is not detectable by this assay.

COMMENTS
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by Rong Mao, M.D.

## Test Information

i1: BRCA Interp

BACKGROUND INFORMATION: BRCA1 and BRCA2-Associated HBOC Syndrome Panel, Sequencing and Deletion/Duplication

CHARACTERISTICS: Individuals with a single germline BRCA1 or BRCA2 pathogenic variant have an increased risk for breast (female and male), ovarian, fallopian tube, peritoneal,pancreatic, and prostate cancers. Additionally, BRCA2 carriers may be at increased risk for melanoma.

EPIDEMIOLOGY: 1 in 40 individuals of Ashkenazi Jewish decent or 1 in 400 individuals in the general population have a germline BRCA1 or BRCA2 pathogenic variant; 5-10 percent of breast cancers and 10-15 percent of ovarian cancers are associated with a hereditary cause.

## Test Information

i1: BRCA Interp

CAUSE: Pathogenic germline variants in the tumor suppressor genes BRCA1 and BRCA2 cause hereditary breast and ovarian cancer (HBOC) syndrome. Approximately 20-60 percent of inherited breast and/or ovarian cancers are due to pathogenic germline variants in BRCA1 and BRCA2.
INHERITANCE: Autosomal dominant
CLINICAL SENSITIVITY: Greater than 90 percent of BRCA1 and BRCA2 pathogenic variants.
GENES TESTED: BRCA1 (NM_007294), BRCA2 (NM_000059)
METHODOLOGY: Multiplex ligation-dependent probe amplification (MLPA) of the BRCA1 and BRCA2 genes. Capture of all coding exons and exon-intron junctions of the BRCA1 and BRCA2 genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and confirm reported variants.
ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity for MLPA is 99 percent. The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.
LIMITATIONS: A negative result does not exclude a heritable form of cancer. This test only detects variants within the coding regions and intron-exon boundaries of the BRCA1 and BRCA2 genes. Regulatory region variants and deep intronic variants will not be identified and breakpoints of large deletions/duplications will not be determined. 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 may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Non-coding transcripts were not analyzed.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS