

SAMPLE REPORT

Client/Sending Facility:
LABCORP OF AMERICA
CMB&P
1912 ALEXANDER DR
RESEARCH TRIANGLE, NC 27709
Ph: (919)361-7700
Fax: (919) 361-7798 NCB-13

LCLS Specimen Number: 123456789

Patient Name: TEST, AJPOS

Date of Birth: 01/01/1975

Gender: N

Patient ID: 123456789

Account Number: 123456789

Ordering Physician: Dr. 101

Specimen Type: BLOOD

Date Collected: 12/10/2013

Date Received: 12/11/2013

Test: BRCA Ashkenazi Jewish Panel

Date Reported: 12/12/2013

Result: Positive for BRCA1 Variant, Pathogenic

Results Summary:

Gene	Variant Detected	Zygosity	Classification
BRCA1	c.68_69delAG (p.Glu23fsX17)	Heterozygous	Pathogenic
BRCA2	Not detected		

Interpretation:

The most significant variant detected was c.68_69delAG, which was present in the heterozygous state. The variant detected in BRCA1 has been classified as associated with an increased risk for the Hereditary Breast and Ovarian Cancer Syndrome.

Recommendation:

Genetic counseling is recommended to discuss the clinical implications of this result. Genetic counselors are available for health care providers to discuss this result further at (800)345-GENE.

Comments:

The three Ashkenazi Jewish founder mutations in BRCA1 (c.68_69delAG and c.5266dupC) and BRCA2 (c.5946delT) are also known by their previous nomenclature, namely 187delAG and 5382insC in the BRCA1 gene and 6174delT in the BRCA2 gene.

Only certain BRCA1 and BRCA2 regions were sequenced that are believed to harbor the variant(s) of interest associated with Hereditary Breast and Ovarian Cancer Syndrome in this individual's family. It cannot be excluded that pathogenic variants were missed due to limitations inherent in the sequence analysis method used here (see Methods/Limitations section). In addition, the presence of the Hereditary Breast and Ovarian Cancer Syndrome due to a different genetic cause can also not be ruled out. Any interpretation given here should be clinically correlated with available information about presentation and relevant family history of the patient.

Methods/Limitations:

Selected regions of the coding sequence of BRCA1 and BRCA2 are amplified by polymerase chain reaction and each PCR product (amplicon) then sequenced bi-directionally, using Sanger sequencing. Nucleotide and codon number are based on the mRNA isoform NM_007294 for BRCA1 gene and NM_000059 for BRCA2 gene.

References:

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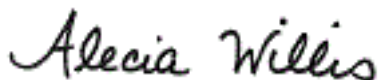
Date Received: 12/11/2013

1. National Comprehensive Cancer Network. Clinical practice guidelines in oncology, genetic/familial high-risk assessment: breast and ovarian. Available at: www.nccn.org. 2010. Accessed 5.29.13.

2. American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility. J Clin Oncol. 2003 Jun 15; 21(12):2397-406.

Disclaimer:

Unless stated otherwise in this report, this test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. LabCorp, is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing.



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Arundhati Chatterjee, MD
Medical Director

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