Integrated GENETICS
LabCorp Specialty Testing Group

SAMPLE REPORT

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

LCLS Specimen Number:	123456789	Account Number:	123456789
Patient Name:	**TEST, AJPOS**	Ordering Physician:	**Dr. 101**
Date of Birth:	01/01/1975	Specimen Type:	**BLOOD**
Gender:	N	Date Collected:	12/10/2013
Patient ID:	123456789	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 heterzygous 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 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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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.

Alecia Willis

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Arundhati Chatterjee, MD
Medical Director

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