myGenomics, LLC
11535 Park Woods Circle • Suite E • Alpharetta • Georgia • 30005
Phone 1-855-647-4363

BRCA Variant Report

Sample ID: MGxxxx

Procedure Performed: Illumina DNA sequencing libraries were prepared and subjected to BRCA1 and BRCA2 targeted gene sequencing. The targeted sequences include the exons and 10bp flanking sequence. The sequencing data was subjected to nucleotide-level variation analysis and the variants filtered for variants of known or suspected clinical significance to include possible disease-causing genetic variations. Please note that each analysis method or filter has its own limitations and confirmation for the functional effects of these genetic variations will need further investigations. The results must be interpreted by a trained professional with sufficient knowledge in related fields of studies.

For Research Use Only. *This test was developed and its performance characteristics determined by myGenomics, LLC in Alpharetta, GA. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. The data contained within this report is not intended for use in diagnosis of disease or other conditions, including a determination of the state of health, in order to cure, mitigate, treat, or prevent disease or its sequelae. False positives may be included in this report due to intentional use of loose filtering algorithm criteria. This report is intended to aid the end user in discovering disease-causing mutations.*

Summary of Results

The filtered list of mutations detected are listed in the Table of Sample Variants on page 2 of this report.

One (1) variant was detected in BRCA1, of unknown significance. The variant detected is predicted to result in an amino acid change to the BRCA1 protein which may or may not affect function.
One (1) mutation was detected in BRCA2, of unknown significance. The variant detected is predicted to result in an amino acid change to the BRCA1 protein which may or may not affect function.
It is important to note that sequencing may not detect all mutations in some regions of the gene.

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Table of Sample Variants

Variant coordinates	Gene	dbSNP Identifier	Sequence change[1]	Protein change[1]	Zygosity[2]	Reference Reads	Variant Reads	Overall Coverage	1000 Genomes Frequency	SIFT Prediction[3]	Polyphen Prediction[3]	dbSNP Clinical Significance
Chr17: 41246480	BRCA1	rs1799950	1067A>G	Gln356Arg	Het	126	123	249	0.028	Not tolerated (0.00)	Probably damaging (0.998)	Other
Chr13: 32950815	BRCA2	--	8869delC	Thr2881Lysfs	Het	139	111	250	--	Not tolerated	Probably Damaging	--

[1] Position of variants numbered based on NM_007294.3 and NP_009225.1 for *BRCA1*, and NM_000059.3 and NP_000050.2 for *BRCA2*.
[2] Homo = homozygous. Het = Heterozygous. For homozygous variants, this ratio should be close to 1. For heterozygous variants, the closer this ratio is to 1/2, the more confident the heterozygous call is.
[3] SIFT and Polyphen predict whether an amino acid substitution affects protein function.

Report Prepared By: JJ
Date: 11 July 2014

Approved By: NJM