Gene Tox Lab Solutions

Patient Information	Specimen Details	Provider Information
Name: Jane Doe	**Specimen Type:**	**Physician:**
Acc#: GM14637_HCP	**Collection Date:**	**Report Date:** Not yet approved
DOB: 05/01/2017	**Received Date:**	
Sex: Female		

Hereditary Cancer - 29 Genes Report

Indication for Testing

Test Result

⊕ Positive Result Pathogenic/Likely pathogenic variant(s) detected.

Test Details

GENE	RELATED DISEASE	VARIANT / POSITION	ZYGOSITY	CLASSIFICATION
BRCA1	Breast-Ovarian Cancer, Familial, Susceptibility To, 1	g.41234451G>A c.4327C>T p.Arg1443Ter	Heterozygous	Pathogenic

Summary

***BRCA1* p.Arg1443Ter**

This patient is heterozygous for variant p.Arg1443Ter in the *BRCA1* gene.

This gene encodes a nuclear phosphoprotein that plays a role in maintaining genomic stability, and it also acts as a tumor suppressor. The encoded protein combines with other tumor suppressors, DNA damage sensors, and signal transducers to form a large multi-subunit protein complex known as the BRCA1-associated genome surveillance complex (BASC). This gene product associates with RNA polymerase II, and through the C-terminal domain, also interacts with histone deacetylase complexes. This protein thus plays a role in transcription, DNA repair of double-stranded breaks, and recombination. Mutations in this gene are responsible for approximately 40% of inherited breast cancers and more than 80% of inherited breast and ovarian cancers. Alternative splicing plays a role in modulating the subcellular localization and physiological function of this gene. Many alternatively spliced transcript variants, some of which are disease-associated mutations, have been described for this gene, but the full-length natures of only some of these variants has been described. A related pseudogene, which is also located on chromosome 17, has been identified. [provided by RefSeq, May 2009]

Disclaimer

Limitations: This test will not detect all the known alleles that result in altered or inactive tested genes. This test does not account for all individual variations in the individual tested. Absence of a detectable gene mutation does not rule out the possibility that a patient has different phenotypes due to the presence of an undetected polymorphism or due to other factors such as drug-drug interactions, comorbidities, and lifestyle habits.

Methodology: Single-nucleotide polymorphism (SNP)-based assays detect listed alleles, including all common and most rare variants with known clinical significance at analytical sensitivity and specificity >99%. Genotyping analysis was performed on Illumina Global Screening Array, which covers about 657,000 SNPs. 3,804 SNP information located in 29 genes, which are related to hereditary cancer, were used for this test.

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About the Report

This report offers insights on pathogenic/likely pathogenic variants found within the set of 29 hereditary cancer genes. These variants were elucidated from extracted genomic material analyzed on an Illumina Global Screening Array-based assay designed to detect clinically relevant single-nucleotide polymorphisms (SNPs). Detected variants were evaluated using genotype quality, minor allele frequency, and curated clinical database for pathogenicity.

Genes tested

APC	APC, WNT signaling pathway regulator
ATM	ATM serine/threonine kinase
BARD1	BRCA1 associated RING domain 1
BLM	Bloom syndrome RecQ like helicase
BMPR1A	bone morphogenetic protein receptor type 1A
BRCA1	BRCA1, DNA repair associated
BRCA2	BRCA2, DNA repair associated
BRIP1	BRCA1 interacting protein C-terminal helicase 1
CDH1	cadherin 1
CDK4	cyclin dependent kinase 4
CDKN2A	cyclin dependent kinase inhibitor 2A
CHEK2	checkpoint kinase 2
EPCAM	epithelial cell adhesion molecule
MLH1	mutL homolog 1
MRE11A	MRE11 homolog, double strand break repair nuclease
MSH2	mutS homolog 2
MSH6	mutS homolog 6
MUTYH	mutY DNA glycosylase
NBN	nibrin
PALB2	partner and localizer of BRCA2
PMS2	PMS1 homolog 2, mismatch repair system component
PTEN	phosphatase and tensin homolog
RAD50	RAD50 double strand break repair protein
RAD51C	RAD51 paralog C
RAD51D	RAD51 paralog D
SMAD4	SMAD family member 4
STK11	serine/threonine kinase 11
TP53	tumor protein p53
VHL	von Hippel-Lindau tumor suppressor