Abnova
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Report Date Aug 29, 2019
Status -

Prostate Cancer Mutation DNA Panel

Specimen Information

Date of Birth
Age
Sex Male
Ethnicity East Asian
Symptoms Not Applicable
Indication Hereditary Disorder

Accession ID
Specimen blood
Collection Aug 15, 2019
Accession Aug 27, 2019

Result: Positive

1	0	0
Pathogenic	Likely Pathogenic	Secondary Findings

Variant Summary

Gene / Variant	Genotype	Assessment	Mode of Inheritance	Phenotype
BRCA2 c.5722_5723delCT p.L1908fs*2	Heterozygous	Pathogenic	dominant	Hereditary breast and/or ovarian cancer

The QIAGEN® GeneReader™ is intended for research use only (RUO). This product is not intended for the diagnosis, prevention or treatment of a disease.

Individual Variant Interpretations

Gene	***BRCA2***
Exon	11
Nucleotide	NM_000059.3: c.5722_5723delCT
Amino Acid	p.L1908fs*2
Genotype	Heterozygous
Assessment	Pathogenic

Interpretation
BRCA2 is a tumor suppressor that is involved in genetic stability through aberrant DNA damage repair [1]. Deletions, loss of heterozygosity, and loss-of-function mutations cause BRCA2 inactivation [3, 2].

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Genes Tested

BRCA1, BRCA2, ATM, PALB2, FANCA

Methods and Limitations

QIAGEN Clinical Insight (QCI™) Interpret software includes the following underlying databases, data reference sets and tools; QIAGEN Clinical Insight-Interpret (5.5.20190807), Ingenuity Knowledge Base (Utopia 190806.000), CADD (v1.4), Allele Frequency Community (2018-12-15), EVS (ESP6500SI-V2), Refseq Gene Model (2018-07-10), JASPAR (2013-11), Ingenuity Knowledge Base Snapshot Timestamp (2019-08-06 05:06:57.0), Vista Enhancer hg18 (2012-07), Vista Enhancer hg19 (2012-07), Clinical Trials (Utopia 190806.000), PolyPhen-2 (v2.2.2), 1000 Genome Frequency (phase3v5b), ExAC (0.3.1), iva (Jun 28 11:10 iva-1.0.1085.jar), PhyloP hg18 (2009-11), PhyloP hg19 (2009-11), DbSNP (151), TargetScan (7.2), GENCODE (Release 28), CentoMD (5.3), OMIM (May 26, 2017), gnomAD (2.0.1), BSIFT (2016-02-23), TCGA (2013-09-05), Clinvar (2019-01-02), DGV (2016-05-15), COSMIC (v87), HGMD (2018.4), SIFT4G (2016-02-23)

1. Wellconn Genomics has no responsibility to the results, if the sample provided by the referring facility is inadvisable to carry out the test analysis.
2. This report is for clinical reference and research use only, and is not to be used in diagnostic and treatment procedures.

Selected Citations

1. Farmer H, McCabe N, Lord CJ, Tutt AN, Johnson DA, Richardson TB, Santarosa M, Dillon KJ, Hickson I, Knights C, Martin NM, Jackson SP, Smith GC, Ashworth A (2005) Targeting the DNA repair defect in BRCA mutant cells as a therapeutic strategy. Nature. 2005 Apr 14;434(7035):917-21 (PMID: 15829967)

2. Walsh T, Casadei S, Coats KH, Swisher E, Stray SM, Higgins J, Roach KC, Mandell J, Lee MK, Ciernikova S, Foretova L, Soucek P, King MC (2006) Spectrum of mutations in BRCA1, BRCA2, CHEK2, and TP53 in families at high risk of breast cancer. JAMA. 2006 Mar 22;295(12):1379-88 (PMID: 16551709)

3. Wooster R, Bignell G, Lancaster J, Swift S, Seal S, Mangion J, Collins N, Gregory S, Gumbs C, Micklem G (1995) Identification of the breast cancer susceptibility gene BRCA2. Nature. 1995 Dec 21-28;378(6559):789-92 (PMID: 8524414)

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