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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 16, 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
ATM c.8435_8436delCT p.S2812fs*2	Heterozygous	Pathogenic	recessive	Ataxia-telangiectasia

Individual Variant Interpretations

Gene	***ATM***
Exon	58
Nucleotide	NM_000051.3: c.8435_8436delCT
Amino Acid	p.S2812fs*2
Genotype	Heterozygous
Assessment	Pathogenic

Interpretation
ATM is a tumor suppressing serine/threonine protein kinase involved in DNA damage and repair [1, 4, 5]. Loss-of-function mutations and gene deletions cause ATM inactivation [3, 2].

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. Berger ND, Stanley FKT, Moore S, Goodarzi AA (2017) ATM-dependent pathways of chromatin remodelling and oxidative DNA damage responses. Philos Trans R Soc Lond B Biol Sci. 2017 Oct 05;372(1731) (PMID: 28847820)

2. Choi M, Kipps T, Kurzrock R (2016) ATM Mutations in Cancer: Therapeutic Implications. Mol Cancer Ther. 2016 Aug;15(8):1781-91. Epub 2016 Jul 13 (PMID: 27413114)

3. Sanmartín E, Muñoz L, Piqueras M, Sirerol JA, Berlanga P, Cañete A, Castel V, Font de Mora J (2017) Deletion of 11q in Neuroblastomas Drives Sensitivity to PARP Inhibition. Clin Cancer Res. 2017 Nov 15;23(22):6875-6887. Epub 2017 Aug 22 (PMID: 28830922)

4. Stracker TH (2017) Chaperoning the DNA damage response. FEBS J. 2017 Aug;284(15):2375-2377 (PMID: 28782203)

5. Xu Y, Gao P, Lv X, Zhang L, Zhang J (2017) The role of the ataxia telangiectasia mutated gene in lung cancer: recent advances in research. Ther Adv Respir Dis. 2017 Sep;11(9):375-380. Epub 2017 Aug 21 (PMID: 28825373)