Germline Cancer Predisposition Panel-focused

Test Description

The MolQ *BRCA* Germline mutation test helps assess your risk of developing cancer by detecting a potentially harmful change (mutation) in *BRCA1* and *BRCA2* genes.

Patient Demographic

Name: Mr. Abdblaziz Mustafa Hassan Mahgoub
Sex: Male
Date of Birth/Age: 28 years
Disease: Asymptomatic

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Dr AV Institute of Personalized Cancer Therapy and Research
Pathologist: Not Provided

Specimen

Booking ID: 012210260098
Site: NA
Sample Type: Blood
Date of Collection: 26-10-2022
Date of Booking: 26-10-2022

CLINICAL SYNOPSIS

The index patient, Ms. Nagwa Ashri Mohamed Hussien's Morphology and immunoprofile favors high grade papillary serous carcinoma of ovary. Tumor cells are immunoreactive for ER, WT-1 and CK7 [as per the histopathological report dated 14-04-2021 provided along with Test Requisition Form]. The tumor was identifiable in the FFPE block [H-1974/21A]. She was found to harbor a heterozygous variant, **c.156del** in *BRCA2* gene. Son of the index patient is being evaluated for the same variation.

RESULTS

Variant is confirmed to be absent by Sanger sequencing.

Gene (Transcript) #	Location	Variant	Relationship to Index Patient	Clinical condition of family member	Variation reported in family member*
BRCA2 (ENST0000544455.1)[1]	Exon 3	chr13:g.32893302del (HET); c.156del; (p.His52GlnfsTer28)	Son	Asymptomatic	Absent

*The exon number and nucleotide numbers will differ based on the reference file chosen and the database used.

CLINICAL CORRELATION AND VARIANT INTERPRETATION

Variant description: A frameshift deletion in exon 3 of the *BRCA2* gene (**chr13:g.32893302del; c.156del**) that results in a premature truncation of the protein 28 amino acids downstream to codon 52 (**p.His52GlnfsTer28**) was detected in the index patient (Sample ID: 7268712, Date of report: 28th October 2021) by Sanger sequencing.

The same variant was not detected in the asymptomatic son of the index patient, Mr. Abdblaziz Mustafa Hassan Mahgoub (Figure 1).

RECOMMENDATION

Genetic counselling is recommended to interpret the significance of the results.

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APPENDIX 1: TEST METHODOLOGY

Method

Targeted gene sequencing by Next Generation Sequencing: Selective capture and sequencing of the protein coding regions of the genome/genes is performed using NGS platform. The sequences obtained are aligned to human reference genome (GRCh37/hg19) using BWA program and analyzed using Picard and GATK-version 3.6 to identify variants detected in the individuals tested in NGS. Variant classification follows the tenets of American College of Medical Genetics (ACMG) guidelines[2].

DISCLAIMER

The results generated after Custom amplicon sequencing for the variation in exon 3 of the *BRCA2* gene (**chr13:g.32893302del; c.156del; p.His52GlnfsTer28**) for SID: 7731979 were not worth reporting, so Custom Amplicon Sequencing was done for the same.

REFERENCES

1. ENSEMBL: http://www.ensembl.org.
2. Green R. C., et al., American College of Medical Genetics and Genomics. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genet Med. 2013 Jul;15(7):565-74

Figure 1: Integrative Genomic Viewer (IGV) snapshot showing the variation in exon 3 of the *BRCA2* gene (chr13:g.32893302del; c.156del; p.His52GlnfsTer28) is not detected in the son of the index patient, Mr. Abdblaziz Mustafa Hassan Mahgoub.