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: Ms Nagwa Ashri Mohamed Hussien
Sex: Female
Date of Birth/Age: 60 years
Disease: High Grade Papillary Serous Carcinoma of Ovary

Clinician

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

Specimen

Booking ID: 012109170159
Site: NA
Sample Type: Blood
Date of Collection: 17-09-2021
Date of Booking: 17-09-2021

CLINICAL SYNOPSIS

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-1974/21 A). The NGS data analysis of this patient has identified a variation in *BRCA2* gene and the same variation is being validated by Sanger sequencing.

RESULTS

Variant is confirmed to be present by Sanger sequencing.

	Analysis For: Variation in *BRCA2* Gene	**Gene Name: *BRCA2* (Exon 3)**
S.No.	**Variation Detected in NGS**	**Sanger Validation Result**
1.	chr13:g.32893302del; c.156del; p.His52GlnfsTer28	Present (Heterozygous)

*The variant analysis in Sanger sequencing is based on the *BRCA2* reference sequence ENST00000544455.1[1]. 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 Ms. Nagwa Ashri Mohamed Hussien by NGS.

The same variation was detected in heterozygous condition in this patient by Sanger sequencing (Figure 1)

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Head, Molecular Biology & Genomics

Dr. Gulshan Yadav, MD
Head, Pathology

APPENDIX 1: TEST METHODOLOGY

Method

Targeted gene Sanger sequencing: Exon 3 of the *BRCA2* gene was PCR-amplified and the products were sequenced using Sanger sequencing. In case of mosaicism in leucocytes, the detection limits of Sanger sequencing for presence of variation is ~12%. The sequence was aligned to available reference sequence ENST00000544455.1[1] to detect variation using variant analysis software programs.

REFERENCES

1. ENSEMBL: http://www.ensembl.org.

Figure 1: Sequence chromatogram and alignment to the reference sequence showing the variation in exon 3 of the *BRCA2* gene (chr13:g.32893302del; c.156del; p.His52GlnfsTer28) detected in heterozygous condition in Ms. Nagwa Ashri Mohamed Hussien.