Germline Cancer Predisposition Panel-focused

Test Description

The MolQ Germline Cancer Predisposition Panel-focused include *BRCA1* and *BRCA2* genes associated with breast and ovarian hereditary cancers.

Patient Demographic

Name: Ms Shikha Goel
Sex: Female
Date of Birth/Age: 35 years
Disease: Asymptomatic

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Max Hospital
Pathologist: Not Provided

Specimen

Booking ID: 011909070007
Site: NA
Sample Type: Blood
Date of Collection: 07-09-2019
Date of Booking: 07-09-2019

CLINICAL SYNOPSIS

The index patient, Ms. Renu Grover, was a case of stage III carcinoma ovary. She was found to harbor a heterozygous variation in the *BRCA1* gene. Her daughter is being evaluated for the pathogenic variations.

RESULTS

Pathogenic variant is detected.

Gene (Transcript) #	Location	Variant	Zygosity	Clinical condition of family member	Classification	Variation reported in family member
BRCA1 (-) (ENST00000471181.2)	Exon 17	chr17:g.41219664del; c.5098del (p.Leu1700Ter)	Heterozygous	Asymptomatic	Pathogenic	Present

CLINICAL CORRELATION AND VARIANT INTERPRETATION

Variant description: A heterozygous single base pair deletion in exon 17 of the *BRCA1* gene (chr17:g.41219664del; c.5098del) that results in a stop codon and premature truncation of the protein at codon 1700 (p.Leu1700Ter) was detected in index patient, Ms. Renu Grover (Sample ID: 293187) by NGS.

The same pathogenic variation was detected in heterozygous condition in the asymptomatic daughter of the index patient, Ms. Shikha Goel (Figure 1). Incomplete penetrance and variable age of cancer development have been reported in *BRCA1* gene variants[1]. Hence the results must be carefully correlated with clinical condition of the individuals tested.

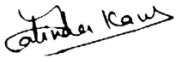
RECOMMENDATIONS

- Genetic counselling is advised to discuss the significance of this test. Kindly email us at contact@molq.in for post-test counselling.

REFERENCES

1. Petrucelli N, Daly MB, Pal T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer. 1998 Sep 4 [Updated 2016 Dec 15]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019.

Germline Cancer Predisposition Panel-focused



Jatinder Kaur, PhD
Head, Molecular Biology & Genomics

Dr. Gulshan Yadav, MD
Head, Pathology

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.

Figure 1: Integrative Genomic Viewer (IGV) snapshot showing the variation (chr17:g.41219664del; c.5098del; p.Leu1700Ter) in exon 17 of *BRCA1* gene not detected in the daughter of the index patient, Ms. Shikha Goel.

303022 – Daughter of index patient (Ms. Shikha Goel)