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MEDGENOME

DNA TEST REPORT– MEDGENOME LABORATORIES

Order ID/Sample ID:	**103744/250536**	Sample Type:	Blood (2.5ml) in EDTA tube
Full Name/Ref no:	**Mr. Mast Ramji**	Date & time of Sample Collection:	NA
Referring Clinician:	Dr. Amit Verma, Molecular Quest Healthcare Pvt.Ltd, Haryana.	Date & time of Sample Receipt: Order date & time: Date & Time of report:	06-03-2019; 04:25 PM 18-03-2019; 04:16 PM 09-04-2019; 06.30 PM
Test Requested:	**Additional family member (investigational) testing (MGM277)**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

The index patient, Ms. Krishnadevi Bhardwaj was found to harbor a heterozygous variation in *BRCA2* gene and father of the index patient is being evaluated for the same variation.

RESULT SUMMARY

Analysis for: **Variation detected by Next Generation Sequencing in the *BRCA2* gene of Ms. Krishnadevi Bhardwaj (90971)**

Sl. no.	Sample ID	Name, Gender, Age	Relationship to the index patient	Gene Name	Exon / Intron	Variation reported in index patient	Variation detected in family member*	Clinical condition of family member
1.	**250536**	**Mr. Mast Ramji, Male, 85yrs**	**Father**	***BRCA2***	**Exon 3**	**chr13:32893238G>G/A (HET); c.92G>G/A; p.Trp31Ter**	**Absent**	**Asymptomatic**

* The variant analysis in Sanger sequencing is based on the *BRCA2* reference sequence ENST00000544455 [1]. The exon number and nucleotide numbers will differ based on the reference file chosen and the database used.

INTERPRETATION

A heterozygous nonsense variation in exon 3 of the *BRCA2* gene **(chr13:32893238G>G/A; c.92G>G/A)** that results in a stop codon and premature truncation of the protein at codon 31 **(p.Trp31Ter)** was detected in Ms. Krishnadevi Bhardwaj (Sample ID: 90971) by NGS and was further validated by Sanger sequencing (Fig. 1 A).

The same pathogenic variation was not detected in the asymptomatic father of the index patient, Mr. Mast Ramji (Fig. 1 B).

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TEST METHODOLOGY

Exon 3 of *BRCA2* gene was PCR-amplified and the product was sequenced using Sanger sequencing. The detection limits of Sanger sequencing for presence of variation is ~12%. The sequence was aligned to available reference sequence ENST00000544455 [1] to detect variation using variant analysis software programs.

Report prepared by:	**Results verified by:**	**Report released by:**
Gargi G. Nanda, PhD	**Dr. Sheetal Sharda, MD (Paediatrics), DM (Medical Genetics), MNAMS.**	**V L Ramprasad, PhD**
Senior Genome Analyst	Consultant - Clinical Geneticist	Chief Operating Officer

REFERENCE

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

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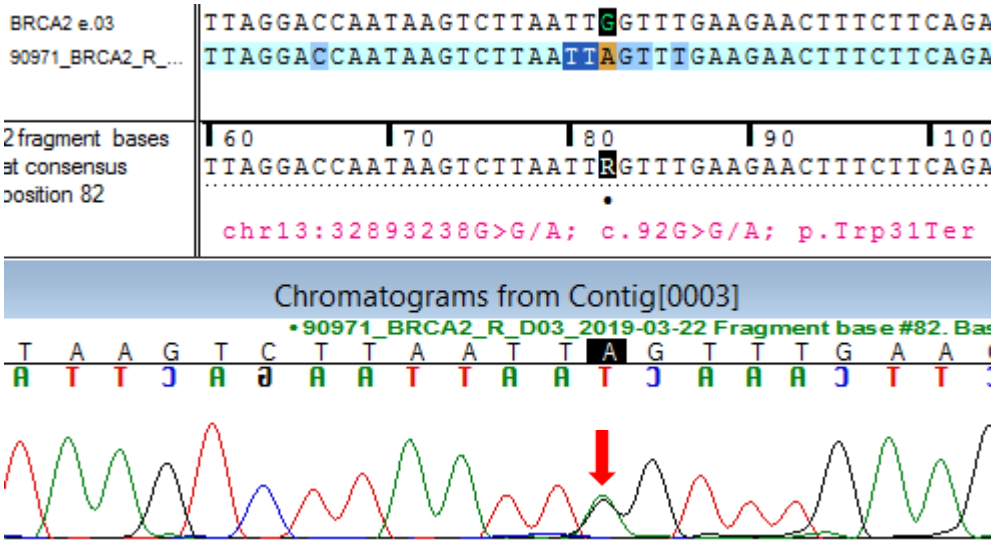
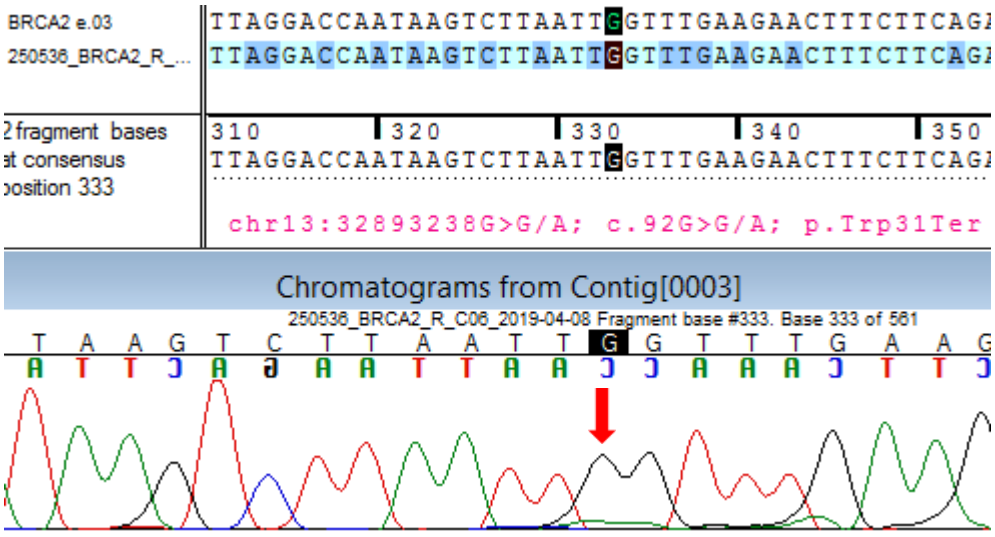
ANNEXURE-I

Fig.1: Sequence chromatogram and alignment to the reference sequence showing the variation in exon 3 of *BRCA2* gene (chr13:32893238G>G/A; c.92G>G/A; p.Trp31Ter) detected in heterozygous condition in the index patient, Ms. Krishnadevi Bhardwaj (A) and not detected in the father of the index patient, Mr. Mast Ramji (B).

A. 90971 – Index patient (Ms. Krishnadevi Bhardwaj)



B. 250536 – Father of the index patient (Mr. Mast Ramji)



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