Peter Smith | Med-V4I2H-S

Age: 22 years | Gender: MALE

Diagnosis: **Hereditary breast and ovarian cancer syndrome (BRCA1-related)**

Impactomics

MGM537 : Tumor BRCA1 & BRCA2 Gene Analysis

Report Details

Sample ID :	Med-V4I2H-S
Collection Date:	11th Jan 2026
Date Received:	8th Jan 2026
Report Date & Time:	2nd Feb 2026 6:10 PM

Specimen Information

Specimen Site:	
Specimen Received:	106
Specimen Tested:	FFPE-006
Tumor Content (%):	36

Ordering Clinician

Clinician:	Vivek G
Organization:	Rainbow hospital
Serviced By:	N/A
Report Status:	DRAFT

Clinical Summary

Patient referred for tumor BRCA1 & BRCA2 gene analysis. FFPE block received; NGS performed. Clinically relevant variant(s) identified; see AMP classification and result table.

Test Result Summary

Next Generation Sequencing (NGS) Results Positive

Gene	Findings	Gene	Findings
BRCA2	Not Detected	*BRCA1*	Single Nucleotide Variant

Please refer to the complete variant details in the result table in page 2.

Next Generation Sequencing (NGS) Results Positive

Clinically relevant variants were detected.

AMP Classification^	CDS variant details	Interpretation	Treatment Recommendations	Treatment Response$
BRCA1 imr5050 (snv) Variant Allele Frequency - 0.8%				
Tier 1	{newVal=c.-20+221G>A, oldVal=c.-20 +221G>A}	N/A	N/A	Not Effective

* **Clinically Significant** term in this report refers to the mutation that has potential to alter the medical intervention.

^ Refer to AMP-ASCO-CAP classification criteria section for the AMP classification criteria details.

Note: Decisions regarding treatment action plan should not be solely based on this test results. These findings are highly recommended to be correlated with the patient's clinical, pathological, and family history for decisions on diagnosis, prognosis or therapeutics.

Additional Biomarkers Detected (Variants of Uncertain Significance)

This section provides information about variants that do not have any therapeutic value. However, these variants may or may not have a likely oncogenic effect.

No other biomarkers that warrants to be reported was detected.

Disclaimer

- **Decisions regarding treatment action plan should not be solely based on these test results. These findings are highly recommended to be correlated with the patient's clinical, pathological, radiological and family history for decisions on diagnosis, prognosis, or therapeutics.**
- The therapy information provided in this report is based on FDA approved drugs data, NCCN guidelines, peer-reviewed published literature, standard clinical databases, and strength of biomarker results till date. These therapies may or may not be suitable /beneficial to a particular patient. This clinical report summarizes potentially effective medications, potentially ineffective medications, and medications that may pose a higher risk of adverse reactions by mapping the patient's genetic alterations to the biomedical reference information. The report may also provide prognostic and diagnostic biomarkers detected or shown for the given disease context.
- The clinical trials information provided in this report is compiled from www.clinicaltrials.gov as per currently available data, however, completeness of information provided herein cannot be guaranteed. This information should only be used as a guide and specific eligibility criteria should be reviewed thoroughly for the concerned patient. Medgenomes does not guarantee or promise an enrolment in any clinical trials.
- The identification of a genomic biomarker does not necessarily imply pharmacological effectiveness or ineffectiveness. The medications identified by the treating physician may or may not be suitable for use on a particular patient. Thus, the clinical report does not guarantee that any particular agent will be effective in the treatment of any particular condition. Also, the absence of a treatment option does not determine the effectiveness or predict an ineffective or safety-relevant effect of a medication selected by the treating physician.
- The classification and clinically relevant information for the reported variants is based on peer-reviewed publications, public clinical databases, medical guidelines (WHO, NCCN, ASCO, AMP) or other publicly available information and it has been ensured that the information provided is up to date at the time of report generated, however continuous updates may happen in public domains. Also, the classification of variants can change based on the updated literature evidence. Re-analysis of the results can be requested at additional cost.
- This test is performed on the patient's tumour sample without a paired blood sample; therefore, it may include variations which may be of germline origin. However, this test is designed and validated for the detection and reporting of somatic genomic variants only and does not discriminate between germline and somatic variants. If clinically warranted, appropriate germline testing and genetic counselling for the patient should be considered for further evaluation.
- Due to poor quality of FFPE tissue blocks, the QC parameters from extracted DNA may not pass to proceed further with the testing, therefore there is a possibility of assay failure at various steps (DNA QC, Library QC, Bioinformatics QC) or compromised results that include low gene coverage and low variant depth. However, sample status in such scenarios shall be sent through mail to the ordering clinician.
- The test has been validated at Medgenomes and the limit of detection (LOD) of allele fraction for SNVs and InDels is 5%. However, the report may include, at the discretion of laboratory director, the variants with lower allele burden (3-5%) having strong or potential clinical significance or those have been reported earlier in the patient. Variants with <1% allele fraction and variant s of uncertain significance with <5% allele fraction are not routinely reported. however, possibility of false negative or false positive variants are not detected in this assay.
- Large deletions and copy number variations and deep intronic variations are beyond the scope of this test.
- Additional case specific disclaimer: None

Test Description

The Medgenomes's *BRCA1* & *BRCA2* panel is a high throughput next-generation sequencing based single assay that covers complete coding regions and splice boundaries of these genes to detect single nucleotide variations (SNVs), small insertions and deletions (InDels) and splice variants. The test is performed on the tumor biopsy FFPE block/curls which enables detection of somatic and germline variations that may provide treatment benefit to the patients. The positive cases are recommended to be screened for germline predisposition through blood genetic testing followed by genetic counselling.

Test Methodology

Sample type: FFPE Specimen; A histopathologic review is performed to determine the tumor content in the FFPE block/curls.

Extraction and Library Preparation: Tumor nucleic acid is extracted from FFPE (Formalin fixed) tissue block and used to perform targeted gene capture using a custom hybrid capture kit for HRR genes (complete coding region).

Sequencing: The QC-passed libraries are sequenced to a minimum depth of 250X (post UMI collapse) on a validated Illumina sequencing platform.

Data Analysis: The sequences are processed using a customized and validated UMI-based analysis pipeline designed to accurately detect all classes of genomic alterations (SNVs and InDels).

Variant Annotation and Reporting: The variants are annotated using our in-house annotation pipeline. Reportable genomic alterations are prioritized, classified, and reported based on AMP-ASCO-CAP guidelines [PMID: 27993330] and NCCN guidelines.

Limit of Detection (LOD): The LOD for SNVs and InDels is 5% Variant allele Frequency (VAF).

This test was developed, and its performance characteristics determined by Medgenomes

#The transcript used for clinical reporting generally represents the canonical transcript (according to Ensembl release 99 human gene model), which is usually the longest coding transcript with strong/multiple supporting evidence. However, clinically relevant variants annotated in alternate complete coding transcripts could also be reported. Variants annotated on incomplete and nonsense mediated decay transcripts will not be reported.

AMP-ASCO-CAP Classification Criteria

Genetic test results are reported based on the somatic variant classification recommendations of College of American Pathologists (CAP) / American society for Clinical Oncology (ASCO) / Association of Molecular Pathologists (AMP) [PMID: 27993330] as described in the table below:

Tier	Criteria
Tier I	Variants of strong clinical significance.
Tier II	Variants of potential clinical significance.
Tier III	Variants of unknown clinical significance.
Tier IV	Benign or likely benign variants.

Reference: PMID: 27993330

Signatures

Dr. Syed Muqlisur Rehman
MD Path
KMC Reg No. 71468

Aparna Natarajan
Ph.D

Genes Analyzed

Total Genes: 2

BRCA1 *BRCA2*