

BRCA1/2 Mutation Analysis, Next-Generation
Sequencing, Tumor

Patient ID SA00174965	Patient Name TESTINGRNV, ABNORMAL	Birth Date 2005-05-05	Sex F	Age 19
Order Number SA00174965	Client Order Number SA00174965	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 28 Mar 2025 00:00		

BRCA1/2 Mutation Analysis, Tumor

Result

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Provided diagnosis: adenocarcinoma involving liver

The following CLINICALLY RELEVANT VARIANT was detected:

Gene: BRCA1

DNA Change: c.2594del (Exon 10)

Amino Acid Change: p.K865Sfs*28 (Lys865Serfs*28)

Variant Allele Frequency: 90.3%

No other reportable sequence variants were detected within the analyzed regions of the tested genes listed in the method description.

Interpretation

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BRCA1 c.2594del (p.K865Sfs*28) (Exon 10)

BRCA1 encodes the protein Brca1, which is involved in the maintenance of genomic stability, including DNA repair, cell cycle checkpoint, and chromosome segregation [PMID:20400477]. Inactivating mutations of BRCA1 can lead to the inability to repair DNA damage and loss of cell cycle checkpoints, which can lead to tumorigenesis [PMID:24638981, PMID:28079255].

Inactivating BRCA1 and BRCA2 alterations have been reported to predict sensitivity to platinum-based chemotherapy and PARP inhibitors, some of which are FDA-approved for specific indications [PMID:30110579, PMID:28546758, PMID:26187614, PMID:27908594, PMID:27717299].

Germline (inherited) mutations in BRCA1 are associated with a hereditary cancer syndrome. This test does not distinguish between germline and somatic (not inherited) genetic variants. If a hereditary cancer syndrome is suspected for this patient, clinical correlation with their personal and family history as well as a referral for genetic counseling is recommended.

Additional Information

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CLINICAL TRIALS

Possible clinical trials of benefit for this patient can be found at the following sites:

1) ClinicalTrials.gov:

www.clinicaltrials.gov/ct2/search/advanced

2) Mayo Clinic: www.mayo.edu/research/clinical-trials/ 3) National Cancer Institute: www.cancer.gov/clinicaltrials/search

REFERENCE TRANSCRIPT

Sequence variant nomenclature is based on the following RefSeq accession number (build GRCh37 (hg19)):BRCA1 NM_007294.

Specimen

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Tissue, Tumor

Tissue ID

MCR

12345

Method

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Microscopic examination is performed by a pathologist to identify areas of tumor for enrichment by macrodissection. DNA is extracted from FFPE or cytology slides, and next generation sequencing is performed to evaluate the presence of a mutation in all coding regions and exon/intron boundaries of the BRCA1 and BRCA2 genes.

Variant nomenclature is based on build GRCh37 (hg19). For details about gene transcripts (RefSeq accession numbers), specific targeted regions of each gene, and additional information on this test, see www.mayocliniclabs.com (Test ID BRCAT).

Disclaimer

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This test cannot differentiate between somatic and germline alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292

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DNA variants of uncertain significance may be identified.

A negative result does not rule out the presence of a variant that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay for sequence reportable alterations is 5% mutant allele frequency with a minimum coverage of 500X in a sample with at least 20% tumor content.

Point mutations and small insertion/deletion mutations will be detected in the BRCA1 and BRCA2 genes only. This test may detect single exon deletions but does not detect multi-exon deletions, duplications, larger-scale genomic copy number variants, copy neutral loss of heterozygosity (LOH), or epigenetic modifications such as promoter methylation. DELINS of 1,000 bp or less are detectable with at least 50 or more supporting reads.

Variant allele frequency (VAF) is the percentage of sequencing reads supporting a specific variant divided by the total sequencing reads at that position. In somatic testing, VAF should be interpreted in the context of several factors including, but not limited to: tumor purity/heterogeneity/copy number status (ploidy, gains/losses, loss of heterozygosity) and sequencing artifact/misalignment [PMID: 27144058, PMID: 29769535].

Rare polymorphisms may be present that could lead to false-negative or false-positive results.

The presence or absence of a variant may not be predictive of response to therapy in all patients.

Test results should be interpreted in the context of clinical, tumor sampling, histopathological, and other laboratory data. If results obtained do not match other clinical or laboratory findings, contact the laboratory for discussion. Misinterpretation of results may occur if the information provided is inaccurate and/or incomplete.

Reliable results are dependent on adequate specimen collection and processing. This test has been validated on cytology slides and formalin-fixed, paraffin-embedded tissues; other types of fixatives are discouraged. Improper treatment of tissues, such as decalcification, may cause PCR failure.

Released By

Sounak Gupta, M.B.B.S., Ph.D.

MCR

Received: 29 Mar 2025 09:50

Reported: 06 Apr 2025 11:28

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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