Patient ID	Patient Name		Birth Date	Sex	Age
SA00174983	TESTINGRNV, ABNORMAL		1988-02-04	F	37
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174983	SA00174983	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

MayoComplete Bladder/Prostate Panel

Result

MCR

Provided diagnosis: prostate tumor

The following CLINICALLY RELEVANT VARIANT was detected:
Gene: BRCA2
DNA Change: c.5073dup (Exon 11)
Amino Acid Change: p.W1692Mfs*3 (Trp1692Metfs*3)
Variant Allele Frequency: 40.5%

Microsatellite Instability (MSI) status: Stable (MSS)

No other reportable sequence variants were detected within the analyzed regions of the tested gene(s) listed in the method description.

Interpretation

MCR

1) BRCA2 c.5073dup (p.W1692Mfs*3) (Exon 11)

BRCA2 encodes the Brca2 tumor suppressor. Brca2 aids in DNA double-strand break repair in response to chromosomal damage, in part by binding and regulation of Rad51 [PMID:12228710]. Inactivating mutations of BRCA2 can lead to the inability to repair DNA damage and loss of cell cycle checkpoints, which can lead to tumorigenesis [PMID:21731065, PMID:28079255].

BRCA2 alterations have been reported in 4–5% of prostate adenocarcinoma samples and 2–13% of urothelial carcinoma samples [COSMIC, Prostate Adenocarcinoma (TCGA, PanCancer Atlas), cBioPortal for Cancer Genomics].

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].

Additional studies are needed to assess the efficacy of approved targeted therapy in other tumor types.

Germline (inherited) mutations in BRCA2 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.

MSI: Stable (MSS)
NO evidence of microsatellite instability was detected suggesting the presence of normal DNA mismatch repair function within the tumor. Current data suggest that advanced stage solid tumors with intact mismatch repair (MSS) are less likely to be responsive to treatment with immunotherapies such as anti-PD-1 therapies [PMID 28596308, PMID 29355075].

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)): BRCA2 NM_000059.

Specimen

MCR

Tissue, Tumor

Tissue ID

MCR

12345

Method

MCR

Microscopic examination is performed by a pathologist to identify areas of tumor for enrichment by macrodissection.

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

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174983	TESTINGRNV, ABNORMAL		1988-02-04	F	37
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
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C7028846 DLMP Rochester		28 Mar 2025 00:00			

Next generation sequencing is performed to test for the presence of microsatellite instability and a mutation within most coding regions and exon/intron boundaries of the following genes: APC, AR, ARID1A, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CDK12, CDKN2A, CHD1, CHEK1, CHEK2, CTNNB1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCL, FGFR1, FGFR2, FGFR3, FOXA1, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51B, RAD51C, RAD51D, RAD54L, RB1, SPOP, TERT, and TP53

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 MCBPP).

Disclaimer

[1] MCR

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.

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 APC, AR, ARID1A, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CDK12, CDKN2A, CHD1, CHEK1, CHEK2, CTNNB1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCL, FGFR1, FGFR2, FGFR3, FOXA1, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51B, RAD51C, RAD51D, RAD54L, RB1, SPOP, TERT, and TP53 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.

This test cannot reliably determine if a variant identified in PMS2 exons 11–15 originated from PMS2 or the highly homologous pseudogene PMS2CL. In the instance that a reportable variant is detected in PMS2 exons 11–15, additional testing will be recommended in the patient report.

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

MCR

HUSSAM AL KATEB

Received: 29 Mar 2025 11:29 **Reported:** 08 Apr 2025 14:07

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

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Sex	Age
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SA00174983	SA00174983	CLIENT,CLIENT			
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C7028846 DLMP Rochester		28 Mar 2025 00:00			

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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