Patient ID	Patient Name		Birth Date	Sex	Age
321	IMPLEMENTATION, TEST T		1973-05-01	F	52
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100515187	X100515187	UNKNOWN,PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		01 Dec 2025 10:00			

MayoComplete Sarcoma Panel

Result

MCR

Provided diagnosis: spindle cell neoplasm involving uterine contents

No Fusion detected

The following CLINICALLY RELEVANT VARIANT was detected:
Gene: MED12
DNA Change: c.122_139del (Exon 2)
Amino Acid Change: p.V41_N47delinsD (Val41_Asn47delinsAsp)
Variant Allele Frequency: 33.3%

Microsatellite Instability (MSI) status: Stable (MSS)

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

Interpretation

MCR

1) MED12 c.122_139del (p.V41_N47delinsD) (Exon 2)

MED12 encodes Med12, which is part of a Cdk8 kinase-containing Mediator complex module that contributes to both the activation and repression of transcription and has been shown to be the subunit responsible for direct physical interactions with proteins such as Sox9, beta-catenin, Gli3, and REST [PMID:12136106, PMID:16565090, PMID:17000779, PMID:17716226, PMID:24342527]. Mutation and loss of MED12 have been reported in several types of cancer; it has been suggested that inactivation of Med12 may promote cancer through the dysregulation of several signaling pathways, including TGF-beta, MEK, ERK and hormone receptors [PMID:22532225, PMID:23836153, PMID:24342527].

MED12 mutations have been reported in 1–30% of sarcoma samples, mostly commonly in smooth muscle tumors [COSMIC, cBioPortal for Cancer Genomics].

There are currently no known clinically approved therapies that specifically target MED12 mutations.

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

MCR

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)):MED12 NM_005120.

Specimen

MCR

Tissue, Tumor

Tissue ID

MCR

13245

Method

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

Next generation sequencing is performed to test for the presence of microsatellite instability and a mutation within all coding regions and exon/intron boundaries of the following 31

Performing Site Legend

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MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292

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targeted genes (unless otherwise specified): ALK, APC, BAP1, BCOR (exon 15 only), BRAF, CDKN2A, CTNNB1, DICER1, EED, EGFR, FGFR4, GNA11, GNA14, GNAQ, GNAS, H3–3A (excludes exon 4), H3–3B, KIT, MDM2, MED12 (excludes exon 43), MYOD1, NF1, PDGFRA, PDGFRB, PTPRD, ROS1, SMARCB1, SUZ12 (excludes exons 3, 4, 6, and 9), TERT (promoter), TP53, and TSC2.

Next-generation sequencing is performed to test for the presence of gene rearrangements involving 1,445 genes, transcript variants in the MET and EGFR genes and in-tandem duplications within exon 15 of the BCOR gene.

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 MCSRC).

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.

Variants and fusions of uncertain significance may be identified.

A negative result does not rule out the presence of a variant that may be present below the sensitivity 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 20% or more tumor content.

The sensitivity of this assay for gene fusions depends on several variables including decreased sensitivity with decreased tumor percentage, and decreased sensitivity with decreased level of expression of a variant. A negative result does not rule out the presence of a gene fusion, splice variant, or BCOR exon 15 internal tandem duplication that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay for rearrangements is a minimum coverage of 5 unique variant molecules in a sample with at least 10% tumor content.

Point mutations and small deletion-insertion mutations will be detected in the ALK, APC, BAP1, BCOR, BRAF, CDKN2A, CTNNB1, DICER1, EED, EGFR, FGFR4, GNA11, GNA14, GNAQ, GNAS, H3–3A, H3–3B, KIT, MDM2, MED12, MYOD1, NF1, PDGFRA, PDGFRB, PTPRD, ROS1, SMARCB1, SUZ12, TERT-promoter, TP53, and TSC2 genes. 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 assay can detect in-frame and out-of-frame fusions involving 1,445 genes. Sensitivity for detecting out-of-frame fusions such as exon-intron, intron-intron or big insertions, may be lower due to bioinformatics detection limitations. This assay will only detect fusions involving at least 1 gene in the defined gene fusion target list of interest. This assay may not detect fusions involving deep intron or intergenic regions and will not detect chromosomal rearrangements that do not create a fusion transcript (i.e. enhancer repositioning). Variants not expressed, or expressed at very low level, are not detected by this assay.

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 variants, or alterations derived from the production of a gene fusion, may be present that could lead to false-negative or false-positive results.

The presence or absence of a variant or fusion 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 or incomplete.

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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 polymerase chain reaction failure.

RNA is particularly labile and degrades quickly. Rapid preservation of the tumor sample after collection reduces the likelihood of degradation, but sometimes, there are biological factors, such as tumor necrosis, that interfere with obtaining a high-quality RNA specimen despite rapid preservation.

Released By MCR

HUSSAM AL KATEB

Received: 02 Dec 2025 12:37 **Reported:** 04 Dec 2025 14:33

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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