

Patient ID 321	Patient Name IMPLEMENTATION, TEST T	Birth Date 1973-05-01	Sex F	Age 52
Order Number X100515181	Client Order Number X100515181	Ordering Physician UNKNOWN, PROVIDER	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 01 Dec 2025 09:00		

MayoComplete Targeted RNAseq Panel

Result

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Provided diagnosis: sarcoma

The following CLINICALLY RELEVANT VARIANT was detected:

Fusion: NAB2::STAT6 (exon 4::exon 2)

Unique supporting reads: 1002

NO additional reportable CLINICALLY RELEVANT FUSIONS/TRANSCRIPT VARIANTS were detected involving the tested genes.

Interpretation

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1) NAB2::STAT6 (exon 4::exon 2)

NAB2::STAT6 fusions are oncogenic and typically result from an intrachromosomal (12q13) inversion that truncates the NAB2 transcriptional repressor domain and juxtaposes its early growth response 1 (EGR1)-binding domain to the activation domain of STAT6. These fusions lead to attenuation of the repressive activity of NAB2 with constitutive activation of EGR1-target genes, converting NAB2 from "transcriptional repressor" into a "transcriptional activator" of EGR1 [PMID:23313952, PMID:23313954, PMID:26927892].

NAB2::STAT6 fusions are a diagnostic molecular biomarker for solitary fibrous tumor/hemangiopericytoma (SFT/HPC) [WHO Classification of Tumours Editorial Board. Soft tissue and bone tumours. Lyon (France): International Agency for Research on Cancer; 2020. (WHO classification of tumours series, 5th ed.; vol. 3).].

However, the occurrence of this fusion in other tumor types cannot be excluded.

There are currently no known clinically approved therapies that specifically target NAB2 or STAT6 fusions.

If additional guidance is desired as to the significance of this fusion event, surgical pathology consultation is available from

Mayo Clinic Laboratories at the following address: Attention: Pathology, Mayo Clinic Laboratories, 3050 Superior Dr. NW, Rochester, MN 55901-1195. Please include relevant radiographic studies for cases involving bone.

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(S) AND FUSION/TRANSCRIPT VARIANT JUNCTIONS

Fusion/transcript variant (i.e. rearrangement) junction location and sequence variant nomenclature is based on the following RefSeq accession number(s) (build GRCh37 (hg19)): NAB2 NM_005967 and STAT6 NM_003153.

The rearrangement junction location and the corresponding genomic coordinates within the NAB2 gene occur in exon 4 at genomic position chr12(GRCh37):g.57486751 and within the STAT6 gene occur in exon 2 at genomic position chr12(GRCh37):g.57502082.

Specimen

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Tissue, Tumor

Tissue ID

MCR

1345

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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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 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 MCRSP).

Disclaimer

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This assay is not validated for the detection of single nucleotide variations, deletions-insertions, copy number alterations, or gene expression.

Fusions of uncertain significance may be identified.

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 is a minimum coverage of 5 unique variant molecules in a sample with at least 10% tumor content.

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.

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 fusion may not be predictive of response to therapy in all patients.

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

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

Released By

MCR

HUSSAM AL KATEB

Received: 02 Dec 2025 12:24

Reported: 04 Dec 2025 14:22

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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