

## MayoComplete Sarcoma Mutation Panel, Next-Generation Sequencing, Tumor

|                                                       |                                           |                                            |                 |                  |
|-------------------------------------------------------|-------------------------------------------|--------------------------------------------|-----------------|------------------|
| Patient ID<br><b>SA00175006</b>                       | Patient Name<br><b>TESTINGRNV, NORMAL</b> | Birth Date<br><b>2000-03-19</b>            | Sex<br><b>M</b> | Age<br><b>25</b> |
| Order Number<br><b>SA00175006</b>                     | Client Order Number<br><b>SA00175006</b>  | Ordering Physician<br><b>CLIENT,CLIENT</b> | Report Notes    |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |                                           | Collected<br><b>28 Mar 2025 00:00</b>      |                 |                  |

## MayoComplete Sarcoma Mutation Panel

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| <p><b>Result</b> <span>MCR</span></p> <p><b>Provided diagnosis:</b> bone osteogenic tumor</p> <p><b>Microsatellite Instability (MSI) status:</b> Stable (MSS)</p> <p>No reportable sequence variants were detected within the analyzed regions of the tested genes listed in the method description.</p> <p>This assay only evaluates mutations and small insertion/deletions. FUSION testing was NOT PERFORMED. If fusion testing is desired, consider Sarcoma Targeted Gene Fusion/Rearrangement Panel, Next-Generation Sequencing, <b>Tumor (Mayo Test ID: SARCP)</b> or FISH assays. Refer to <a href="http://mayocliniclabs.com">mayocliniclabs.com</a> or call 1-800-533-1710 for more information about specimen requirements for this test.</p> <p><b>Interpretation</b> <span>MCR</span></p> <p>No reportable gene mutations informative for diagnosis, prognosis or predictive of response to therapy were identified within the analyzed genes. If a specific fusion is suspected, consider ordering SARCP (Sarcoma Targeted Gene Fusion Panel). Test results should be interpreted in the context of clinical findings, tumor sampling, histopathology, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory.</p> <p>MSI: Stable (MSS)<br/>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].</p> | <p><b>Additional Information</b> <span>MCR</span></p> <p><b>CLINICAL TRIALS</b></p> <p>Possible clinical trials of benefit for this patient can be found at the following sites:</p> <p>1) ClinicalTrials.gov: <a href="http://www.clinicaltrials.gov/ct2/search/advanced">www.clinicaltrials.gov/ct2/search/advanced</a></p> <p>2) Mayo Clinic: <a href="http://www.mayo.edu/research/clinical-trials/">www.mayo.edu/research/clinical-trials/</a> 3) National Cancer Institute: <a href="http://www.cancer.gov/clinicaltrials/search">www.cancer.gov/clinicaltrials/search</a></p> <p><b>Specimen</b> <span>MCR</span></p> <p>Tissue, Tumor</p> <p><b>Tissue ID</b> <span>MCR</span></p> <p>12345</p> <p><b>Method</b> <span>MCR</span></p> <p>Microscopic examination is performed by a pathologist to identify areas of tumor for enrichment by macrodissection.</p> <p>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 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.</p> <p>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 <a href="http://www.mayocliniclabs.com">www.mayocliniclabs.com</a> (Test ID MCSMP).</p> <p><b>Disclaimer</b> <span>1 MCR</span></p> <p>This test cannot differentiate between somatic and germline</p> |
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### 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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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 ALK, APC, BAP1, BCOR (exon 15), 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 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

MCR

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

**Received:** 29 Mar 2025 16:23

**Reported:** 02 May 2025 08:03

#### 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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