

Patient ID SA00174990	Patient Name TESTINGRNV, NORMAL	Birth Date 1984-02-16	Sex M	Age 41
Order Number SA00174990	Client Order Number SA00174990	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 28 Mar 2025 00:00		

MayoComplete GIST Panel

Result

MCR

Provided diagnosis: gastrointestinal stromal tumor

Microsatellite Instability (MSI) status: Stable (MSS)

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

Interpretation

MCR

Current data suggests that tyrosine kinase inhibitor therapies (i.e. imatinib) are most effective in patients with GISTs that have activating KIT or PDGFRA mutations. Thus, the absence of a detectable mutation suggests that such therapies may have limited therapeutic value for this patient [PMID 14645423, PMID 22089421, PMID 24925196, PMID 24790651, PMID 24384849].

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

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. 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 11 targeted genes: APC, BRAF, HRAS, KIT, KRAS, NF1, NRAS, PDGFRA, PIK3CA, SETD2, 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 MCGST).

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, BRAF, HRAS, KIT, KRAS, NF1, NRAS, PDGFRA, PIK3CA, SETD2, 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.

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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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

ANDE RUMILLA

Received: 29 Mar 2025 12:25

Reported: 01 May 2025 02:41

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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