

Patient ID SA00174991	Patient Name TESTINGRNA, ABNORMAL	Birth Date 1988-02-23	Sex F	Age 37
Order Number SA00174991	Client Order Number SA00174991	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

The following CLINICALLY RELEVANT VARIANT was detected:

Gene: KIT

DNA Change: c.1651_1662del (Exon 11)

Amino Acid Change: p.P551_E554del (Pro551_Glu554del)

Variant Allele Frequency: 33%

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

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1) KIT c.1651_1662del (p.P551_E554del) (Exon 11)

KIT encodes a tyrosine kinase receptor also known as c-Kit or CD117 that is expressed on the cell surface of a variety of cell types, including hematopoietic stem cells and melanocytes. Binding of the Kit ligand stem cell factor (SCF, or steel factor) leads to Kit dimerization and activation of the PI3K/Akt and Ras/MAPK signaling pathways that regulate cellular proliferation and survival [PMID:10582339]. KIT activation through mutations is oncogenic [PMID:15175998].

KIT mutations have been reported in 55–65% of gastrointestinal stromal tumor samples [COSMIC, cBioPortal for Cancer Genomics].

Current data suggests that the efficacy of tyrosine kinase inhibitor therapies (i.e. imatinib) in patients with GIST is specific to tumors with activating KIT mutations, particularly in exon 9 and exon 11 [PMID:15897563, PMID:22089421, PMID:24925196, PMID:24790651].

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)):KIT NM_000222.

Specimen

MCR

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 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.

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 12:27
Reported: 01 May 2025 02:43
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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