

Specimen Collected: 09-Sep-21 10:15

Gastrointestinal Stromal Tumor Mutations | Received: 10-Sep-21 13:26 Report/Verified: 10-Sep-21 13:28

Procedure	Result	Units	Reference Interval
GIST Interpretation	Detected * <sup>i1</sup>		

**Test Information**

i1: GIST Interpretation  
Test information: Gastrointestinal Stromal Tumor Mutation

CHARACTERISTICS: This assay is designed to detect mutations in the KIT gene exons 9, 11, 13, 14, 17 or 18 and in PDGFRA gene exons 12, 14 or 18. Mutations in the KIT and PDGFRA genes may indicate responsiveness to certain targeted therapies. For specific treatment recommendations please refer to NCCN Clinical Practice Guidelines in Oncology for Soft Tissue Sarcoma (Gastrointestinal Stromal Tumor section) ([www.nccn.org](http://www.nccn.org)).

GENES TESTED: KIT gene (NM\_000222.2) exons 9, 11, 13, 14, 17, and 18 and PDGFRA gene (NM\_006206.4) exons 12, 14, and 18

METHODOLOGY: Genomic DNA is isolated from microscopically-guided dissection of tumor tissue and then enriched for the targeted regions of the tested genes. The mutation status of the targeted genes is determined by massively parallel sequencing (next generation sequencing). The hg19 (GRCh37) reference sequence is used as a reference for identifying genetic mutations.

LIMITATIONS: This test will not detect mutations in other locations within the KIT or PDGFRA genes, in other genes, or below the limit of detection. This test evaluates for variants in tumor tissue only and cannot distinguish between somatic and germline variants. It is possible that some large insertion/deletion mutations (especially those greater than 60bp) may not be detected. Tissue samples yielding at least 10ng are acceptable but may yield suboptimal results if yield is less than 50ng.

LIMIT OF DETECTION: 5 percent mutant allele frequency for single nucleotide variants (SNV) and small to medium sized multi-nucleotide variants (MNV) (insertions/deletions less than 60bp).

ANALYTICAL SENSITIVITY (PPA): Analytical sensitivity for all variant classes is available through this link: <http://ltd.aruplab.com/Tests/Pdf/294>.

CLINICAL DISCLAIMER: Results of this test must always be interpreted within the clinical context and other relevant data and should not be used alone for a diagnosis of malignancy. This test is not intended to detect minimal residual disease.

\*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

*Unless otherwise indicated, testing performed at:*

**ARUP Laboratories**

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Tracy I. George, MD

**ARUP Accession:** n/a

**Report Request ID:** 15047734

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**Test Information**

i1: GIST Interpretation

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

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