

### **Laboratory Service Report**

### 1-800-533-1710

Patient Name TEST,IMPLEMENTATION TESTING	Patient ID 321	Age 57	Gender F	<b>Order #</b> R1057509
Ordering Phys TESTING				<b>DOB</b> 05/23/1956
Client Order # R1057509	Account Information			Report Notes
<b>Collected</b> 06/06/2013 06:00	C7028846-DLMP Rochester 3050 Superior Drive			
<b>Printed</b> 07/17/2013 11:47	Rochester, MN 55901			

Reference Perform Test Flag Results Unit Value Site\* AP Special Studies Review **RECEIVED:** 06/06/2013 15:23 **REPORTED:** 06/07/2013 12:27 Accession Number HR13-193 MCR Specimen: MCR A:AP Slide Review Material: MCR 1 Block SLIDE DISPOSITION: Final Diagnosis: MCR Molecular Anatomic Pathology Report: Reason for referral: Gastrointestinal Stromal Tumor Specimen source: Stomach Negative for PDGFRA exon 18 mutation by PCR and sequencing. All controls worked appropriately. Interpretation: Several tumors can harbor KIT mutations, including gastrointestinal stromal tumor (GIST), mast cell disease, melanoma, seminoma, acute

myeloid leukemia, myeloproliferative neoplasms, and lymphomas. In addition, occasional cases of GIST can also harbor mutations in PDGFRA, a gene structurally related to KIT. The frequency and type of mutations vary among these tumors and portent distinct clinical implications. The result does not rule out the presence of a mutation that may be present but below the limit of detection for this assay (approximately 30%). GIST PDGFRA exon 18 mutations have been associated with susceptibility to tyrosine kinase inhibitors, except p. Asp842Val, which confers resistance to tyrosine kinase inhibitors. Mutational status should be correlated with clinical data. The ordering physician is responsible for the diagnosis and management of disease and decisions based on the data provided. False-negative results may occur in specimens when tumor cells comprise <60% of the cell population. Tumor cells are routinely enriched by macrodissection to avoid false-negative results. Clinical diagnosis and/or therapy should not be based solely on this assay. The results should be considered in conjunction with clinical information, histologic evaluation, and/or additional

Method:

Mutation analyses were performed using polymerase chain reaction (PCR) and sequencing for PDGFRA exon 18 on DNA extracted from paraffin-embedded tissue.

Cautions:

diagnostic tests.

This test is unable to distinguish between a somatic and a germline KIT (or PDGFRA) mutation. Germline KIT (or PDGFRA) mutations are

## \*\*\*Performing Site Legend on Last Page of Report\*\*\*

Patient Name	Collection Date and Time	Report Status
TEST,IMPLEMENTATION TESTING	06/06/2013 06:00	Final
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Unit

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rare and their clinical relevance has been described in more detail by Robson ME et al. Clin Cancer Res 2004; 10:1250-4 and Li FP et al. J Clin Oncol 2005; 23:2735-43. Testing of a peripheral blood specimen from this individual would be required to distinguish a germline from a somatic mutation. This test is currently not offered at Mayo Clinic.

Flag Results

Laboratory Developed Test!

Test

Signing Pathologist: See Below

Result:6/7/2013 12:27 Interpreted by: Pathologist X. Test, M.D.

Report electronically signed by Melissa L. Skjeveland

Transcribed by: mls 6/7/2013 12:27:14

Slide Review Performed MCR

**RECEIVED:** 06/06/2013 15:23 **REPORTED:** 06/07/2013 12:27

PDGFRA, Mutation Analysis, Ex18 Performed

**RECEIVED:** 06/06/2013 15:23 **REPORTED:** 06/07/2013 12:27

### \* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
TEST,IMPLEMENTATION TESTING	06/06/2013 06:00	Final
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<sup>\*</sup> Report times for Mayo performed tests are CST/CDT