

## **Laboratory Service Report**

## 1-800-533-1710

Patient Name TEST,IMPLEMENTATION TESTING	Patient ID 321	<b>Age</b> 57	Gender F	<b>Order #</b> R1057502
Ordering Phys TESTING				<b>DOB</b> 05/23/1956
Client Order # R1057502	Account Information			Report Notes
<b>Collected</b> 06/06/2013 06:00	C7028846-DLMP Rochester 3050 Superior Drive			
<b>Printed</b> 07/17/2013 08:46	Rochester, MN 55901			

Perform Reference Test Flag Results Unit Value Site\* AP Special Studies Review RECEIVED: 06/06/2013 12:49 REPORTED: 06/06/2013 18:46 Accession Number HR13-178 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 KIT exon 8 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%). KIT exon 8 mutations are very rare. 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 diagnostic tests. Method: Mutation analyses were performed using polymerase chain reaction (PCR) and sequencing for KIT exon 8 on DNA extracted from paraffin-embedded tissue. Cautions: This test is unable to distinguish between a somatic and a germline KIT (or PDGFRA) mutation. Germline KIT (or PDGFRA) mutations are rare and their clinical relevance has been described in more detail

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

by Robson ME et al. Clin Cancer Res 2004; 10:1250-4 and Li FP et al.

Patient Name	Collection Date and Time	Report Status
TEST,IMPLEMENTATION TESTING	06/06/2013 06:00	Final
Page 1 of 2		>> Continued on Next Page >>



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Reference

Perform

Test

Flag Results

Unit

Value

Site\*

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.

Laboratory Developed Test

Signing Pathologist: See Below

MCR

Result:6/6/2013 18:45 Interpreted by: Pathologist X. Test, M.D.

Report electronically signed by Melissa L. Skjeveland

Transcribed by: mls 6/6/2013 18:45:21

Slide Review

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**RECEIVED:** 06/06/2013 12:49 **REPORTED:** 06/06/2013 18:46

KIT, Mutation Analysis, Ex8

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**RECEIVED:** 06/06/2013 12:49 **REPORTED:** 06/06/2013 18:46

## \* Performing Site:

MCR Mayo Clinic Laboratories - Rochester Main Campus Lab Director: Franklin R. Cockerill, III, M.D.

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Page 2 of 2		** End of Report **