MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TEST,IMPLEMENTATION TESTING	321	57	F	R1055976
Ordering Phys TEST,DOCTOR				**DOB** 05/23/1955
Client Order # R1055976	**Account Information** C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			**Report Notes**
Collected 05/24/2012 06:00				
Printed 05/24/2012 15:10				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Pre-Analytic Process, MAP Lab			REPORTED 05/24/2012 12:57		
DNA/RNA Extraction, MAP Lab		Performed			MCR
AP Special Studies Review			REPORTED 05/24/2012 12:57		
Accession Number		HR12-160			MCR
Referring Pathologist/Physician					MCR
Doctor Test Jr., M.D.					
Ref Path/Phys Address					MCR
Methodist Hospital					
200 1st Street SW					
Rochester, MN 55905					
507-266-0740					
Specimen:					MCR
A:AP Slide Review					
Material:					MCR
1 block S-12-0000123					
SLIDE DISPOSITION:					
1 block returned 5/24/12. asb					
Final Diagnosis:					MCR

Molecular Anatomic Pathology Report:
Reason for referral: Gastrointestinal Stromal Tumor
Specimen source: Peritoneal Mass
Result:
Positive for KIT exon 11 c.1669_1674del p.Trp557_Lys558del 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 KIT exon 11 mutations have been associated with susceptibility 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.
Clinical diagnosis and/or therapy should not be based solely on this assay. The results should be considered in conjunction with clinical information and/or additional diagnostic tests.
Method:

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Mutation analyses were performed using polymerase chain reaction (PCR) and sequencing for the KIT exon 11 gene 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 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. Laboratory Developed Test					
Signing Pathologist:		See Below			MCR
Result: 5/24/2012 12:56 Interpreted by: Pathologist X. Test, M.D. Report electronically signed by Angie S. Beckel Transcribed by: asa05 5/24/2012 12:56:17					
				REPORTED 05/24/2012 12:57	
KIT, Mutation Analysis, Ex11		Performed			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

* Report times for Mayo performed tests are CST/CDT