MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MSH6M	SA00050445	37	M	SA00050445
Ordering Phys				**DOB** 06/15/1975
Client Order # SA00050445	**Account Information**			**Report Notes**
Collected 11/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 01/04/2013 15:39				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MSH6 Mutation Screen, B			REPORTED 11/19/2012 19:03		
Reason For Referral					MCR
Tumor from patient shows absence of protein expression for MSH6 only. Test for the presence of a mutation in the MSH6 gene and large deletions involving the TACSTD1/EPCAM gene.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR

This result does not rule out the diagnosis of Lynch syndrome. We predict that some individuals who have a diagnosis of Lynch syndrome and the involvement of MSH6 may have a mutation that is not identified by the methods described (e.g. promoter mutations and deep intronic mutations).

The absence of protein expression for MSH6 previously detected for the tumor of this patient could be the result of a somatic alteration rather than a germline mutation. Additionally, the clinical phenotype that is observed in this patient might be due to disease causing mutations in one of the other genes involved in DNA mismatch repair.

Consider germline testing for MSH2 gene mutations if this has not already been done.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Method					MCR
Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the MSH6 gene. Additionally, gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the MSH6 gene (all exons) and the TACSTD1/EPCAM gene (exons 3 and 8, 2 probes in 3-prime UTR, 1 probe 3kb downstream of TACSTD1/EPCAM, and 1 probe 2.5kb upstream of MSH2). Mutation nomenclature for MSH6 is based on GenBank accession number, NM_000179. Mutation nomenclature for TACSTD1/EPCAM is based on GenBank accession number, NM_002354.					
Specimen		Blood			MCR
Reviewed By:		D Brian Dawson PhD			MCR
Release Date		19 Nov 2012 19:02			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.