MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MSH2M	SA00041692	45	F	SA00041692
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00041692	**Account Information**			**Report Notes**
Collected 12/12/2011 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 12/15/2011 08:42				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MSH2 Mutation Screen			REPORTED 12/13/2011 10:01		
Specimen		Blood			MCR
Specimen ID		1037490			MCR
Order Date		13 Dec 2011 09:47			MCR
Reason For Referral					MCR

Tumor from patient demonstrates the absence of protein expression for MSH2 and MSH6. Test for the presence of MSH2 gene mutations and large deletions involving the TACSTD1/EPCAM gene.

Method MCR

DNA sequence analysis was performed to test for the presence of a mutation in all 16 exons of the MSH2 gene. Additionally, gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the MSH2 gene (all exons) and the TACSTD1/EPCAM gene (exons 3 and 8, 2 probes in the 3-prime untranslated region, 1 probe 3kb downstream of TACSTD1/EPCAM, and 1 probe 2.5kb upstream of MSH2). Mutation nomenclature for MSH2 is based on GenBank accession number, NM_000251. Mutation nomenclature for TACSTD1/EPCAM is based on GenBank accession number, NM_002354.

Result MCR

A mutation was NOT detected.

Interpretation MCR

This result does not rule out the diagnosis of HNPCC/Lynch syndrome. We predict that some individuals who have a diagnosis of HNPCC/Lynch syndrome and the involvement of MSH2 or TACSTD1/EPCAM may have a mutation that is not identified by the methods described above (e.g.mutations in the promoter, intron, etc.).

The absence of protein expression for MSH2 and 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 MSH6 gene mutations if this has not already been done.

A genetic consultation may be of benefit.

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*
A list of common polymorphisms identified for this patient is 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. 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.					
MLPA Performed?		Yes.			MCR
See #89853, MLH1/MSH2 Large Deletion/Duplication, MLPA, for billing information.					
Reviewed By:					MCR
Kandelaria Margarita Rumilla MD					
Release Date		13 Dec 2011 10:00			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.