

## **Laboratory Service Report**

## 1-800-533-1710

MCR

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Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MSH2M	SA00050440	37	M	SA00050440
Ordering Phys		·		<b>DOB</b> 06/15/1975
Client Order # SA00050440	Account Information			Report Notes
<b>Collected</b> 11/06/2012	3050 SUPERIOR DRIV	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE		
Printed 01/04/2013 15:32	ROCHESTER,MN 5590	01		

Test Flag Results Unit Value Site\*

MSH2 Mutation Screen REPORTED 11/19/2012 19:03

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.
Result

A mutation was NOT detected.

Interpretation

Reason For Referral

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 MSH2 or TACSTD1/EPCAM may have a mutation that is not identified by the methods described above (e.g. promoter mutations or deep intronic mutations).

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.

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.

#### CAUTTONS

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

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

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<sup>\*</sup> Report times for Mayo performed tests are CST/CDT



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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.

Method

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries 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 deletion of 2 probes in the 3'prime untranslated region of the TACSTD1/EPCAM gene. 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.

Specimen Blood
Reviewed By: D Brian Dawson PhD
Release Date 19 Nov 2012 19:01

### \* Performing Site:

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