MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MLHMS N	SA00058832	46	F	SA00058832
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058832	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 15:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*

MLH1 Mutation Screen
RECEIVED: 06/07/2013 12:14 **REPORTED:** 06/10/2013 16:03

Reason For Referral MCR

Tumor from patient demonstrates the absence of protein expression for MLH1 and PMS2. Test for the presence of a mutation in the MLH1 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 involving the MLH1 gene 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 MLH1 and PMS2 previously detected for the tumor of this patient could be the result of a somatic alteration rather than a germline mutation. MLH1 Hypermethylation and BRAF Mutation Analyses (Mayo test ID MLBRF/87931) may help to distinguish a somatic versus germline event. This testing can be performed on colon tumor that demonstrates loss of MLH1 protein expression if available for this individual. 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.

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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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 MLH1 gene. Additionally, gene dosage analysis (MLPA) is used to test for the presence of large deletions and duplications in the MLH1 gene. Mutation nomenclature is based on GenBank accession number, NM_000249.3.					
Specimen		Blood			MCR
Reviewed By:					MCR
Emily Christine Lauer					
Release Date		10 Jun 2013 16:00			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: