MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MLHKM A	SA00058830	46	F	SA00058830
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058830	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 15:31				

Test	Flag	Results	Unit	Reference Value	Perform Site*

MLH1 Known Mutation
RECEIVED: 06/07/2013 12:12 **REPORTED:** 06/10/2013 15:59

Reason For Referral MCR

Family history of Hereditary Nonpolyposis Colon Cancer Lynch Syndrome. Test for the presence of a mutation in the MLH1 gene.

Result MCR

The following deletion was detected:
Exon: 16
DNA change: c.1772_1775delATAG
Amino acid change: p.D591VfsX24 (Asp591ValfsX24)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of Lynch Syndrome. Appropriate screening procedures and/or prophylactic measures should be considered.

Identification of a mutation for this individual means that predictive testing can be performed for at risk family members.

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.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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marrow transplant.					
Laboratory developed test.					
Method					MCR
DNA sequence analysis was used to test for the presence of the c.1772_1775delATAG (p.D591VfsX24) mutation in exon 16 of the MLH1 gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. 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 15:57			MCR

* Performing Site:

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

TEST SITE