MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MLH3K	SA00057766	58	M	SA00057766
Ordering Phys CLIENT,CLIENT				**DOB** 06/15/1954
Client Order # SA00057766	**Account Information**			**Report Notes**
Collected 05/24/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/27/2013 12:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MLH3 Gene, Known Mutation					
RECEIVED: 05/24/2013 09:45 **REPORTED:** 06/10/2013 12:03					
Reason For Referral					MCR

An MLH3 mutation was previously identified in an affected family member. Test for the presence of a mutation in the MLH3 gene.

Result MCR

The following heterozygous deletion was detected:
Exon: 2
DNA change: c.2579delA
Amino Acid change: p.N860IfsX14 (Asn860IlefsX14)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

Mutations found in MLH3 are associated with an increased risk for colorectal cancer. In addition to colorectal cancer, individuals with a mutation in MLH3 can be at an increased risk for endometrial and esophageal cancers. Therefore, the cancer in this individual and/or family may be attributed to this result. Appropriate screening procedures 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 the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

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

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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considered.					
A previous bone marrow transplant from an allogenic donor 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
DNA sequence analysis was used to test for the presence of the c.2579delA (p.N860IfsX14) mutation in exon 2 of the MLH3 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_001040108.1.					
Specimen		Blood			MCR
Reviewed By					MCR
Matthew John Ferber PhD					
Release Date		10 Jun 2013 12:00			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.