Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MSH6M	SA00050449	34	M	SA00050449
Ordering Phys				**DOB** 06/15/1978
Client Order # SA00050449	**Account Information**			**Report Notes**
Collected 11/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 01/04/2013 15:40				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MSH6 Mutation Screen, B			REPORTED 11/19/2012 19:05		
Reason For Referral					MCR

Tumor from patient shows absence of protein expression for MSH6 only. Test for the presence of a mutation in the MSH6 gene and large deletions involving the TACSTD1/EPCAM gene.

Result MCR

The following deletion was detected:
Exon: 8
DNA change: c.3699_3702delAGAA
Amino acid change: p.K1233NfsX6 (p.Lys1233AsnfsX6)
Classification: DELETERIOUS

A mutation was not detected in TACSTD1/EPCAM.

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of Lynch syndrome for this individual. Appropriate screening measures and/or prophylactic measures should be considered.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing is available at Mayo Medical Laboratories by ordering MSH6K/83706 MSH6 Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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

Performing Site Legend on Last Page of Report

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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					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 MSH6 gene. Additionally, gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the MSH6 gene (all exons) and the TACSTD1/EPCAM gene (exons 3 and 8, 2 probes in 3-prime UTR, 1 probe 3kb downstream of TACSTD1/EPCAM, and 1 probe 2.5kb upstream of MSH2). Mutation nomenclature for MSH6 is based on GenBank accession number, NM_000179. Mutation nomenclature for TACSTD1/EPCAM is based on GenBank accession number, NM_002354.					
Specimen		Blood			MCR
Reviewed By:		D Brian Dawson PhD			MCR
Release Date		19 Nov 2012 19:02			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.