Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MLH12 N	SA00058928	47	F	SA00058928
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058928	**Account Information**			**Report Notes**
Collected 06/10/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/12/2013 10:59				

Test	Flag	Results	Unit	Reference Value	Perform Site*

MLH1/MSH2 Mutation Screen
RECEIVED: 06/11/2013 15:59 **REPORTED:** 06/12/2013 10:48

Reason For Referral MCR

Possible diagnosis of Lynch syndrome. Test for the presence of a mutation in the MLH1 and MSH2 genes as well as large deletions involving the TACSTD1/EPCAM gene.

Result MCR

A mutation was NOT detected in MLH1, MSH2, or TACSTD1/EPCAM.

Interpretation MCR

This result reduces the likelihood but does not rule out the diagnosis of Lynch syndrome for this individual. We predict that some individuals who have a diagnosis of Lynch syndrome and the involvement of MLH1, MSH2, or TACSTD1/EPCAM may have a mutation that is not identified by the methods described above (e.g. promoter mutations and deep intronic mutations).

Importantly, the clinical phenotype that is observed in this patient and/or family might be due to disease causing mutations in other genes involved in DNA mismatch repair. Testing the tumor from this individual or from an affected individual in this family for the presence of microsatellite instability (MSI) and for the expression of the DNA mismatch repair proteins MLH1, MSH2, MSH6, and PMS2 by immunohistochemistry (IHC) would be helpful for determining if defective mismatch repair is contributing to the clinical phenotype in this individual or family. In approximately 70-80% of cases where MSI and IHC demonstrate defective mismatch repair, a mutation is found in either MLH1 or MSH2.

If tumor is not available, consider germline analysis for mutations in MSH6 and PMS2.

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

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MLH12 N	SA00058928	47	F	SA00058928
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058928	**Account Information**			**Report Notes**
Collected 06/10/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/12/2013 10:59				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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 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 and MSH2 gene. Additionally, gene dosage analysis (MLPA)was used to test for the presence of large deletions and duplications in the MLH1 and MSH2 genes (all exons) and the TACSTD1/EPCAM gene (2 probes in the 3-prime untranslated region). Mutation nomenclature for MLH1 is based on GenBank accession number, NM_000249.3. Mutation nomenclature for MSH2 is based on GenBank accession number, NM_000251.1. Mutation nomenclature for TACSTD1/EPCAM is based on GenBank accession number, NM_002354.2.					
Specimen		Blood			MCR
Reviewed By:					MCR
Emily Christine Lauer					
Release Date		12 Jun 2013 10:47			MCR

* Performing Site:

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