Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00049760	11D	F	SA00049760
Ordering Phys				**DOB** 10/01/2012
Client Order # SA00049760	**Account Information**			**Report Notes**
Collected 10/12/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 11/08/2012 07:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*
PMS2 Gene, Full Gene Analysis			REPORTED 10/15/2012 09:17		
Reason for Referral					MCR

Tumor from patient shows absence of protein expression for PMS2 only. Test for the presence of a mutation in the PMS2 gene.

Result MCR

The following sequence change in PMS2 was detected:
Exon: 6
DNA change: c.697C>T
Amino acid change: p.Q233X (Gln233X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of Lynch syndrome for this individual. Appropriate screening procedures 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 PMS2K/61174 PMS2 Gene, 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 clinical findings, additional testing should be considered.

Bone marrow transplants from allogenic donors will interfere

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Method					MCR
Long range PCR and bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the PMS2 gene. Additionally, gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the PMS2 gene (all exons). Mutation nomenclature is based on GenBank accession number, NM_000535.5.					
Specimen		Blood			MCR
Reviewed By		Victoria Marie Waugh			MCR
Release Date		15 Oct 2012 09:15			MCR

* Performing Site:

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