Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00049762	11D	F	SA00049762
Ordering Phys				**DOB** 10/01/2012
Client Order # SA00049762	**Account Information**			**Report Notes**
Collected 10/12/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 11/08/2012 07:52				

Test	Flag	Results	Unit	Reference Value	Perform Site*
PMS2 Gene, Known Mutation			REPORTED 10/15/2012 09:33		
Reason for Referral					MCR

Family history of Lynch syndrome. 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. Appropriate screening procedures and/or prophylactic measures should be considered.

Identification of a mutation for this individual means that predictive testing can be erformed for other 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 marrow transplant.

Performing Site Legend on Last Page of Report

MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

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Ordering Phys				**DOB** 10/01/2012
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Printed 11/08/2012 07:52				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Method					MCR
DNA sequence analysis was used to test for the presence of the c.697C>T (p.Q233X) mutation in exon 6 of the PMS2 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_000535.5.					
Specimen		Blood			MCR
Reviewed By		Victoria Marie Waugh			MCR
Release Date		15 Oct 2012 09:32			MCR

* Performing Site:

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

* Report times for Mayo performed tests are CST/CDT