| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| TESTINGRNV,REPORTS | SA00049759 | 11D | F | SA00049759 |
| **Ordering Phys** | | | | **DOB** 10/01/2012 |
| **Client Order #** SA00049759 | **Account Information** | | | **Report Notes** |
| **Collected** 10/12/2012 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 11/08/2012 07:43 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **PMS2 Gene, Full Gene Analysis** | | | REPORTED 10/15/2012 09:15 | | |
| 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 |
| A mutation was NOT detected. | | | | | |
| Interpretation | | | | | MCR |

This result does not rule out the diagnosis of Lynch syndrome. We predict that some individuals who have a diagnosis of Lynch syndrome and the involvement of PMS2 may have a mutation that is not identified by the methods described (e.g. promoter mutations and deep intronic mutations).

The absence of protein expression for PMS2 previously detected for the tumor of this patient could be the result of a somatic alteration rather than a germline mutation. Additionally, the clinical phenotype that is observed in this patient might be due to disease causing mutations in one of the other genes involved in DNA mismatch repair.

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***

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# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| TESTINGRNV,REPORTS | SA00049759 | 11D | F | SA00049759 |
| **Ordering Phys** | | | | **DOB** 10/01/2012 |
| **Client Order #** SA00049759 | **Account Information** | | | **Report Notes** |
| **Collected** 10/12/2012 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 11/08/2012 07:43 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| 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 09:14 | | | MCR |

* Performing Site:

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



* Report times for Mayo performed tests are CST/CDT