

Patient Name SAMPLEREP,MLH3S	Patient ID SA00057772	Age 67	Gender M	Order # SA00057772
Ordering Phys CLIENT,CLIENT				DOB 06/12/1945
Client Order # SA00057772	Account Information			Report Notes
Collected 05/24/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/27/2013 12:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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MLH3 Gene, Full Gene Analysis
RECEIVED: 05/24/2013 11:25 **REPORTED:** 06/10/2013 12:56

Reason For Referral

MCR

Test for the presence of a mutation in the MLH3 gene.

Result

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The following heterozygous deletion was detected:

Exon: 2

DNA change: c.2579delA

Amino Acid change: p.N860IifsX14 (Asn860IleifsX14)

Classification: DELETERIOUS

Interpretation

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This alteration is a known deleterious mutation.

Mutations found in MLH3 are associated with an increased risk for colorectal cancer. In addition to colorectal cancer, individuals with a mutation in MLH3 can be at an increased risk for endometrial and esophageal cancers. Therefore, the cancer in this individual and/or family may be attributed to this result. Appropriate screening procedures should be considered.

Since a mutation has been identified, predictive testing of at risk family members is possible. Mutation-specific testing for MLH3 is available at Mayo Medical Laboratories by ordering MLH3K/61473 MLH3 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 the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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the clinical findings, additional testing should be considered.

A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Laboratory developed test.

Method

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the MLH3 gene. Additionally, array comparative genomic hybridization (aCGH) was used to test for the presence of large deletions and duplications. Mutation nomenclature for MLH3 is based on GenBank accession number NM_001040108.1.

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Array Billed?

Yes.

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See COLDB, Hereditary Colon Cancer CGH Array, for billing information.

Specimen

Blood

MCR

Reviewed By

Matthew John Ferber PhD

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Release Date

10 Jun 2013 12:53

MCR

* Performing Site:

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