MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

MSH2Z

MSH2 Gene, Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00000850	SAMPLEREPDMGLM, VLD20150713A0111		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00000850	SA00000850	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 15:45			

Result Summary

MCR

Positive

Result

MCR

The following heterozygous intronic alteration in MSH2 was identified:
DNA change: c.942+3A>T (g.47641560)
Classification: PATHOGENIC

Interpretation

1 MCR

The c.942+3A>T alteration is a known pathogenic 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, genetic testing of at risk family could be considered. Mutation specific testing is available at Mayo Medical Laboratories by ordering FMTT / Familial Mutation, Targeted Testing. 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.

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the MSH2 gene (GenBank accession number NM_000251;build GRCh37 (hg19)). Array comparative genomic hybridization (aCGH) was used to test for the presence of large deletions and duplications in the MSH2 gene and the TACSTD1/EPCAM gene (GenBank accession number NM_002354;build GRCh37 (hg19)). An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the 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 false-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.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

QA Environment

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

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Specimen MCR

WB Whole Blood

Released By MCR

EMILY LAUER

Array Billed? MCR

Yes. See COLAB, Hereditary Colon Cancer CGH Array, for billing information.

Received: 13 Jul 2015 20:22 **Reported:** 27 Jul 2015 12:50

QA Environment

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905