MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

MSH6Z

MSH6 Gene, Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00001010	SAMPLEREPDMGLM, VLD20150713A0271		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00001010	SA00001010	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 18:59			

Result Summary

MCR

Negative

Result

MCR

No reportable variants were identified.

Interpretation

1 MCR

This result decreases the likelihood but does not rule out the presence of a germline mutation in the MSH6 gene for this individual. We predict that there are individuals with a diagnosis of Lynch syndrome who have a pathogenic mutation that is not detectable by the methods described (e.g. promoter mutations or deep intronic mutations).

Additionally, this assay does not rule out the presence of mutations in other genes involved in DNA mismatch repair. Consider germline testing for MSH2 gene mutations if this has not already been done (MSH2Z / MSH2 Gene, Full Gene Analysis).

The absence of protein expression for MSH6 reported for the tumor of this individual could also be the result of a somatic alteration rather than a germline mutation. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

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 MSH6 gene (GenBank accession number NM_000179;build GRCh37 (hg19)). Array comparative genomic hybridization (aCGH) was used to test for the presence of large deletions and duplications in this gene. 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.

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 20:28 **Reported:** 24 Jul 2015 20:41

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