MAYO CLINIC LABORATORIES

1-800-533-1710

MLHPB

MLH1 Hypermethylation Analysis, Blood

Patient ID	Patient Name		Birth Date	Gender	Age
SA00000842	SAMPLEREPDMGLM, VLD20150713A0102		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00000842	SA00000842	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 15:32			

MLH1 Hypermethylation Analys, Blood

Result Summary

MCR

POSITIVE FOR GERMLINE METHYLATION

Result

MCR

Blood: Positive for MLH1 promoter hypermethylation

Interpretation

[1] MCR

This result is consistent with germline (constitutional) inactivation of MLH1 by promoter hypermethylation.

Individuals with germline/constitutional MLH1 promoter hypermethylation are at increased risk for other HNPCC/Lynch syndrome related tumors. Germline/constitutional hypermethylation has been reported in both familial and sporadic cases; therefore, other family members may be at risk.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

A PCR-based assay is used to test peripheral blood DNA for the presence of MLH1 promoter hypermethylation. 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 Clinic 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.

Reason for Referral

MCR

Tumor demonstrates loss of MLH1 protein expression and MLH1 promoter hypermethylation. Evaluate peripheral blood DNA for the presence of germline MLH1 promoter hypermethylation.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

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

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	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292