



Patient ID SA00000839	Patient Name SAMPLEREPDMGLM, VLD20150713A0099	Birth Date 1981-01-01	Gender M	Age 34
Order Number SA00000839	Client Order Number SA00000839	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Jul 2015 15:29		

MLH1 Hypermethylation Analys, Tumor

Result Summary

MCR

MLH1 HYPERMETHYLATION PRESENT

Result

MCR

Tumor tissue: Positive for MLH1 promoter hypermethylation

Interpretation

1 MCR

The loss of MLH1 protein expression observed in this patient's tumor is most likely due to the somatic/epigenetic inactivation of MLH1 (promoter hypermethylation). This patient is very unlikely to have a germline MLH1 mutation.

Testing for the presence of the BRAF V600E mutation may provide additional information (BRAFT / BRAF Mutation Analysis(V600E), Tumor). The BRAF V600E mutation is rarely detected in patients with a germline MLH1 gene mutation.

It is important to note that this result does not distinguish between somatic and germline hypermethylation. Testing for germline hypermethylation of the MLH1 Gene (MLHPB / MLH1 Hypermethylation Analysis, B) on an additional blood sample may help to distinguish between these two possibilities. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 for more information regarding germline testing for MLH1 hypermethylation.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Microscopic examination was performed by a pathologist only to identify areas of tumor for enrichment by macrodissection. A PCR-based assay is used to test tumor DNA for the presence of MLH1 promoter hypermethylation. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Reason for Referral

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Tumor demonstrates loss of MLH1 protein expression. Determine the likelihood of a germline MLH1 mutation versus the somatic/epigenetic inactivation of MLH1.

Specimen

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Tissue, Tumor

Tissue ID

MCR

S15-5999

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:22

Reported: 24 Jul 2015 16:00

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