MAYO CLINIC LABORATORIES

1-800-533-1710

BRMLH

MLH1 Hypermethylation and BRAF Mutation Analysis, Tumor

Patient ID	Patient Name		Birth Date	Sex	Age
SA00168951	TESTING, BRMLH ATALAS		1990-06-12	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00168951	SA00168951	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		14 Jun 2024 11:00			

MLH1 Hypermethylation/BRAF Mutation

MLH1 Hypermethylation/BRAF Mutation

Result Summary MCR

MLH1 HYPERMETHYLATION PRESENT/BRAF MUTATION IDENTIFIED

Result MCR

Provided diagnosis: colorectal adenocarcinoma

Methylation status: Positive for MLH1 promoter hypermethylation
BRAF status: Mutant (V600E, GTG>GAG)

Interpretation 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). In addition, the BRAF V600E mutation is rarely detected in patients with a germline MLH1 gene mutation. Taken together, these results suggest that a germline mutation within the MLH1 gene is very unlikely for this patient.

To date, our data suggests that the possibility of germline hypermethylation is very unlikely in tumors showing MLH1 promoter hypermethylation along with the BRAF mutation.

A genetic consultation may be of benefit.

Specimen MCR

Tissue, Tumor

Tissue ID MCR

1234

Method MCR

Microscopic examination was performed by a pathologist only to identify areas of tumor for enrichment by macrodissection. A PCR based assay was used to test tumor DNA for the presence of MLH1 promoter hypermethylation and tumor DNA for the presence of the V600E (Val600Glu) mutation in the BRAF gene.

Disclaimer [1] MCR

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.

Rare polymorphisms exist that could lead to false-negative or false-positive results.

A negative result does not rule out the presence of the V600E mutation that may be present but below the limits of detection for this assay (approximately 10%).

Released By MCR

ANDE RUMILLA

BRAF Analysis MCR

Performed

MLH1 Hypermethylation Analysis MCR

Performed

Received: 14 Jun 2024 13:24 **Reported:** 16 Jun 2024 15:22

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	Nikola Baumann Ph.D.	24D0404292