

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 7/19/1961
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

BRAF Codon 600 Mutation Detection with Reflex to MLH1 Promoter Methylation

ARUP test code 0051750

BRAF codon 600 Mutation Detection

Not Detected

A mutation in BRAF codon 600 was not detected. MLH1 promoter methylation testing will be performed.

This result has been reviewed and approved by [REDACTED]

INTERPRETIVE INFORMATION: BRAF codon 600 Mutation Detection with Reflex to MLH1 Promoter Methylation

Presence of a BRAF c.1799T>A, p.Val600Glu (V600E) mutation in a microsatellite unstable colorectal carcinoma indicates that the tumor is probably sporadic and not associated with Lynch syndrome (HNPCC). However if a BRAF mutation is not detected, the tumor may either be sporadic or Lynch syndrome associated. It should be noted that there have been rare reports of BRAF mutations in Lynch syndrome associated tumors, so the presence of a BRAF mutation does not completely exclude the possibility of Lynch syndrome.

Methodology:
DNA is isolated from microdissected tumor tissue and amplified for exon 15 of the BRAF gene. Mutation status is determined by pyrosequencing.

Limitations: Mutations in other locations within the BRAF gene or in other genes will not be detected.
Limit of detection: 10 percent mutant alleles.

Clinical Disclaimer: Results of this test must always be interpreted within the clinical context and other relevant data, and should not be used alone for a diagnosis of malignancy. This test is not intended to detect minimal residual disease.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

MLH1 Promoter Methylation (Reflex Only)

ARUP test code 0051747

MLH1 Promoter Methylation

Positive *

H=High, L=Low, *=Abnormal, C=Critical

This result has been reviewed and approved by Parisa Khalili, M.D.

MLH1 promoter methylation was detected.

TEST INFORMATION: MLH1 Promoter Methylation, Paraffin

MLH1 methylation is common in sporadic microsatellite unstable tumors, like colorectal cancer and endometrial cancer, and rarely occurs in Lynch syndrome (hereditary non-polyposis colon cancer or HNPCC). Therefore, the presence of MLH1 methylation suggests that the tumor is sporadic and not associated with Lynch syndrome. However, since there have been rare reports of Lynch syndrome-associated MLH1 methylation, all results should be interpreted within the clinical context. The lack of MLH1 methylation in a mismatch repair deficient tumor suggests that it may be associated with Lynch syndrome, and germline evaluation is suggested. Finally, low level MLH1 methylation is not reported as positive, since it does not correlate with MLH1 inactivation and microsatellite instability.

METHODOLOGY: DNA is isolated from tumor tissue microdissected from prepared slides. DNA is treated with sodium bisulfite, followed by amplification of a segment of the MLH1 promoter region using methylation specific real-time PCR. The MLH1 methylation level is calculated by comparison to the amplification of a reference gene.

LIMITATIONS: Methylation at locations other than those covered by the primers and probes will not be detected. Results of this test must always be interpreted within the clinical context and other relevant data, and should not be used alone for a diagnosis of malignancy. This test is not intended to detect minimal residual disease.

ANALYTICAL SENSITIVITY: Methylation levels below 10 percent are reported as negative.

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H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
BRAF codon 600 Mutation Detection	21-315-402299	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
MLH1 Promoter Methylation	21-315-402299	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 21-315-402299
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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