ARUP LABORATORIES | www.aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

HNPCC/Lynch Syndrome (MSH2) Sequencing and Deletion/Duplication

ARUP test code 0051654

MSH2 FGA Specimen	Whole Blood
MSH2 Full Gene Analysis	Negative

TEST PERFORMED - 0051654
TEST DESCRIPTION - HNPCC/Lynch Syndrome (MSH2) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the MSH2 gene.

INTERPRETATION
No pathogenic MSH2 gene variants were detected by sequencing the coding regions and intron/exon boundaries or deletion/duplication analysis. This result does not exclude Lynch syndrome/HNPCC as this individual may have an unidentified causative variant in MSH2 (e.g., deep intronic or regulatory region variant) or in another mismatch repair gene.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. If suspicion remains for a hereditary gastrointestinal cancer syndrome, consideration should be given to ordering the Gastrointestinal Hereditary Cancer Gene Panel (ARUP test code 2013449). Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000251.1 (MSH2)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Benign variants are not included in this report but are available upon request.

REFERENCES

This result has been reviewed and approved by Rong Mao, M.D.

H – high L – low * – abnormal C – critical

BACKGROUND INFORMATION: HNPCC/Lynch Syndrome (MSH2)
Sequencing and Deletion/Duplication
CHARACTERISTICS: Increased risk of colorectal and extra-colonic cancers including endometrial, renal pelvis, ureter, ovary, stomach, small intestine and hepatobiliary tract.
INCIDENCE: 1-2 percent of colorectal cancer is due to mismatch repair gene mutations.
INHERITANCE: Autosomal dominant
PENETRANCE: 80 percent lifetime risk of colorectal cancer; 20-60 percent risk for endometrial cancer.
CAUSE: Pathogenic Germline MLH1 MSH2, MSH6, and PMS2 gene mutations.
METHODS: Bi-directional sequencing of MSH2 coding regions and intron-exon boundaries; multiplex ligation-dependent probe amplification (MLPA) to detect large MSH2 exonic deletions and EPCAM (TACSTD1) exon 9 deletions.
TEST LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. The breakpoints of large/duplications will not be determined. Regulatory region mutations, deep intronic mutations and mutations in genes other than MSH2 will not be detected.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
MSH2 FGA Specimen	17-313-105390	11/9/2017 11:06:00 AM	11/13/2017 11:20:14 AM	11/13/2017 11:30:50 AM
MSH2 Full Gene Analysis	17-313-105390	11/9/2017 11:06:00 AM	11/13/2017 11:20:14 AM	11/13/2017 11:30:50 AM

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