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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:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

HNPCC/Lynch Syndrome (MSH6) Sequencing and Deletion/Duplication

ARUP test code 0051656

MSH6 FGA Specimen	Whole Blood
Lynch Syndrome (MSH6) Interpretation	Negative

TEST PERFORMED - 0051656
TEST DESCRIPTION - HNPCC/Lynch Syndrome (MSH6) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the MSH6 gene.

INTERPRETATION
No pathogenic MSH6 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 MSH6 (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_000179.2 (MSH6)
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 (MSH6)
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 OF MSH6 MUTATIONS: Risk of colorectal cancer is 40 percent in men and 20 percent in women up to age 80. Women also have a 40 percent risk for endometiral cancer up to age 80.
CAUSE: Pathogenic germline MLH1, MSH2, MSH6, and PMS2 gene mutations.
GENE TESTED: MSH6
CLINICAL SENSITIVITY: Approximately 5 percent of Lynch syndrome is due to MSH6 mutations.
METHODOLOGY: Bidirectional sequencing of MSH6 coding regions and intron-exon boundaries; multiplex ligation-dependent probe amplification (MLPA) to detect large MSH6 exonic deletions.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
TEST LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. The breakpoints of large deletions/duplications will not be determined. Regulatory region, deep intronic mutations and mutations in genes other than MSH6 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
MSH6 FGA Specimen	17-317-106848	11/13/2017 11:50:00 AM	11/13/2017 11:56:31 AM	11/13/2017 2:16:27 PM
Lynch Syndrome (MSH6) Interpretation	17-317-106848	11/13/2017 11:50:00 AM	11/13/2017 11:56:31 AM	11/13/2017 2:16:27 PM

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