ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	1/19/1962
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

HNPCC/Lynch Syndrome (PMS2) Sequencing and Deletion/Duplication

ARUP test code 0051737

PMS2 FGA Specimen	Whole Blood
Lynch Syndrome (PMS2) Interpretation	Negative

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.
TEST PERFORMED - 0051737
TEST DESCRIPTION - HNPCC/Lynch Syndrome (PMS2) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Predictive Testing

RESULT
No pathogenic variants were detected in the PMS2 gene.

INTERPRETATION
No pathogenic PMS2 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 PMS2 (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. These results should be interpreted in conjunction with results from MSH6, MSH2, and MLH1 sequencing and deletion/duplication tests, reported separately (see ARUP accessions 20-295-400938, 20-295-400931, and 20-295-400927). If suspicion remains for a hereditary cancer syndrome, consideration should be given to ordering the Hereditary Cancer Panel, Sequencing and Deletion/Duplication (ARUP test code 2012032). Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000535.5 (PMS2)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report, but are available upon request.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: HNPCC/Lynch Syndrome (PMS2) 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: Unknown for PMS2 mutations
CAUSE: Pathogenic germline MLH1, MSH2, MSH6, and PMS2 gene mutations.
GENE TESTED: PMS2
CLINICAL SENSITIVITY: Less than 5 percent of Lynch syndrome cases are due to PMS2 mutations.
METHODOLOGY: Bidirectional sequencing of PMS2 coding regions and intron-exon boundaries; multiplex ligation-dependent probe amplification (MLPA) to detect large PMS2 exonic deletions.
ANALYTICAL SENSITIVITY & SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations and deep intronic mutations will not be detected. Mutations in genes other than PMS2 are not evaluated. This assay is not designed to detect somatic variants associated with malignancy. Interpretation of this test result may be impacted if the patient has had an allogeneic stem cell transplantation.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
PMS2 FGA Specimen	20-295-400945	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lynch Syndrome (PMS2) Interpretation	20-295-400945	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