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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	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

HNPCC/Lynch Syndrome Deletion/Duplication

ARUP test code 2001728

HNPCC DD Specimen	Whole Blood
HNPCC/Lynch Syndrome, MMR Gene	PMS2
HNPCC Deletion/Duplication Interp	Negative

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

RESULT
No pathogenic mutations detected in the PMS2 gene.

INTERPRETATION
No large deletions or duplications in the PMS2 gene were detected by deletion/duplication analysis. This result does not exclude Lynch syndrome/HNPCC as base pair substitutions and small deletions/duplications in PMS2, as well as mutations in other mismatch repair genes, are not detected by this assay.

RECOMMENDATION
Medical screening and management should rely on clinical findings and family history. Sequence analysis for PMS2 is recommended to detect the majority of pathogenic PMS2 mutations. Genetic consultation is recommended.
Benign variants are not included in this report but are available upon request.

BACKGROUND INFORMATION: HNPCC/Lynch Syndrome (PMS2) 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: PMS2 penetrance is possibly lower than the other mismatch repair genes.
CAUSE: Germline MLH1, MSH2, MSH6, and PMS2 gene mutations.
GENE TESTED: PMS2.
METHODOLOGY: Multiplex ligation-dependent probe amplification (MLPA) to detect large PMS2 exonic deletions.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Rare diagnostic errors can occur due to probe site mutations. Single base pair substitutions, small deletion/duplications, regulatory region mutations, and deep intronic mutations will not be detected. The breakpoints of large deletions/duplications will not be determined. Mutations in genes other than PMS2 are not evaluated.

This result has been reviewed and approved by Wei Shen, Ph.D.

BACKGROUND INFORMATION: HNPCC/Lynch Syndrome Deletion/Duplication

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HNPCC DD Specimen	19-273-101572	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HNPCC/Lynch Syndrome, MMR Gene	19-273-101572	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HNPCC Deletion/Duplication Interp	19-273-101572	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