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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	[redacted]
Gender:	Male
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	MSH2	
HNPCC Deletion/Duplication Interp	**Deletion**	*

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

Positive for the requested deletion in the MSH2 gene.

DNA MUTATIONS
Pathogenic
Nucleic Acid Change: deletion of exon 8; Heterozygous

INTERPRETATION
The familial MSH2 gene deletion of exon 8 was detected by deletion/duplication analysis. This mutation was previously reported to be associated with Lynch syndrome/HNPCC in his/her family; therefore, this individual is predicted to be affected.

RECOMMENDATION
Genetic consultation, including a discussion of medical screening and management, is indicated.

COMMENTS
Reference Sequence: GenBank # NM_000251.1

Note: Since a familial positive control was not availible, a non-familial positive control was tested.

BACKGROUND INFORMATION: HNPCC/Lynch syndrome (MSH2) 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 to 60 percent risk for endometrial cancer.
CAUSE: Germline MLH1, MSH2, MSH6, and PMS2 gene mutations.
GENE TESTED: MSH2.
METHODOLOGY: Multiplex ligation-dependent probe amplification (MLPA) to detect large MSH2 exonic deletions and EPCAM (TACSTD1) exon 9 deletions.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Rare diagnostic errors can occur due to probe binding site mutations. Deletion/duplication breakpoints will not be determined. Single base pair substitutions and small deletion/duplication mutations will not be detected.

This result has been reviewed and approved by Rong Mao, M.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	19-102-401077	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HNPCC/Lynch Syndrome, MMR Gene	19-102-401077	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HNPCC Deletion/Duplication Interp	19-102-401077	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