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MLH1, MSH2, MSH6, EPCAM Analysis Results

PHYSICIAN	SPECIMEN	PATIENT
John Smith, MD **Comprehensive Medical Center** **1100 Grand Ave** **Away, GA 12345**	Specimen: **Blood** Draw date: **Aug 01, 2010** Accession date: **Aug 02, 2010** Report Date: **Aug 12, 2010**	Name: **Doe, Jane** Date of Birth: **April 1, 1492** Patient ID: **000000** Gender: **Female** Accession #: **00000000-BLD** Requisition #: **000000**

Test Results and Interpretation

POSITIVE FOR A DELETERIOUS MUTATION

Test Performed:	Result:	Interpretation:
EPCAM rearrangement analysis	No Mutation Detected	No Mutation Detected
MLH1 sequencing	No Mutation Detected	No Mutation Detected
rearrangement analysis	No Mutation Detected	No Mutation Detected
MSH2 sequencing	No Mutation Detected	No Mutation Detected
rearrangement analysis	No Mutation Detected	No Mutation Detected
MSH6 sequencing	No Mutation Detected	No Mutation Detected
rearrangement analysis	del exons 2-4	**Deleterious**

Analysis includes sequencing of all exons and adjacent intronic regions and large rearrangement (LR) testing of the MLH1, MSH2, and MSH6 genes and LR testing of the EPCAM gene. LR testing is performed by quantitative multiplex PCR and multiplex ligation-dependent probe amplification (MLPA) for MLH1 and MSH2 and MLPA for MSH6 and the 3' region of EPCAM. MLPA reagents used for this test have not been approved or cleared by the FDA. However, Myriad Genetic Laboratories, Inc. has validated the performance characteristics of this test. Rare interfering variants may exist which could lead to false positive or negative results. Testing at Myriad found that LR mutations account for ~17% of MLH1 and ~37% of MSH2 mutations. The classification and interpretation of all variants identified in this assay reflect the current state of scientific understanding at the time this report was issued. The classification and interpretation of such variants may change as new information becomes available.

It is our understanding that this patient was identified for testing due to a personal or family history suggestive of Lynch syndrome (hereditary non-polyposis colorectal cancer, HNPCC). The results of this analysis are consistent with the germline MSH6 mutation del exons 2-4, resulting in a deletion of exons 2-4 and premature truncation of the MSH6 protein. Although the exact risk of cancer conferred by this specific mutation has not been determined, deleterious mutations in MSH6 confer as much as a 69% risk of colorectal cancer in men and as much as an 30% risk of colorectal cancer in woman, and a 71% risk of endometrial cancer by age 70 (Hendriks YM et al. Gastroenterology 2004;127(1):17-25). First-degree relatives of this individual each have a one-in-two chance of having this mutation. Family members can be tested for this specific mutation with a single site analysis.

Please contact Myriad Professional Support at 1-800-469-7423 to discuss any questions regarding this result.

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These test results should only be used in conjunction with the patient's clinical history and any previous analysis of appropriate family members. It is strongly recommended that these test results be communicated to the patient in a setting that includes appropriate counseling. The accompanying Technical Specifications summary describes the analysis, method, performance characteristics, nomenclature, and interpretive criteria of this test. This test may be considered investigational by some states. This test and its performance characteristics were determined by Myriad Genetic Laboratories. It has not been reviewed by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.