CONFIDENTIAL

Single Site Analysis Result

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:
198insC *MLH1*	198insC	**Deleterious**

This test is designed to detect the specific mutation(s) or variant(s) indicated above. The classification and interpretation of all variants identified in this assay reflects the current state of scientific understanding at the time this report was issued. In some instances, the classification and interpretation of such variants may change as new scientific 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 MLH1 mutation 198insC, resulting in premature truncation of the MLH1 protein at amino acid position 78. Although the exact risk of cancer conferred by this specific mutation has not been determined, deleterious mutations in MLH1 confer as much as an 82% risk of colorectal cancer and a 60% risk of endometrial cancer by age 70. Other increased cancer risks include as much as a 13% risk of gastric cancer and a 12% risk of ovarian cancer by age 70. (Vasen HFA et al. Gastroenterology 1996;110:1020-1027; Aarnio M et al. International Journal of Cancer 1999;81:214-218). 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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