CONFIDENTIAL

Comprehensive COLARIS AP®
APC Analysis and *MYH* Mutation Panel 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 TWO COPIES OF A *MYH* MUTATION
COLORECTAL POLYPOSIS AND CANCER RISK MARKEDLY INCREASED

Test Performed:	Result:	Interpretation:
APC sequencing	No Mutation Detected	No Mutation Detected
comprehensive rearrangement	No Mutation Detected	No Mutation Detected
G382D (1145G>A) *MYH*	G382D (1145G>A), 2 copies	} **Deleterious**
Y165C (494A>G) *MYH*	No Mutation Detected	

Analysis consists of sequencing of all exons and immediately adjacent intronic regions of the APC gene and a comprehensive rearrangement test of APC by Southern blot, as well as analysis of the specific MYH mutations 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.

This analysis detected two (homozygous) copies of the germline MYH mutation, G382D (1145G>A). MYH-associated polyposis and colorectal cancer is a recessive disease. Since two copies of an MYH mutation are present, this individual is at increased risk for MYH-associated polyposis and colorectal cancer. Although the exact risks are unknown, the presence of two deleterious MYH mutation has been documented in recent literature to be associated with colorectal polyposis and cancer (Nat Genet 2002;30:227-232; Hum Mol Genet 2002;11:2961-2967). It has been recommended that the medical management of individuals who have two deleterious MYH mutations be similar to individuals known to carry a germline APC mutation, including regular colonoscopies and upper GI screening for duodenal polyps (N Engl J Med 2003; 348:791-799). Full siblings of this individual have at least a 25% chance of having inherited both copies of this MYH mutation. Genetic testing should be considered in these individuals.

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

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Qualifications Here

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.