

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 A DELETERIOUS MUTATION

<u>Test Performed:</u>	<u>Result:</u>	<u>Interpretation:</u>
APC sequencing comprehensive rearrangement	3804del3insG No Mutation Detected	Deleterious No Mutation Detected
G382D (1145G>A) MYH	No Mutation Detected	No Mutation Detected
Y165C (494A>G) MYH	No Mutation Detected	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.

The results of this analysis are consistent with the germline APC mutation 3804del3insG, resulting in premature truncation of the APC protein at amino acid position 1274. Although variation in polyp number occurs among individuals within families with identical APC mutations, without intervention, deleterious mutations in the APC gene confer as much as a 93% risk of colorectal cancer by age 50 (Bussey HJR, Familial Polyposis Coli; 1975, Baltimore, Johns Hopkins Press). Such mutations also confer a 5-12% risk of duodenal or periampullary cancer and increased albeit small risks of other tumors (Burt RW et al, Gastroenterology 2000;119:837-853). First degree relatives of this individual each have a one-in-two chance of having this mutation. It has been recommended that genetic testing for first degree relatives of an affected FAP patient be initiated at age ten (AGA Statement, Gastroenterology 2001;121:195-197). 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.

Director Name Here
Qualifications Here

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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.