CONFIDENTIAL

Comprehensive BRACAnalysis®
BRCA1 and *BRCA2* 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: **Jun 22, 2011**	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:
BRCA1 sequencing	No Mutation Detected	No Mutation Detected
comprehensive rearrangement	No Mutation Detected	No Mutation Detected
BRCA2 sequencing	S1970X (6137C>A)	**Deleterious**
comprehensive rearrangement	No Mutation Detected	No Mutation Detected

It is our understanding that this patient was identified for testing due to a personal or family history suggestive of hereditary breast and ovarian cancer. Analysis consists of sequencing of all translated exons and immediately adjacent intronic regions of the BRCA1 and BRCA2 genes, a test for five specific BRCA1 rearrangements, and a comprehensive rearrangement test of both BRCA1 and BRCA2 by quantitative PCR analysis (BRACAnalysis Rearrangement Test, BART). 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 BRCA2 mutation S1970X, resulting in premature truncation of the BRCA2 protein at amino acid position 1970. Although the exact risk of breast and ovarian cancer conferred by this specific mutation has not been determined, studies of this type of mutation in high-risk families indicate that deleterious mutations in BRCA2 may confer as much as an 84% risk of breast cancer and a 27% risk of ovarian cancer by age 70 in women (Am. J. Hum. Genet. 62:676-689, 1998). Mutations in BRCA2 have been reported to confer a 12% risk of a second breast cancer within five years of the first (J Clin Oncol 17:3396-3402, 1999), as well as a 16% risk of subsequent ovarian cancer (J Natl Cancer Inst 91:1310-1315, 1999). Additionally, studies have shown that BRCA2 mutations confer as much as a 7% risk of pancreatic cancer by age 80 (J Med Genet 42:711-9, 2005); however, this risk may be higher in families in which pancreatic cancer has previously been diagnosed (Cancer Res 64:2634-2638, 2004). This mutation may also confer up to an 8% risk of male breast cancer and 20% risk of prostate cancer by age 80 (J Natl Cancer Inst 99:1811-4, 2007; J Natl Cancer Inst 91:1310-1315, 1999), as well as increased (albeit low) risks of some other cancers. Each first degree relative of this individual has 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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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.