Single Site Analysis Result

PHYSICIAN

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Away, GA 12345

SPECIMEN

Specimen: **Blood**

Draw date: Aug 01, 2010 Aug 02, 2010 Accession date:

Report Date: Jun 22, 2010 **PATIENT**

Doe, Jane Name: April 1, 1492 Date of Birth: 000000 Patient ID:

Gender: Accession #: 00000000-BLD

Female

Requisition #: 000000

Test Results and Interpretation

POSITIVE FOR A DELETERIOUS MUTATION

Test Performed: Result: Interpretation: R1443X (4446C>T) BRCA1 R1143X (4446C>T) **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.

The results of this analysis are consistent with the germline BRCA1 mutation R1443X, resulting in premature protein truncation at codon 1443 of the BRCA1 protein. Although the exact risk of breast and ovarian cancer conferred by this specific mutation has not been determined, studies in high-risk families indicate that deleterious mutations in BRCA1 may confer as much as an 87% risk of breast cancer and a 44% risk of ovarian cancer by age 70 in women (Lancet 343:692-695, 1994). Mutations in BRCA1 have been reported to confer a 20% risk of a second breast cancer within five years of the first (Lancet 351:316-321, 1998), as well as a ten-fold increase in the risk of subsequent ovarian cancer (J Clin Oncol 16:2417-2425, 1998). This mutation may also confer an increased (albeit low) risk of male breast cancer (Am J Hum Genet 62:676-689, 1998), as well as 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.

Director Name Here **Qualifications Here**

Director Name Here 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.