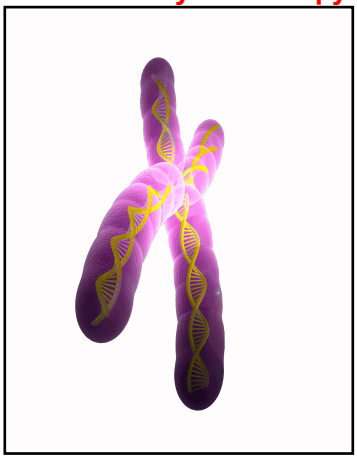
MEDICAL DIAGNOSTIC LABORATORIES L.L.C.
2439 KUSER ROAD, HAMILTON, NJ 08690-3303
TL: 609-570-1000, FX: 609-570-1050, TF: 877-269-0090
www.mdlab.com

Final

MDL#: 4328684 **Test Results** **Physician Copy**

Genetic Counselor Information:

Specimen Type:	Mouthwash
Date Collection:	1/25/2015
Date Processed:	1/26/2015
Date Reported:	3/2/2015



Patient Information: **SSN:** XXX-XX-5555 **DOB:** 1/1/1993 (Age: 22)

DOE, JANE
90 TRENTON ROAD
DAYTON, NJ 08690

Home: (142) 141-4113 **Patient ID:** 4444444

Ordering Physician/Lab: **NPI:** 2121212121

JOHN DOE MD
JOHN DOE, MD
202 ANY STREET
DAYTON, NJ 08810

Tel: 555-555-5551
Fax: 555-555-5555

Results Faxed To:
JANE DOE HOSPITAL

BRCAcare™ BRCA1 and BRCA2 Analysis Results, COMPREHENSIVE

Interpretation Summary:

POSITIVE FOR A PATHOGENIC MUTATION

Test Performed	Reference Sequence	Common Name	cDNA Change	Amino Acid Change	Exon	References	Interpretation
BRCA1 Sequencing						-	NO ANOMALIES DETECTED
BRCA1 Deletion / Duplication Analysis						-	NO ANOMALIES DETECTED
BRCA2 Sequencing	NC_000013.10	Y3308X	c.9924C>G	p.Tyr3308Ter	27	-	PATHOGENIC
BRCA2 Deletion / Duplication Analysis						-	NO ANOMALIES DETECTED

Comprehensive Interpretation:

Test Interpretation:
Sequencing of the coding regions and splice junction sites of the BRCA2 gene was done and was POSITIVE for the Y3308X change in the BRCA2 gene. This change has been associated with the Hereditary Breast Ovarian Cancer Syndrome (HBOC) and is considered to be PATHOGENIC. This change was identified by NGS and classified based on MDL BRCA variant classification system. This change has and others have been associated with 60-80% risks for the HBOC cancers.

In addition to the gene sequencing assay, a multiplex ligation-dependent probe amplification (MLPA) analysis which detects deletions and/or duplications involving one or more exon, including those that affect the entire BRCA1 and BRCA2 gene, was completed. No deletions or duplications were detected.

The classification and interpretation of all genetic variants identified as a result of this genetic testing is based on the current scientific information available. As new scientific information becomes available, in some circumstances, the classification and interpretation of the genetic variants may change.

Genetic counseling is advised to learn the full meaning of the test results and to discuss risks to other family members. Relatives should consider genetic counseling and testing. All test results should be interpreted by physician or genetic counselor in the context of the personal/family cancer history, and clinical and laboratory data.

View: M

Mail:	Yes	USPS
	All	Yes

Fax:	Yes	***Manual***
	All	No

Medical Director, Dante A. Ragasa, MD.

BR

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BRCAcare™ BRCA1 and BRCA2 Analysis Results, COMPREHENSIVE

Comprehensive Interpretation (continued):

Methods and Variant Classification:

The entire gene coding region of the BRCA1/BRCA2 genes, as well as all flanking non-coding regions, were analyzed by Next Generation Sequencing. The multiple-ligation-probe amplification assay (MLPA) was also performed to detect copy number variations (gross deletions and duplications) in the BRCA1 and BRCA2 genes. The MDL BRCA variant classification system is based on the 5-tier system recommendations for the interpretation of sequence variants proposed by the American College of Medical Genetics and Genomics (ACMG). To classify each variant, MDL assigns weight to each piece of available evidence, including literature review, reputable database reports, population frequencies, and computational evidence and prediction. Each identified variant is classified as Benign, Likely Benign, a Variant of Unknown Signficance, Likely Pathogenic, or Pathogenic. Variants determined to be benign are not reported, but are available upon request. MDL variant results are reported using numbering and nomenclature recommended by the Human Genome Variation Society (HGVS http://hgvs.org). Nucleotide and codon number are based on the reference sequence NC_000017.10 for the BRCA1 gene and NC_000013.10 for the BRCA2 gene.

Test Limitations:

This assay cannot detect mutations affecting gene regions not examined in the assay (e.g. most of the intronic regions).

Disclaimer:

This test was developed and its performance characteristics have been determined by Medical Diagnostic Laboratories, LLC. Performance characteristics refer to the analytical performance of the test. It is not been reviewed by the US Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

View: M

Mail:	Yes	USPS
	All	Yes

Fax:	Yes	***Manual***
	All	No

Medical Director, Dante A. Ragasa, MD.

BR

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3/3/2015
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