BRCAcare

DNA-based Next Generation Sequencing (NGS) technologies provide high sensitivity and accuracy for the detection of BRCA and other gene mutations.

Tests Available:

- BRCA1/2: Comprehensive BRCA Analysis
- BRCA1/2: Ashkenazi Jewish 3-site Mutation Analysis
- BRCA1 & BRCA2: Specific Site Analysis
- Testing for Hereditary Breast & Ovarian Cancer Syndrome (HBOC)
 - Breast Cancer High Risk Extended Panel Plus: 14 genes (BRCA1, BRCA2, CDH1, PTEN, TP53, STK11, ATM, CHEK2, PALB2, BARD1, BRIP1, MUTYH, RAD51C, RAD51D)
 - Breast Cancer High Risk Extended Panel (No BRCA1, BRCA2): 12 genes (CDH1, PTEN, TP53, STK11, ATM, CHEK2, PALB2, BARD1, BRIP1, MUTYH, RAD51C, RAD51D)
 - BRCA1/2: Ashkenazi Jewish 3-site Mutation Analysis (Reflex to Breast Cancer High Risk Extended Panel Plus if negative)
- Testing for Lynch Syndrome genes:



Comprehensive Hereditary Breast and Gynecologic Cancer Panel: 19 genes analyzed by Gene Sequencing and/or Deletion/Duplication Analysis (BRCA1, BRCA2, ATM, BARD1, BRIP1, CDH1, CHEK2, MUTYH, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53, EPCAM, MLH1, MSH2, MSH6, PMS2).

Targets genes of elevated risk for the development of breast, ovarian, and endometrial uterine cancer.

Advantages:

- Non-invasive
- Ease of collection
- Rapid turnaround time
- High sensitivity and specificity
- Genetic counseling can be arranged
- Patient friendly pricing

UNLOCK YOUR



C A D E











MEDICAL DIAGNOSTIC LABORATORIES L.L.C.

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Patient Information: SSN: XXX-XX-5555 DOB: 1/1/1993 (Age: 22) 4328684 **Test Results** MDL#: **Physician Copy** Senetic Counselor Information: 90 TRENTON ROAD **DAYTON, NJ 08690** Home: (142) 141-4113 Patient ID: 4444444 NPI: 2121212121 Ordering Physician/Lab: JOHN DOE MD JOHN DOE, MD 202 ANY STREET DAYTON, NJ 08810 1/25/2015 Date Collection Results Faxed To: Date Processed: 1/26/2015 555-555-5551

BRCAcare™ BRCA1 and BRCA2 Analysis Results, COMPREHENSIVE

Interpretation Summary:

Date Reported:

3/2/2015

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.

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View: M

Mail: Yes USPS
All Yes

Fax: Yes **Manual** All No

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MDL#: 4328684 3/3/2015 BR Final