BRCAcare

Las tecnologías de Secuenciación de nueva generación (NGS) de ADN proporcionan una alta sensibilidad y precisión para la detección de genes BRCA y otras mutaciones genéticas.

Pruebas disponibles:

- **BRCA1/2: Análisis integral de BRCA**
- **BRCA1/2: Análisis de la mutación judío-Ashkenazi en 3 regiones**
- **BRCA1 y BRCA2: Análisis de región específica**
- **Pruebas para el Síndrome de cáncer de mama y ovario hereditario (HBOC)**
  - **Panel ampliado para identificación de alto riesgo de cáncer de mama Plus:** (BRCA1, BRCA2, CDH1, PTEN, TP53, STK11, ATM, CHEK2, PALB2, BARD1, BRIP1, MUTYH, RAD51C, RAD51D)
  - **Panel ampliado para identificación de alto riesgo de cáncer de mama (Sin BRCA1, BRCA2): 12 genes** (CDH1, PTEN, TP53, STK11, ATM, CHEK2, PALB2, BARD1, BRIP1, MUTYH, RAD51C, RAD51D)
  - **BRCA1/2: Análisis de la mutación judío-Ashkenazi en 3 regiones** (reflejo del panel ampliado para identificación de cáncer de mama de alto riesgo Plus si es negativo)
- **Pruebas para los genes del Síndrome de Lynch:**
  - **Panel integral para identificación de cáncer de mama y ginecológico hereditario: 19 genes** analizados por secuenciación génica y/o análisis de deleción/duplicación (BRCA1, BRCA2, ATM, BARD1, BRIP1, CDH1, CHEK2, MUTYH, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53, EPCAM, MLH1, MSH2, MSH6, PMS2).

Ya en disponibilidad

Se enfoca en genes de alto riesgo para el desarrollo de cáncer de mama, de ovario y endometrial.

Ventajas:

- **No invasivo**
- **Facilidad de recogida**
- **Tiempo de entrega rápido**
- **Alta sensibilidad y especificidad**
- **Se puede organizar asesoría genética**
- **Precios amigables para el paciente**

UNLOCK YOUR BRCA CODE™

IH0188 Upd: 9_2023

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Medical Diagnostic Laboratories
www.mdlab.com • 877.269.0090

A DIVISION OF GENESIS CLINICAL DIAGNOSTICS

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

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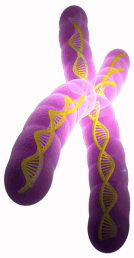
TL: 609-570-1000, FX: 609-570-1050, TF: 877-269-0090
www.mdlab.com

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, Jing Jing Yang, M.D.

BR

MDL#: 4328684
3/3/2015
Final