Now Available

Genetic Carrier Screening

Available from whole blood.

ACOG Recommends Offering Carrier Screening to All Women, Regardless of Ethnicity or Family History…

1274 Genetic Carrier Screening Panel (2 genes) includes:

- Cystic Fibrosis Core Test (23 major CFTR variants approved by ACOG/ACMG)
- Spinal Muscular Atrophy

1. American College of Obstetricians and Gynecologists Committee on Genetics. ACOG Committee Opinion No. 691: Carrier Screening for Genetic Conditions. *Obstet Gynecol* 2017 Mar;129(3):e41-e55.

IH0012 Upd: 9_2023

mdl
A MEMBER OF GENESIS BIOTECHNOLOGY GROUP

Medical Diagnostic Laboratories
www.mdlab.com • 877.269.0090

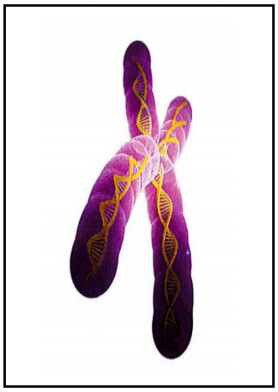
A DIVISION OF GENESIS CLINICAL DIAGNOSTICS

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

MDL#: 9649360 Final Test Results Physician Copy

Patient Information: SSN: XXX-XX-0001 DOB: 1/2/1974 (Age: 44)
DOE, JANE
123 MAIN ROAD
ANYTOWN, NJ 55555
Home: (555) 555-5555 **Patient ID:**

Genetic Counselor Information:



Specimen Type:	Blood
Date Collection:	8/28/2023
Date Processed:	8/29/2023
Date Reported:	9/4/2023

Ordering Physician/Lab: NPI: 1234567890
DOE WOMANS GROUP
JOHN DOE, MD
555 SMITH STREET
ANYTOWN, NJ 55555
Tel: (555) 555-5551
Fax: (555) 555-5552
Results Faxed To: /N

Interpretation Summary:

CARRIER OF A CF-CAUSING MUTATION;

Test Performed	Gene Transcript	Variant	Zygosity	Location	Disease	Inheritance	Parental Origin	Reference	Classification
CFTR Sequencing	NM_000492.3	c.350G>A (p.Arg117His)	Heterozygous	Exon4	Cystic Fibrosis	Autosomal recessive	Unknown	-	CF-Causing

Comprehensive Interpretation:

Test Interpretation:
The 23 Cystic Fibrosis-causing genetic sites recommended by the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) were tested and were determined to be **POSITIVE for the R117H**. This individual is a carrier of a CF-Causing Mutation.

Test Methodology:
The MDL Cystic Fibrosis Comprehensive Test is a next generation sequencing (NGS)-based CFTR gene analysis screen for 191 gene variants, including the 23 major mutations recommended by ACOG/ACMG for Cystic Fibrosis screening and the variants approved by the FDA for determining Ivacaftor treatment effectiveness. The complete list of all CF-causing mutations detected by this assay can be found at: http://www.mdlab.com/testing-menu. When a mutation from this list is detected, it is confirmed PCR amplification followed by Sanger DNA Sequencing prior to reporting.

Genetic testing was completed utilizing **Ion Torrent** (**Torrent Suite** v5.0.4, **variantCaller** v5.0.4.0) software, and using variant assembly **Genome Reference Consortium GRCh37** (UCSC version: hg19, rel. Feb. 2009).

Test Limitations:
Test Limitations:This assay cannot detect mutations affecting gene regions not examined by this assay. The 191 CFTR gene variants do not represent the complete list of possible CF-causing mutations. The prevalence of particular CF-causing mutations varies based upon the ethnic background of the patient.

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 has 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:			Fax:		
	Yes	USPS		Yes	*Manual*
	All	Yes		All	No

Medical Director, Jing-Jing Yang, M.D.

MDL#: 9649360 53085
9/4/2023
BR Final