Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003826 F 12/05/1988 34 Y

Referral Testing

Collected: 09/28/2023 15:29 Received: 09/28/2023 15:29

Test Name	Result	Flag	Ref-Ranges	Units	Site
Gaucher Disease, Mutation Analysis	See Below				WMQC

Test Name	Result	Flag	Ref Range
Gaucher Disease	NEGATIVE		

RESULT: NO PATHOGENIC VARIANT DETECTED

Interpretation: DNA testing indicates that this individual is negative for the pathogenic variants examined (see below) in the Gaucher disease (GD) gene (GBA). These pathogenic variants account for approximately 95% of GD pathogenic variants in the Ashkenazi-Jewish population. This result does not rule out the possibility that this individual carries a rare pathogenic variant in the GBA gene.

Gaucher disease (GD) is an autosomal recessive, lysosomal storage disease characterized by bone disease, hepatosplenomegaly, anemia and thrombocytopenia, lung disease, and the absence (Type 1) or presence (Types 2 and 3) of primary central nervous system disease. GD is caused by a deficiency of acid beta-glucosylceramidase activity, and the cellular accumulation of the glycolipid glucosylceramide (glucocerebroside). The acid beta-glucosylceramidase gene (GBA) is located on chromosome 1. GD is common in the Ashkenazi-Jewish population, with an estimated carrier frequency of 1 in 15. Four common pathogenic variants, N370S (c.1226A>G), IVS2+1G>A (c.115+1G>A), L444P (c.1448T>C), and 84GG (c.84dupG), in the GBA gene account for approximately 95% of GD pathogenic variants in Ashkenazi-Jews. The residual risk for an individual of full Ashkenazi-Jewish heritage to be a carrier of a GD pathogenic variant, after testing negative for the four common GD pathogenic variants is approximately 1 in 281. This assay tests for the four common GD pathogenic variants listed above. This assay also tests for four other, rare GD pathogenic variants, V394L (c.1297G>T), D409H (c.1342G>C), R496H (c.1604G>A), and del 55bp (c.1263 1317del55).

The eight GD pathogenic variants listed above are detected by multiplex-PCR amplification of specific gene regions, followed by nucleotide sequence analysis on a massively parallel sequencing platform.

Although rare, false positive or false negative results may occur. All results should be interpreted in the context of clinical findings, relevant history, and other laboratory data.

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Health care providers, please contact your local Quest Diagnostics' genetic counselor or call 1-866-GENEINFO (1-866-436-3463) for assistance with interpretation of these results.
Laboratory testing supervised and results monitored by Tina Hambuch-Hawks, Ph.D., DABMGG, CGMB.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Performed at Quest Diagnostics Nichols Institute
33608 Ortega Highway San Juan Capistrano, CA 92675-2042
Laboratory Director: I Maramica MD, PhD, MBA

Performing Site:
WMQC: QUEST DIAGNOSTICS REFERENCE LAB CAPISTRANO 33608 Ortega Highway San Juan Capistrano CA 92675

Reported Date: 2023.09.28 15:30

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F328000027
WX0000003826
Printed D&T: 09/28/23 15:30

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002353

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1