Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 09/27/2023 11:46 Received: 09/27/2023 11:46

Test Name	Result	Flag	Ref-Ranges	Units	Site
Hemochromatosis, Hereditary	See Below				QCRL

RESULT: POSITIVE FOR TWO COPIES OF THE HFE GENE PATHOGENIC VARIANT: C282Y/C282Y (HOMOZYGOTE)

Interpretation: Two copies of the C282Y pathogenic variant in the HFE gene were detected. This patient is negative for the H63D pathogenic variant. Approximately 60% to 90% of individuals with a biochemical diagnosis of hereditary hemochromatosis (HH) have this genotype. Therefore, this result is consistent with a diagnosis of HH in an individual with clinical evidence of HH. However, this genotype does not predict a diagnosis of HH in an asymptomatic individual, as not all individuals with this genotype will develop symptoms or clinical evidence of this disorder. Disease diagnosis can only be made by demonstration of elevated iron stores. Consider genetic counseling and DNA testing for at-risk family members.

Laboratory testing supervised and results monitored by Tina Hambuch-Hawks, Ph.D., DABMGG, CGMB.

DETAILED ASSAY INFORMATION: Hereditary hemochromatosis (HH) is an autosomal recessive disorder of iron metabolism that can result in iron overload and potential organ failure. It is one of the most common genetic disorders in individuals of European-Caucasian ancestry, with an estimated carrier frequency of 10%. HH is caused by pathogenic variants in the HFE gene. Most individuals with HH (60-90%) are homozygous for the C282Y pathogenic variant. A smaller percentage of affected individuals are either compound heterozygous for the C282Y and H63D pathogenic variants (3%-8%), or homozygous for the H63D pathogenic variant (approximately 1%).

METHODOLOGY: This assay detects two pathogenic variants in the HFE gene, C282Y (NM 000410.2: c.845G>A, p.Cys282Tyr) and H63D (NM 000410.2: c.187C>G, p.His63Asp), that are commonly associated with HH. These variants are detected by multiplex-polymerase chain reaction (PCR) amplification, followed by restriction enzyme digestion and capillary electrophoresis.

LIMITATIONS: This assay does not detect other pathogenic variants in the HFE gene that may be associated with HH. 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.

Health care providers, please contact your local Quest Diagnostics' genetic counselor for assistance with the interpretation of these results.

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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.

For more information, please refer to (This link is being provided for informational/educational purposes only.)
Reviewed and signed by Laboratory testing supervised and results monitored by Tina Hambuch-Hawks, Ph.D., DABMGG, CGMB
Test Performed at:
Quest Diagnostics Nichols Institute
33608 Ortega Highway
San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

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

Reported Date: 2023.09.27 11:46

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

F327000027
WX0000003827
Printed D&T: 09/27/23 11:47

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002365

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