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 11:48 Received: 09/27/2023 11:48

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

RESULT: NEGATIVE

Interpretation: DNA testing indicates that this individual is negative for the C282Y and H63D pathogenic variants in the HFE gene. This negative result significantly reduces the likelihood of hereditary hemochromatosis (HH) in this individual. However, it does not rule out the presence of other pathogenic variants within the HFE gene or a diagnosis of HH. The risk of this individual to carry an HFE pathogenic variant other than those tested in this assay depends greatly on family and clinical history as well as ethnicity. This assay does not test for other primary or secondary iron overload disorders.

Laboratory results and submitted clinical information reviewed by Guity Ghaffari, Ph.D., FACMG, HCLD, 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.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols

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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 results and submitted clinical information reviewed by Guity Ghaffari, Ph.D., FACMG, HCLD, 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 11:48

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

F327000028
WX0000003827
Printed D&T: 09/27/23 11:49

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002365

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