Patient ID	Patient Name		Birth Date	Gender	Age
SA00000970	SAMPLEREPDMGLM, VLD20150713A0231		1981-01-01	M	34
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00000970	SA00000970	CLIENT, CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		12 Jul 2015 18:12			

Hemochromatosis HFE Gene Analysis, B

Result Summary

MCR

NEGATIVE

Result

MCR

C282Y: Not detected.
H63D: Not detected.

Interpretation

[1] MCR

This result reduces the risk but does not rule out either a diagnosis of or predisposition for hereditary hemochromatosis (HH).

In the North American Caucasian population, approximately 5 to 8% of individuals with HH do not have either the p.C282Y or p.H63D mutation. For other ethnicities, the proportion of individuals with HH who do not have either the p.C282Y or p.H63D alteration may differ. This assay does not rule out the presence of other disease-causing mutations in the HFE gene or in other genes associated with hemochromatosis.

Genotyping results should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. serum transferrin-iron saturation and serum ferritin). Genetic testing and other laboratory testing of an affected family member can determine if this result is of predictive value for this individual.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Specimen

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WB Whole Blood

Method

MCR

A multiplex PCR based assay utilizing the Agena Mass Array platform was used to test for the following three mutations in the HFE gene; C282Y, H63D, and S65C. Because of the minimal effect on iron metabolism associated with the S65C mutation, it is only reported when it is found with the C282Y mutation (i.e. if the patient has the C282Y/S65C genotype).

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:27 **Reported:** 24 Jul 2015 19:00

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292