Patient ID	Patient Name		Birth Date	Gender	Age
SA00000812	SAMPLEREPDMGLM, VLD20150713A0072		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00000812	SA00000812	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 14:59			

Hemochromatosis HFE Gene Analysis, B

Result Summary

MCR

COMPLEX (SEE RESULT AND INTERPRETATION)

Result

MCR

C282Y: Two copies of the C282Y mutation were identified.
H63D: Not detected.

Interpretation

[1] MCR

This result may be consistent with, but does not confirm a diagnosis of or predisposition for hereditary hemochromatosis (HH).

Gender and serum ferritin level are known to impact the penetrance of the p.C282Y allele. Men homozygous for p.C282Y mutations are at considerably higher risk to exhibit symptoms than women with the same genotype. Individuals with two copies of the p.C282Y mutation and a serum ferritin level greater than 1000 ug/L are at increased risk for symptoms related to iron overload.

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).

Since the p.C282Y mutation has been identified, genetic testing and clinical evaluation of at risk family members could be considered.

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

MCR

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 20:21 **Reported:** 24 Jul 2015 13:24

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