

Patient ID SA00152248	Patient Name TESTINGABNORMAL, HFET	Birth Date 1975-04-02	Sex F	Age 47
Order Number SA00152248	Client Order Number SA00152248	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Apr 2022 12:20		

Hereditary Hemochromatosis HFE Test

Result Summary

MCR

COMPLEX (SEE RESULT AND INTERPRETATION)

Result

MCR

C282Y: Not detected.

H63D: One copy of the H63D variant was identified.

Interpretation

1 MCR

This individual is heterozygous for H63D. This result is consistent with at minimum a carrier status for hereditary hemochromatosis (HH).

The presence of a single H63D variant is unlikely to be of clinical significance in the absence of other disease-causing variants (1, 2).

This assay does not rule out the presence of other disease-causing variants in the HFE gene or in other genes associated with hemochromatosis.

Penetrance of the genotype may be affected by ethnic background and coexisting comorbid and environmental factors. These 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.

REFERENCES:

- Gochee PA, Powell LW, Cullen DJ, Du Sart D, Rossi E, Olynyk JK: A population-based study of the biochemical and clinical expression of the H63D hemochromatosis mutation. Gastroenterology. 2002 Mar;122(3):646-51. (PMID: 11874997)
- Kowdley KV, Brown KE, Ahn J, Sundaram V: ACG Clinical Guideline: Hereditary Hemochromatosis. Am J Gastroenterol. 2019 Aug;114(8):1202-1218. (PMID: 31335359)

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.

One or more in silico tools were used to assist in the interpretation of these results. These tools are updated regularly and predictions for a given variant may change. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated.

Specimen

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WB Whole Blood

Method

MCR

Droplet Digital Polymerase Chain Reaction (ddPCR) was used to test for the following three variants in the HFE gene; C282Y, H63D, and S65C. Because of the minimal effect on iron metabolism associated with the S65C variant, it is only reported when it is found with the C282Y variant (i.e. if the patient has the C282Y/S65C genotype).

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



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Released By

MCR

LINDA HASADSRI

Received: 28 Apr 2022 08:53

Reported: 28 Apr 2022 15:43

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.

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