

Patient Name TESTING,81508	Patient ID	Age	Gender	Order # W2921123
Ordering Phys		DOB		
Client Order # W2921123	Account Information C7999998-STUSTEST 200 FIRST STREET SW ROCHESTER, MN 55901	Report Notes		
Collected 10/14/2009 06:00				
Printed 10/14/2009 16:13	(507)266-5730			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Hemochromatosis HFE Gene Analysis, B			REPORTED 10/14/2009 12:28		
Specimen		Blood			MCR
Specimen ID		756319			MCR
Order Date		09 Oct 2009 11:13			MCR
Method					MCR
<p>A PCR-based assay was utilized 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).</p>					
Results					MCR
<p>C282Y: Homozygous. Two copies of the C282Y mutation identified H63D: not detected.</p>					
Interpretation					MCR
<p>This finding confirms the diagnosis or the predisposition for Hereditary Hemochromatosis (HH). These results need to be interpreted in the context of the clinical presentation and the results of other laboratory tests. Gender and serum ferritin level are known to impact penetrance of C282Y. Men with homozygous C282Y mutations are at considerably higher risk than women with the same genotype to exhibit symptoms. Additionally, individuals with two copies of the C282Y mutation and a serum ferritin level greater than or equal to 1000 ug/L are at increased risk for symptoms related to iron overload.</p> <p>This individual's result has important implications for other family members. A clinical and laboratory evaluation, including genetic testing, should be considered for at risk individuals. However, a number of important issues, such as psychosocial effects, insurability, and employability must be kept in mind prior to genetic testing (presymptomatic or (carrier) for HH.</p> <p>A genetic consultation may be of benefit.</p> <p>CAUTIONS: Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.</p>					

Performing Site Legend on Last Page of Report

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Rare polymorphisms exist that could lead to false negative or 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 Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reviewed By:

MCR

D Brian Dawson PhD

Release Date

14 Oct 2009 09:37

MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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