

1-800-533-1710

PATIENT NAME			PATIENT NU			AGE	SEX	ACCESSION #
TESTING, FECHS		L3MRNG91	L3MRNG9158585		34	F	G9158585	
ORDERING PHYSICIAN		CLIENT ORD	CLIENT ORDER #				ACCOUNT#	
								LIAISONS
COLLECTION	RECEIVED		REPORT PRI	NTED	SPECIMEN INFORMA	TION		
10/25/10 10:40 A	10/25/10 1	0:40 A	11/16/10	10:28 A	DATE OF BIRTH:			
DATE TIME	DATE	TIME	DATE	TIME				
Test Client								
Attn: Mayo Liaisons								
200 First Street SW								
Rochester, MN 55905								
507-284-8202								

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TEST REQUESTED LO REF RANGE PERFORM SITE *

FECH Gene, Full Gene Analysis

SpecimenBloodMCRSpecimen ID1034829MCROrder Date25 Oct 2010 10:44MCRReason for ReferralMCR

Not provided. Test for the presence of mutations in the FECH

Method MCR

DNA sequence analysis was performed to test for the presence of mutations in all 11 exons of the FECH gene. Mutation nomenclature is based on GenBank accession number; NM 000140.3.

Result MCR

A mutation was NOT detected.

Interpretation MCR

This result decreases the likelihood that this individual is a carrier of EPP due to ferrochelatase deficiency. Although a mutation in the FECH gene was not identified, this result does not entirely rule out the possibility that this individual is a carrier of EPP because not all FECH mutations are identifiable by the method described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations).

In addition, this assay does not rule out the presence of mutations in other genes that are associated with EPP, such as ALAS2.

It is often helpful to test the affected individual or the parent of an affected individual. Identification of the disease causing mutations in this family would allow for more direct testing and risk assessment of at risk individuals.

* Perform Site Legend on last page of report

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TESTING, FECHS	Final	10/25/10 10:40 A



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A list of common polymorphisms identified for this patient is available upon request.

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.

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.

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

0.5. Food and Drug Administration.

Extraction Performed? Yes MCR Reviewed By MCR

Michelle Dawn Fjerstad

Release Date 25 Oct 2010 10:49 MCR

* PERFORMING SITE

MCR Mayo Clinic Dpt of Lab Med & Pathology
200 First Street SW Rochester, MN 55905

Lab Director: Franklin R. Cockerill, III, M.D.

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TESTING, FECHS	Final	10/25/10 10:40 A