

Laboratory Service Report

1-8	300	-533-1	710

Patient Name TESTINGRNV,REPORTS	Patient ID SA00124383	Age 24	Gender M	Order # SA00124383
Ordering Phys CLIENT,CLIENT		•	•	DOB 01/01/1990
Client Order # SA00124383	Account Information			Report Notes
Collected 04/02/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support			
Printed 04/08/2014 08:48	Rochester, MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Wilson Disease Mutation Screen		4 17 06			
RECEIVED: 04/02/2014 09:13 REPORT	ED: 04/07/201				MCD
Specimen		Blood 1105319			MCR MCR
Specimen ID			1.0		MCR MCR
Order Date		02 Apr 2014 09:1	10		
Reason For Referral		-+ i 6 Mill			MCR
Patient reported to have f			D.		
disease. Test for the pres	sence or mutat	ions in the AIP/	В		
gene.					Man
Method	. 1		e		MCR
Bi-directional sequence ar			ior		
the presence of mutations	_	-			
intron/exon boundaries of	-	_			
sequence analysis for the					
(-441427del15) was perfo			1S		
based on GenBank accession	number;NM_00	0053.3.			Man
Result	,	1			MCR
The following homozygous s	sequence chang	e was detected:			
Exon: 14					
DNA change: c.3207C>A					
Amino Acid change: p.H1069	~ `	.)			
Classification: DELETERIOU	JS				
Interpretation					MCR
The p.H1069Q alteration is	a known dele	terious mutation	•		
This result is consistent	i+b a diama	sis of Wilson			

This result is consistent with a diagnosis of Wilson disease.

Since mutations have been identified in the ATP7B gene, genetic testing of other at risk family members is possible. Mutation-specific testing for Wilson disease is available at Mayo Medical Laboratories by ordering WDKM/83698 Wilson Disease Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

Mayo Clinic is investigating the use of newborn screening blood spots for the early detection of Wilson disease. Please contact the biochemical genetics laboratory at 1-507-266-8158 for more information about participating in this research study.

A genetic consultation may be of benefit.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status		
TESTINGRNV,REPORTS	04/02/2014 00:00	Final		
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^{*} Report times for Mayo performed tests are CST/CDT



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Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are 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.

Laboratory developed test.

Reviewed By: Release Date D Brian Dawson PhD 07 Apr 2014 17:24

MCR MCR

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status	
TESTINGRNV,REPORTS	04/02/2014 00:00	Final	
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