MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTING,83697				W3001170
Ordering Phys		**DOB**		
Client Order # W3001170	**Account Information** C7999998-STUSTEST 200 FIRST STREET SW ROCHESTER, MN 55901 (507)266-5730	**Report Notes**		
Collected 10/22/2009 06:00				
Printed 10/22/2009 09:37				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Wilson Disease Mutation Screen		REPORTED 10/22/2009 09:28			
Specimen		Blood			MCR
Specimen ID		754047			MCR
Order Date		30 Sep 2009 14:47			MCR
Reason for Referral					MCR
Patient reported to have features suggestive of Wilson disease. Test for the presence of mutation(s) within the ATP7B gene					
Method					MCR
Fluorescent DNA sequence analysis was used to test for the presence of mutations in the 21 exons of the ATP7B gene (GenBank accession number; NM_000053.2). Additionally, sequence analysis for the common Sardinian promoter mutation (-441_-427del15) was performed.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR
This result is not consistent with a diagnosis of Wilson disease. Some individuals with a diagnosis of Wilson disease may have mutations that cannot be detected by this method, although it is unlikely (<0.25%) that an affected individual would have two undetectable, causative mutations.					
There is significant variation in the reporting of the sensitivity of complete gene sequencing assays. Much of this is due to the presence of large genomic rearrangements and alterations outside of the sequenced regions, which would be missed by this methodology. The exact frequency of these types of mutations is unknown, but is estimated to be low in Wilson disease. Therefore, the risk estimate provided is our best approximation given the scientific information available to us at this time.					
This result should be interpreted within the context of all other pertinent clinical and laboratory analyses. Additional laboratory testing (e.g. Ceruloplasmin, serum; 24hr urine copper, copper, serum) and clinical evaluations may help to clarify the diagnosis for this individual.					
A genetic consultation may be of benefit.					
A list of common polymorphisms identified for this patient is available upon request.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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.

Reviewed By: MCR
W Edward Highsmith Jr., PhD
Release Date 13 Oct 2009 13:29 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.