MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CFTRK	SA00045691	45	F	SA00045691
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045691	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/14/2012 14:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CFTR Gene, Known Mutation		REPORTED 07/13/2012 09:58			
Specimen		Blood			MCR
Specimen ID		1038144			MCR
Order Date		08 May 2012 08:19			MCR
Reason For Referral					MCR
Family history of Cystic Fibrosis (CF). Test for the presence of mutations in the CFTR gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.R117H (c.482G>A) and p.deltaF508 (c.1653_1655delCTT) alterations in exons 4 and 10, respectively, of the CFTR gene. Testing was performed for these specific alterations because they were previously identified in an affected family member of this individual. Mutation nomenclature is based on GenBank accession number; NM_00492.3.					
Result					MCR
The familial mutations were NOT identified.					
Interpretation					MCR
Absence of the mutation(s) previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population to develop CF.					
This assay does not rule out the presence of other mutations within the CFTR gene. Errors in the information or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of the test results.					
A genetic consultation may be of benefit.					
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: 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.					
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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information provided is inaccurate or incomplete.					
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.					
Extraction Performed?		Yes			MCR
Reviewed By: Melody Elizabeth Kimball					MCR
Release Date		13 Jul 2012 09:55			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: