MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CFTRM	SA00045694	45	F	SA00045694
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045694	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/14/2012 15:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CFTR Gene, Full Gene Analysis		REPORTED 07/13/2012 09:58			
Specimen		Blood			MCR
Specimen ID		1038147			MCR
Order Date		08 May 2012 08:20			MCR
Reason For Referral					MCR
Patient has a possible diagnosis of cystic fibrosis (CF). Test for the presence of mutation(s) in the CFTR gene.					
Method					MCR
Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the CFTR gene. Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications. Mutation nomenclature is based on GenBank accession number; NM_00492.3.					
Result					MCR
The following homozygous deletion was detected: Exon: 10 DNA change: c.1653_1655delCTT Amino acid change: deltaF508 (p.F508del (Phe508del)) Classification: DELETERIOUS Intron 8 polyT alleles: 9T/9T					
Interpretation					MCR
The p.deltaF508 alteration is a known deleterious mutation. This finding is consistent with a diagnosis of CF or a CFTR-related disorder. Since mutations have been identified for this individual, carrier testing of other at risk family members is possible. Intron 8 poly T tract alleles (5T/7T/9T) are benign polymorphic variants. However, the 5T allele has been associated with variant CFTR-related disorders including CBAVD (congenital bilateral absence of the vas deferens). Since mutations have been identified, testing of at risk family members is possible. Mutation-specific testing for CF is available at Mayo Medical Laboratories by ordering CFTRK/88880 CFTR Gene, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test. A genetic consultation may be of benefit.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CFTRM	SA00045694	45	F	SA00045694
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045694	**Account Information**			**Report Notes**
Collected 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/14/2012 15:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*

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.

Extraction Performed?		Yes		MCR
MLPA Performed?		Yes.		MCR
See #89852, CFTR Large Del/Dup, MLPA, for billing information.				
Reviewed By:				MCR
Melody Elizabeth Kimball				
Release Date		13 Jul 2012 09:56		MCR

* Performing Site:

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