

### **Laboratory Service Report**

# 1-800-533-1710

Patient Name SAMPLEREPORT,CFTRK	Patient ID SA00045692	<b>Age</b> 45	Gender F	<b>Order #</b> SA00045692
Ordering Phys				<b>DOB</b> 06/10/1966
Client Order # SA00045692	Account Information			Report Notes
<b>Collected</b> 05/06/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
<b>Printed</b> 09/14/2012 14:59	ROCHESTER,MN 55	901		

Test	Flag	Results	Unit	Reference Value	Perform Site*
FTR Gene, Known Mutation			REPORTED 07	7/13/2012 09:58	
Specimen		Blood			MCR
Specimen ID		1038145			MCR
Order Date		08 May 2012 08:19			MCR
Reason For Referral					MCR
Family history of Cystic	Fibrosis (CF).	Determine carrier			
status.					
Method					MCR
DNA sequence analysis was		_			
the p.R117H (c.482G>A) as	-	• – .			
alterations in exons 4 as					
gene. Testing was perform					
because they were previo	_				
family member of this in					
based on GenBank accessi	on number; NM_0	0492.3.			
Result					MCR
The following heterozygo	us deletion was	detected:			
Exon: 10					
DNA change: c.1653_1655d					
Amino acid change: delta		(Phe508del))			
Classification: DELETERIO	OUS				
Interpretation					MCR
This alteration is a know	wn deleterious	mutation.			

This result indicates that this individual is a carrier of CF. This interpretation assumes this individual is healthy and not clinically affected with CF.

Since a mutation has been identified, testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner to clarify their risk of having a child with CF.

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

## \*\*\*Performing Site Legend on Last Page of Report\*\*\*

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,CFTRK	05/06/2012	Final
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<sup>\*</sup> Report times for Mayo performed tests are CST/CDT



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Reference Perform
Test Flag Results Unit Value Site\*

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 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?
Reviewed By:

Melody Elizabeth Kimball

Release Date 13 Jul 2012 09:55 MCR

Yes

### \* Performing Site:

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

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SAMPLEREPORT,CFTRK	05/06/2012	Final
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