Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CFPB	SA00043755	45	F	SA00043755
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00043755	**Account Information**			**Report Notes**
Collected 02/23/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/24/2012 10:14				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Cystic Fibrosis Mutation Panel		REPORTED 02/24/2012 09:58			
Specimen		Blood			MCR
Specimen ID		1037883			MCR
Source		Blood draw			MCR
Order Date		24 Feb 2012 09:55			MCR
Reason For Referral					MCR
Not provided. Analyze for mutations in the CFTR gene.					
Result					MCR
None of the listed mutations were detected.					
Interpretation					MCR

Having excluded the listed mutations, this result decreases the likelihood but does not exclude the possibility that this individual is a carrier of or affected with cystic fibrosis (CF). The degree to which this result reduces the patient's risk depends on the ethnic background and family history of the patient. Because this information was not provided, we are unable to provide a revised risk assessment at this time.

If there is no family history of CF, the risk that this individual is a carrier of another CF mutation is listed below.

Ethnicity	Risk	(Detection rate, Carrier Freq)
Northern European	1/267	(91%, 1/25)
Mixed European	1/134	(82%, 1/25)
Southern European	1/115	(79%, 1/25)
Eastern European	1/127	(77%, 1/30)
Ashkenazi Jewish	1/801	(97%, 1/25)
French Canadian	1/267	(91%, 1/25)
African American	1/338	(81%, 1/65)
Hispanic American	1/251	(82%, 1/46)
Asian American*	1/194	(54%, 1/90)

* does not apply to individuals of Japanese ancestry

These calculations were based on the mutation detection rates and population carrier frequencies noted in the chart and assume no family history of CF. Because there is little information available about the carrier frequency and mutation detection rates for individuals of other ethnicities, we are unable to provide a revised risk assessment for ethnicities other than those listed.

If the patient has a family history, contact our laboratory

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for a revised risk assessment. If there is a suspected diagnosis of CF, correlation between other laboratory tests and clinical history is recommended. In addition, other genetic testing strategies such as full gene analysis of CFTR should be considered for identifying mutations that are not detected by this assay. Full gene analysis is clinically available (test code 88876). Contact the Molecular Genetics Laboratory at 1-800-533-1710 for further discussion regarding this option.

A genetic consultation may be of benefit.

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 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.

Method MCR

The multiplex PCR based assay utilizing the Sequenom Mass Array platform is used to detect all 23 mutations specified in the American College of Medical Genetics (ACMG) standards for population based carrier screening (deltaF508, deltaI507, G542X, G85E, R117H, W1282X, 621+1 G>T, 711+1 G>T, N1303K (C>A and C>G), R334W, R347P, A455E, 1717-1 G>A, R553X, R560T, G551D, 1898+1 G>A, 2184delA, 2789+5 G>A, 3120+1 G>A, R1162X, 3659delC, and 3849+10kb C>T). Additionally, the deletion of exons 2-3, 296+2 T>A, E60X, R75X, 394delTT, 405+1 G>A, 406-1 G>A, E92X, 444delA, 457TAT>G, R117C, Y122X, 574delA, 663delT, G178R, 711+5 G>A, 712-1 G>T, H199Y, P205S, L206W, 852del22, 935delA, 936delTA, deltaF311, 1078delT, G330X, T338I, R347H, R352Q, Q359K, T360K, 1288insTA, S466X (C>A), S466X (C>G), G480C, Q493X, 1677delTA, C524X, S549N, S549R, Q552X, A559T, 1811+1.6kb

Performing Site Legend on Last Page of Report

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Printed 02/24/2012 10:14				

Test	Flag	Results	Unit	Reference Value	Perform Site*
A>G, 1812-1 G>A, 1898+1 G>T, 1898+1 G>C, 1898+5 G>T, P574H, 1949del84, 2043delG, 2055del9>A, 2105del13ins5, 2108delA, 2143delT, 2183AA>G, 2184insA, R709X, K710X, 2307insA, R764X, Q890X, 2869insG, 3171delC, 3199del6, R1066C, W1089X, Y1092X (C>G), Y1092X (C>A), M1101K, M1101R, D1152H, R1158X, 3667del4, S1196X, W1204X, 3791delC, Q1238X, 3876delA, S1251N, S1255X, 3905insT, and 4016insT mutations are detected. Poly T determination and confirmatory testing of homozygous results are performed as reflex tests when appropriate.					
Extraction Performed?		Yes			MCR
Reviewed By:		Heather Lynn Owen			MCR
Release Date		24 Feb 2012 09:56			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: