MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| SAMPLEREPORT,CFPB | SA00044014 | 45 | F | SA00044014 |
| **Ordering Phys** | | | | **DOB** 06/10/1966 |
| **Client Order #** SA00044014 | **Account Information** | | | **Report Notes** |
| **Collected** 03/14/2012 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 03/15/2012 16:12 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **Cystic Fibrosis Mutation Panel** | | REPORTED 03/15/2012 15:55 | | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1037988 | | | MCR |
| Order Date | | 15 Mar 2012 13:26 | | | MCR |
| Reason For Referral | | | | | MCR |
| Carrier screen for cystic fibrosis (CF). | | | | | |
| Result | | | | | MCR |
| One copy of the deltaF508 mutation in exon 10 was identified. | | | | | |
| Interpretation | | | | | MCR |
| This result indicates that this individual is a carrier of CF. This interpretation assumes that this individual is healthy and is not clinically affected with CF. | | | | | |
| Since a mutation has been identified, testing of other at risk family members is possible. CF carrier screening should be offered to this individual's reproductive partner if appropriate. A genetic consultation may be of benefit. | | | | | |
| CAUTIONS:<br>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). | | | | | |

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



* Report times for Mayo performed tests are CST/CDT

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| 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 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: | | | | | MCR |
| Matthew John Ferber PhD | | | | | |
| Release Date | | 15 Mar 2012 15:54 | | | MCR |

* Performing Site:

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|---|---|---|
| MCR | Mayo Clinic Dpt of Lab Med & Pathology<br>200 First St SW Rochester, MN 55905 | Lab Director: |