MAYO CLINIC
Mayo Medical Laboratories

TITLE CHANGE/REPORTING CHANGE
NOTIFICATION DATE: July 28, 2010
EFFECTIVE DATE: September 1, 2010

Cystic Fibrosis Mutation Analysis, 70-Mutation Panel #9497

NOTE: Please note the amended CPT codes.

EXPLANATION: The Cystic Fibrosis Mutation Analysis, 70-Mutation Panel, #9497, will undergo a platform change that will move the testing to a 106-mutation panel. The new platform will affect published name, CPT codes and reporting. The list fee and reporting title will not change.

CURRENT PUBLISHED NAME: Cystic Fibrosis Mutation Analysis, 70-Mutation Panel

NEW PUBLISHED NAME: Cystic Fibrosis Mutation Analysis, 106-Mutation Panel

NEW CPT CODES:
"Cystic Fibrosis Mutation Analysis, 106 Mutation Panel"
83788/x8 mass spectrometry
83890/Molecular isolation or extraction
83900/x8 Amplification, target, multiplex, first 2 nucleic acid sequences
83901/x89 Amplification of patient nucleic acid, multiples, each additional nucleic acid sequence
83912/Interpretation and report
83914/x106 Mutation identification by enzymatic ligation or primer extension, single segment, each segment

"Amniotic Fluid Culture for Genetic Testing"
88235/ tissue culture for amniotic fluid (if appropriate)
88240/Cryopreservation (if appropriate)

"Fibroblast Culture for Genetic Testing"
88233/Tissue culture, skin, or solid tissue biopsy (if appropriate)
88240/Cryopreservation (if appropriate)

LIST FEE: $600.60
For amniotic fluid specimens, the following test will be added at an additional charge:
$554.10 for #80334 "Amniotic Fluid Culture for Genetic Testing"
$1,154.70 =Total List Fee

For chorionic villus specimens, the following test will be added at an additional charge:
$246.10 for #80333 "Fibroblast Culture for Genetic Testing"
$846.70 = Total List Fee

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710

MAYO CLINIC
Mayo Medical Laboratories

TEST DEFINITION

7/29/2010

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
9497	3/20/2009		Cystic Fibrosis Mutation Panel			MCR
		Transport temp : Varies				
				Ambient\Refrig OK\Frozen NO - Blood		
				Refrig\Ambient OK\Frozen NO - Amniotic Fluid		
				Ambient\Refrig OK\Frozen NO - Cultured Amniocytes		
				Ambient\Refrig OK\Frozen NO - Cultured Chorionic Villi		
				Refrig\Ambient NO\Frozen NO - Chorionic Villi		
		16422	Specimen			
			- - - - -			
		16423	Specimen ID			
			- - - - -			
		16424	Source			
			- - - - -			
		16425	Order Date			
			- - - - -			
		16426	Reason For Referral			
			- - - - -			
		16428	Result			
			- - - - -			
		16429	Interpretation			
			- - - - -			
		16427	Method			
			- - - - -			
		29378	Extraction Performed?			
			- - - - -			
		16430	Amendment			
			- - - - -			

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
9497						
		16431	Reviewed By:			
			- - - - -			
		16432	Release Date			

*Performing Site Legend

MCR Mayo Clinic Dpt of Lab Med & Pathology LAB DIRECTOR: Franklin R. Cockerill, III, M.D.
200 First Street SW
Rochester, MN 55905

*** End of Report ***

MML Messages used as normals
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LAB

CODE	TEXT

Total of 0 normals codes

*** End of Report ***

MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
VALIDATION, CFPB	L3MRNG9153287	40Y	M	G9153287

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
07/19/10 02:54 P	07/19/10 02:54 P	07/29/10 01:00 P	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

Validation testing

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

Cystic Fibrosis Mutation Panel

Specimen	**Blood**	**MCR**
Specimen ID	**1034521**	**MCR**
Order Date	**21 Jul 2010 10:22**	**MCR**

Reason For Referral **MCR**

Possible diagnosis of Cystic Fibrosis (CF), patient is of Northern European ancestry. Analyze for mutations in the CFTR gene.

Result **MCR**

None of the listed mutations were detected.

Interpretation **MCR**

These results do not provide evidence for a diagnosis of CF. Based on a mutation detection rate of 91% for Northern European ancestry, approximately 1% of classical CF patients would be predicted to have an unidentified mutation on both chromosomes. For atypical cases of CF or for other ethnic and racial groups, the percentage of patients with an unidentified mutation on both chromosomes may be greater than 1%.

Because this patient may have unidentified mutations on both chromosomes, this test does not rule out the diagnosis of CF. Interpretation of test results and risk calculations are dependent upon the definition of each ethnicity as well as the patient's clinical and family history information. Clinical findings and other laboratory tests, such as sweat chloride testing, is recommended to establish a definite diagnosis. If the diagnosis of CF is still suspected, then other gene testing strategies, such as full gene sequencing of CFTR, should be considered for identifying mutations that are not identified by the above assay. Full gene sequencing is clinically available (test code 88876), contact the

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PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
VALIDATION, CFPB	Final	07/19/10 02:54 P

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
VALIDATION, CFPB	L3MRNG9153287	40Y	M	G9153287

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
07/19/10 02:54 P	07/19/10 02:54 P	07/29/10 01:00 P	DATE OF BIRTH:
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202			Validation testing

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

Molecular Genetics Laboratory at 1-800-533-1710 for further discussion regarding this option.
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.

Method MCR

The multiplex PCR-based assay from Luminex 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, 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 394delTT, E60X, R75X, 405+3 A>C, 406 1 G>A , Y122X, 444delA, R117C, G178R, L206W, 935delA, deltaF311, G330X, R352Q,S364P, 1078delT, R347H, V520F, G480C, Q493X, 1677delTA, A559T, S549N, S549R, 1812 1 G>A, 1898+5G>T, 2183AA>G, 2307insA, 2055del9>A, 2143delT, K710X, Q890X, 2869insG, 3120G>A, 3199del6, Y1092XC>G, Y1092XC>A, M1101K, R1066C, W1089X, D1152H, R1158X, 3791delC, S1196X, 3905insT, S1255X, and 3876delA mutations are detected. Poly T determination and confirmatory testing of homozygous

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1-800-533-1710

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VALIDATION, CFPB	L3MRNG9153287	40Y	M	G9153287

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME
07/19/10 02:54 P	07/19/10 02:54 P	07/29/10 01:00 P

SPECIMEN INFORMATION
DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

Validation testing

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
results are performed as reflex tests when appropriate.			
Extraction Performed?	**Yes**		**MCR**
Reviewed By:	**Keri Jane Kruckeberg**		**MCR**
Release Date	**21 Jul 2010 10:56**		**MCR**

*** PERFORMING SITE**

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
VALIDATION, CFPB	Final	07/19/10 02:54 P