

## **MLYCD Gene, Full Gene Analysis**

### **#89830**

**USEFUL FOR:** Confirmation of diagnosis of malonyl-CoA decarboxylase (MCD) deficiency

Carrier screening in cases where there is a family history of MCD deficiency, but disease-causing mutations have not been identified in an affected individual

**METHODOLOGY:** DNA Sequencing and Gene Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA) of MLYCD Gene and Cell Culture

**REFERENCE VALUES:** An interpretive report will be provided.

**SPECIMEN REQUIREMENTS: Please submit only 1 of the following specimens:**

- **Blood** Draw blood in a lavender-top (EDTA) tube or a yellow-top (ACD) tube, and send 3 mL of EDTA or ACD whole blood in Original VACUTAINER. Invert several times to mix blood. Forward unprocessed whole blood promptly at ambient temperature.
- **Cultured Fibroblasts** 1 T-75 or 2 T-25 flask(s) filled to neck with culture media. **(Specimen received in formalin or fixative preservative is not acceptable.)** Maintain sterility and forward promptly at ambient temperature.
- **Skin Biopsy** Collect sterile skin biopsy (4 mm punch) in a sterile, screw-capped container filled with any standard cell culture media (e.g. minimal essential media, RPMI 1640, etc.). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]). **(Specimen received in formalin or fixative preservative is not acceptable.) Specimen cannot be frozen.** Maintain sterility and forward promptly.

**NOTE:**

- **Specimen must arrive within 96 hours of collection.**
- **"Molecular Genetics-Biochemical Disorders Patient Information Sheet" (Supply T527 or see Special Instructions) is required** for all orders. If not ordering electronically, please submit the above information sheet along with a "Molecular Genetics Request Form" (Supply T245) with the specimen. An "Informed Consent for DNA Testing" (Supply T576) is available. See Special Instructions for a copy of the form.

**CAUTIONS:**

- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- Medical genetic consultation should be considered in complex cases or when the clinical presentation is atypical or uncertain.

- In occasional cases, DNA alterations of undetermined significance may be identified. Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical and biochemical findings, additional testing should be considered. A list of known polymorphisms is available upon request.
- 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.

**LIST FEE:** \$800.00

**CPT CODE:**

"MLYCD Gene, Full Gene Analysis"

83891/Isolation or extraction of highly purified nucleic acid

83892/x5 Enzymatic digestion

83894/x5 Separation by gel electrophoresis

83898/x3 Amplification, target, each nucleic acid sequence

83900/x2 Amplification, target, multiplex, first 2 nucleic acid sequences

83909/x14 Separation and identification by high-resolution technique

83912/Interpretation and report

"Gene Dosage Analysis"

83900/Amplification, target, multiplex, first 2 nucleic acid sequences

83909/Separation and identification by high-resolution technique

83914/x11 Mutation identification by enzymatic ligation or primer extension, single segment, each segment

"Fibroblast Culture for Genetic Testing"

88233/Tissue Culture, skin or solid tissue biopsy (if appropriate)

88240/Cyropreservation (if appropriate)

**ANALYTIC TIME:** 7 days

**DAY(S) SET-UP:** Thursday

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

## TEST DEFINITION

11/2/2009

Code Name  
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89830 MLYCD Gene, Full Gene Analysis

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
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89830	10/8/2009		MLYCD Gene, Full Gene Analysis			MCR
			Transport temp : Ambient\Refrig OK\Frozen NO-Blood			
			Ambient\Refrig <24 hours OK\Frozen NO-Cultured Fibroblasts			
			Refrig\Ambient OK\Frozen NO-Skin Biopsy			
		50902	Specimen			
		50903	Specimen ID			
		50904	Source			
		50905	Order Date			
		50906	Reason For Referral			
		50907	Method			
		50908	Result			
		50909	Interpretation			
		50935	Extraction Performed?			
			Possible result values include : No,Yes,No,Yes			
		50936	MLPA Performed?			
			Possible result values include : No,Yes,No,Yes			
		50910	Amendment			

(continued next page)

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
89830	(continued)					
		50911	Reviewed By			
		50912	Release Date			

\*Performing Site Legend

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MCR      Mayo Clinic Dpt of Lab Med & Pathology      LAB DIRECTOR:  Franklin R. Cockerill, III, M.D.
          200 First Street SW
          Rochester, MN 55905
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CODE            TEXT  
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Total of 0 normals codes  
  
\*\*\* End of Report \*\*\*

1-800-533-1710

<b>PATIENT NAME</b> TESTING, MCDMSDB		<b>PATIENT NUMBER</b>		<b>AGE</b> 33	<b>SEX</b> F	<b>ACCESSION #</b> G9137060
<b>ORDERING PHYSICIAN</b>		<b>CLIENT ORDER #</b>			<b>ACCOUNT #</b> LIAISONS	
<b>COLLECTION</b> 10/09/09 02:02 P <b>DATE TIME</b>	<b>RECEIVED</b> 10/09/09 02:02 P <b>DATE TIME</b>	<b>REPORT PRINTED</b> 11/02/09 03:01 P <b>DATE TIME</b>		<b>SPECIMEN INFORMATION</b> DATE OF BIRTH:		
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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**MLYCD Gene, Full Gene Analysis**

<b>Specimen</b>	Blood	MCR
<b>Specimen ID</b>	1033391	MCR
<b>Order Date</b>	16 Oct 2009 13:18	MCR
<b>Reason For Referral</b>		MCR

Patient reported to have features suggestive of malonyl-coenzyme A decarboxylase (MCD) deficiency. Test for the presence of mutations in the MLYCD gene.

<b>Method</b>		MCR
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DNA sequence analysis was used to test for the presence of a mutation in all 5 exons of the MLYCD gene. Additionally, gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in this gene. GenBank accession number NM\_012213.2 is utilized in reporting all alterations.

<b>Result</b>		MCR
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A mutation was NOT identified

<b>Interpretation</b>		MCR
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These results decrease the likelihood, but do not rule out the diagnosis of MCD deficiency for this individual. We predict that some individuals who have a diagnosis of MCD deficiency may have disease causing mutations in the MLYCD gene not identified by the method described above (e.g. mutations in the promoter and intronic regions).

This assay does not rule out the presence of disease causing mutations within other genes that are associated with metabolic disease. These results should be interpreted in light of clinical findings, family history, and other laboratory data.

A list of common polymorphisms identified for this patient is available upon request.

\* Perform Site Legend on last page of report

<b>PATIENT NAME</b> TESTING, MCDMSDB	<b>ORDER STATUS</b> Final	<b>COLLECTION DATE AND TIME</b> 10/09/09 02:02 P
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1-800-533-1710

<b>PATIENT NAME</b> TESTING, MCDMSDB		<b>PATIENT NUMBER</b>		<b>AGE</b> 33	<b>SEX</b> F	<b>ACCESSION #</b> G9137060
<b>ORDERING PHYSICIAN</b>		<b>CLIENT ORDER #</b>				<b>ACCOUNT #</b> LIAISONS
<b>COLLECTION</b> 10/09/09 02:02 P	<b>RECEIVED</b> 10/09/09 02:02 P	<b>REPORT PRINTED</b> 11/02/09 03:01 P		<b>SPECIMEN INFORMATION</b>		
<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>	<b>DATE</b> <b>TIME</b>	<b>DATE OF BIRTH:</b>			
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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**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.

Extraction Performed?	Yes	MCR
MLPA Performed?	Yes.	MCR
See #89861, MLYCD Large Deletion/Duplication, MLPA, for billing information.		
Reviewed By	Keri Jane Kruckeberg	MCR
Release Date	16 Oct 2009 13:30	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.
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<b>PATIENT NAME</b> TESTING, MCDMSDB	<b>ORDER STATUS</b> Final	<b>COLLECTION DATE AND TIME</b> 10/09/09 02:02 P
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