

MLYCD Gene, Known Mutation #89831

USEFUL FOR:

- Diagnostic confirmation of malonyl-coenzyme A decarboxylase (MCD) deficiency when familial mutations have been previously identified
- Carrier screening of at-risk individuals when a mutation in the *MLYCD* gene has been identified in an affected family member

METHODOLOGY: Polymerase Chain Reaction (PCR)/DNA sequencing and/or gene dosage analysis by multiplex ligation-dependent probe amplification (MLPA) of *MLYCD* gene is utilized for mutations previously identified in an affected family member 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.
- **Amniotic Fluid** Obtain 20 mL of amniotic fluid. Transfer specimen to 2 screw-capped, sterile centrifuge tubes. Send specimen refrigerated. **Specimen cannot be frozen.** A separate culture charge will be assessed under test #80334 "Amniotic Fluid Culture for Genetic Testing." Alternatively, we will accept 2 T-25 flasks of confluent cultured cells from another laboratory sent at ambient temperature.
- **Chorionic Villi** Obtain 20 mg of chorionic villus specimen. Send specimen refrigerated in transport media in 15-mL centrifuge tube. **Specimen cannot be frozen.** A separate culture charge will be assessed under test #80333 "Fibroblast Culture for Genetic Testing." Alternatively, we will accept 2 T-25 flasks of confluent cultured cells from another laboratory sent 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:

- Identification of a mutation in an affected family member is required before this testing will be performed.
- 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.
- 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.
- 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: \$150.00**CPT CODE:**

"MLYCD Gene, Known Mutation"

83891/Isolation or extraction of highly purified nucleic acid

83912/Interpretation and report

"DNA Sequence, Follow-up Analysis"

83892/Enzymatic digestion (if appropriate)

83894/Separation by gel electrophoresis (if appropriate)

83898/Amplification, target, each nucleic acid sequence (if appropriate)

83909/x2 Separation and identification by high-resolution technique (if appropriate)

"MLYCD Gene, Large Deletion/Duplication"

83900/Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)

83909/Separation and identification by high-resolution technique (if appropriate)

83914/x11 Mutation Identification by enzymatic ligation or primer extension, single segment, each segment (if appropriate)

"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)

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

89831 MLYCD Gene, Known Mutation

| ORDER CODE | EFF DATE | TC | TITLE | Checking Normals | Print normals (# coded) | Perform Site * |
|------------|-----------|-------|----------------------------------|--------------------------------------------------------------|-------------------------|----------------|
| 89831 | 10/9/2009 | | MLYCD Gene, Known Mutation | | | MCR |
| | | | Transport temp : | Ambient\Refrig OK\Frozen NO - Blood | | |
| | | | | Refrig\Ambient NO\Frozen NO - Amniotic Fluid, Skin Biopsy | | |
| | | | | Ambient\Refrig <24 hours OK\Frozen NO - Cultured Fibroblasts | | |
| | | | | Ambient\Refrig OK\Frozen NO - Cultured Amniocytes | | |
| | | | | Ambient\Refrig OK\Frozen NO - Cultured Chorionic Villi | | |
| | | | | Refrig\Ambient NO\Frozen NO - Chorionic Villi | | |
| | | 50913 | Specimen | | | |
| | | 50914 | Specimen ID | | | |
| | | 50915 | Source | | | |
| | | 50916 | Order Date | | | |
| | | 50917 | Reason For Referral | | | |
| | | 50918 | Method | | | |
| | | 50919 | Result | | | |
| | | 50920 | Interpretation | | | |
| | | 50937 | Extraction Performed? | | | |
| | | | Possible result values include : | No, Yes, No, Yes | | |
| | | 50921 | Amendment | | | |

(continued next page)

| ORDER CODE | EFF DATE | TC | TITLE | Checking Normals | Print normals (# coded) | Perform Site * |
|---------------|-------------|-------|--------------|------------------|-------------------------|-------------------|
| 89831 | (continued) | | | | | |
| | | 50922 | Reviewed By | | | |
| | | 50923 | 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

Total of 0 normals codes

*** End of Report ***

1-800-533-1710

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|--------------------------------------------------------------------------------------------------|--------------------------------------------------------------|-------------------------------------------------------------------------|--|-----------------------------------------------|-----------------|--------------------------------|
| PATIENT NAME TESTING, MCDKMLD | | PATIENT NUMBER | | AGE 33 | SEX F | ACCESSION # G9137061 |
| ORDERING PHYSICIAN | | CLIENT ORDER # | | | | ACCOUNT # LIAISONS |
| COLLECTION 10/09/09 02:03 P DATE TIME | RECEIVED 10/09/09 02:03 P DATE TIME | REPORT PRINTED 11/02/09 03:01 P DATE TIME | | SPECIMEN INFORMATION 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, Known Mutation

| | | |
|----------------------------|-------------------|-----|
| Specimen | Blood | MCR |
| Specimen ID | 1033392 | MCR |
| Order Date | 16 Oct 2009 13:23 | MCR |
| Reason For Referral | | MCR |

Family history of malonyl-coenzyme A decarboxylase (MCD) deficiency. Test for the presence of mutations in the MLYCD gene.

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|---------------|--|-----|
| Method | | MCR |
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DNA sequence analysis was used to test for the presence of the p.X (c.X) and p.X (c.X) alterations in exons X and X, respectively, of the MLYCD gene (GenBank accession number; NM_012213.2). Analysis for these specific alterations was performed because they have been identified in a family member.

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| Result | | MCR |
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The p.X alteration was NOT detected.

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| Interpretation | | MCR |
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Absence of the mutation(s) previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population for developing symptoms related to MCD deficiency.

This assay does not rule out the presence of other disease causing mutations within this gene or other genes that are associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of test results.

A genetic consultation may be of benefit.

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

CAUTIONS: Rare polymorphisms exist that could lead to false

* Perform Site Legend on last page of report

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|-----------------------------------------|------------------------------|-----------------------------------------------------|
| PATIENT NAME TESTING, MCDKMLD | ORDER STATUS Final | COLLECTION DATE AND TIME 10/09/09 02:03 P |
|-----------------------------------------|------------------------------|-----------------------------------------------------|

1-800-533-1710

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|--------------------------------------------------------------------------------------------------|---------------------------------------------------------|---------------------------------------------------------------|--|-----------------------------------------------|-----------------|--------------------------------|
| PATIENT NAME TESTING, MCDKMLD | | PATIENT NUMBER | | AGE 33 | SEX F | ACCESSION # G9137061 |
| ORDERING PHYSICIAN | | CLIENT ORDER # | | | | ACCOUNT # LIAISONS |
| COLLECTION 10/09/09 02:03 P DATE TIME | RECEIVED 10/09/09 02:03 P DATE TIME | REPORT PRINTED 11/02/09 03:01 P DATE TIME | | SPECIMEN INFORMATION 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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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.

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|-----------------------|----------------------|-----|
| Extraction Performed? | Yes | MCR |
| Reviewed By | Keri Jane Kruckeberg | MCR |
| Release Date | 16 Oct 2009 13:28 | MCR |

MLYCD Gene, Large Del/Dup

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|-------------|-------------------|-----|
| Specimen | Blood | MCR |
| Specimen ID | 1033392 | MCR |
| Order Date | 16 Oct 2009 13:23 | MCR |
| Result | | MCR |

Analysis has been completed. Refer to the mcdkm for results and interpretation.

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| Reviewed By | Keri Jane Kruckeberg | MCR |
| Release Date | 16 Oct 2009 13:29 | MCR |

* PERFORMING SITE

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| 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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|-----------------------------------------|------------------------------|-----------------------------------------------------|
| PATIENT NAME TESTING, MCDKMLD | ORDER STATUS Final | COLLECTION DATE AND TIME 10/09/09 02:03 P |
|-----------------------------------------|------------------------------|-----------------------------------------------------|