

NEW TEST/OBSOLETE TEST ANNOUNCEMENT

NOTIFICATION DATE: September 19, 2012 **EFFECTIVE DATE:** October 23, 2012

Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Mutation Screen

Test ID: MCADS

USEFUL FOR:

- Confirmation of diagnosis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency (as a follow up to biochemical analyses)
- Screening of at-risk carriers of MCAD deficiency when an affected relative has not had molecular testing
- Diagnosis of MCAD deficiency in autopsy specimens

METHODOLOGY: Polymerase Chain Reaction (PCR) Amplification/DNA sequencing

REFERENCE VALUES: An interpretive report will be provided

SPECIMEN REQUIREMENTS: Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Blood **Container/Tube:**

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions:

Invert several times to mix blood.
Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated/Frozen

Specimen Type: Cultured fibroblasts **Container/Tube:** T-75 or T-25 flask

Specimen Volume: 1 full T-75 or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

- 1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours
- 2. Do not expose specimen to heat or direct sunlight.
- 3. Do not stack wet specimens.
- 4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

NOTE:

- Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions.
- New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Varies	

CAUTIONS:

- A small percentage of individuals who are carriers or have a diagnosis of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency may have a mutation that is not identified by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of MCAD. For carrier testing, it is important to first document the presence of an *ACADM* gene mutation in an affected family member.
- In some 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 findings, additional testing should be considered.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
- 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.

FEE: Please contact your Regional Manager for your account's fee information.

CPT CODE:

Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Mutation Screen 83890- Molecular isolation or extraction 83898 x 12-Amplification, target, each nucleic acid sequence 83909 x 22-Separation and identification by high-resolution technique 83912-Interpretation and report

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

DAY(S) SET UP: Wednesday 10am **ANALYTIC TIME:** 10 days

NOTE: The following Mayo test code will become obsolete.

Test Name	Test ID
Medium-Chain Acyl-CoA Dehydrogenase	MCADP
(MCAD) Deficiency Molecular Profile	

NOTE: The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
ACADM Gene analysis in	ZW168	2682	GeneDx, Inc.
Medium Chain Acyl -			
CoA Dehydrogenase			
(MCAD) Deficiency -			
Full Sequencing			