**Disorder:** MCAD deficiency is the most common disorder of fatty acid oxidation affecting 1 in 13,000 newborns and is inherited as an autosomal recessive disorder. This enzyme deficiency results in the inability to catabolize medium-chain (6-12 carbon molecules) fatty acids for energy utilization. MCAD deficiency often presents in the first two years of life after viral illness or fasting. This inability to break down medium-chain lipids as an energy source during times of fasting and metabolic stress can result in hypoglycemia associated with vomiting, lethargy, apnea, coma, encephalopathy and sudden death.

**Indications:**
- Unexplained low blood sugars and metabolic acidosis in an infant
- Abnormal newborn screen suggesting MCAD deficiency
- Abnormal acylcarnitine profile consistent with MCAD deficiency
- Abnormal organic acid profile consistent with MCAD deficiency
- Presymptomatic testing of at-risk siblings
- Prenatal diagnosis of an at-risk fetus, after confirmation of biallelic mutations in the parents (by prior arrangement only)
- Carrier testing in relative of a patient with MCAD deficiency

Additional information and test requisitions are available at: www.cchmc.org/molecular-genetics

**Shipping Instructions**
Please enclose test requisition with sample. All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

**Ship to:**
Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

Molecular Genetics Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: moleculargenetics@cchmc.org
Specimen: At least 3 mLs whole blood in purple/lavender top (EDTA) tube. Label tube with patient’s name, birth date, and date of collection. Phlebotomist must initial tube to verify patient’s identity.

Testing Methodology:
• Genotyping for K329E allele only
  OR
• PCR-based sequencing of all 12 exons and exon/intron boundaries of the ACADM gene.

Test Sensitivity:
• Genotyping for K329E allele: This test detects only the K329E allele which accounts for 80-90% of disease causing mutations in ACADM. Heterozygous and normal test results may be reflexed to full gene sequencing. An additional charge will apply.
• PCR-based sequencing detects >95% of patients with MCAD deficiency. The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed. Multiple exon deletions and insertions may not be identified by this methodology. ACADM is the only gene associated with MCAD deficiency.
MetaboSeq fatty acid oxidation defects panel detects mutations in ACADM as well as 18 other genes involved with FAOD. Please see our website for details.

Turn-Around Time:
• Genotyping for K329E allele: two business days
• ACADM full gene sequence analysis: 28 days

Cost: Please call 1-866-450-4198 for current pricing, assistance with precertification or with any billing questions.

CPT Codes:
• Genotyping for K329E allele: 81400
• ACADM full gene sequence analysis: 81479
• Family specific mutation analysis: 81403

Results: Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

References: