



IMPORTANT MCAD FOLLOW-UP INFORMATION

As of April 1, 2003, all Michigan newborns will be tested for Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency (MCAD), in addition to the other seven disorders. **The current procedure for blood collection between 24-36 hours of age onto filter paper will not change.**

MCAD deficiency is an autosomal recessive inherited fatty acid oxidation disorder (FOD) in which an enzyme defect in the fatty acid metabolic pathway inhibits the body's ability to utilize stored fat. Symptoms include vomiting and lethargy. Hypoglycemia may occur especially under conditions of fasting or intercurrent illness (gastrointestinal or upper respiratory) and can lead to coma, encephalopathy, liver failure or death. Infants appear normal at birth and typically present with symptoms between 3 and 24 months of age, however, later presentation (even into adulthood) is possible.

Early diagnosis and treatment is essential. If left untreated, MCAD deficiency may result in significant disability and ultimately death. Effective treatment for MCAD deficiency includes a low-fat/high-carbohydrate diet and supplemental carnitine. Frequent feedings are instituted to avoid fasting. Acute episodes (during illness) may require aggressive medical management, especially if the infant/child is vomiting or is not receiving adequate nutritional intake. The administration of intravenous glucose and blood sugar monitoring is essential. Children with MCAD deficiency are at risk for speech and development problems if they experience significant hypoglycemia and metabolic decompensation. With early diagnosis and education, these episodes can be avoided and the outcome is excellent.

Newborn screening for MCAD deficiency will be performed using tandem mass spectrometry to detect elevated acylcarnitine levels. False positive and false negative results are possible with this screening. Infants with positive screening results will require a prompt (within 24 hours) repeat newborn screen sent to the MDCH Newborn Screening Laboratory. The specimen should be collected just prior to a feeding if possible. In Addition, positive screening results will be immediately referred to the Pediatric Neurology Metabolic Clinic (PNMC) at the University of Michigan (734) 763-4697 for confirmatory testing and follow up care. When notified of any positive result for MCAD deficiency, the primary care physician should complete an assessment of the infant's clinical status. The parents should be instructed to avoid any significant time gap in feedings (greater than 4 hours in newborns).