Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) Screening
Fact Sheet for Health Care Providers

Newborn Screening Program of the Oklahoma State Department of Health

What are the characteristics of MCAD?

- Autosomal recessive genetic condition.
- Most infants are born to parents who are both unknowingly asymptomatic MCAD carriers and have NO known history of MCAD in the family.
- MCAD has a frequency of 1/15,000 to 1/20,000 live births.
- This disorder can cause metabolic crisis, usually presenting with hypoglycemia, in infants and children during periods of poor feeding, fasting or illness. This crisis can lead to seizures, respiratory failure, cardiac arrest and death. Crisis survivors may experience significant developmental disabilities.
- Treatment involves a special diet, frequent feedings, and special care during times of illness or stress.

What is the screening methodology for Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)?

1. An acylcarnitine profile by Tandem Mass Spectrometry (MS/MS) is performed on each filter paper.
2. C8 is the primary analyte.
3. If C8 is elevated, the following analytes and ratio are assessed: C6, C10, C10:1 & C8/C10 ratio.

What is an in-range (normal) screen result for MCAD?
C8 less than 0.40 µmol/L is NOT consistent with MCAD. See Table 1.

What is an out-of-range (abnormal) screen for MCAD? C8 ≥ 0.40 µmol/L requires further testing.

What screen results will require a repeat filter paper? C8 ≥ 0.40 µmol/L with a C8/C10 Ratio < 3.0 requires a repeat filter paper. Initiation of feeding precautions will be left to provider discretion.

What screen results will require diagnostic testing?
C8 ≥ 0.40 µmol/L with a C8/C10 Ratio ≥ 3.0 will require immediate action. The follow-up program will provide detailed guidance on required actions and an *Emergency Management Protocol* will be provided.

TABLE 1.
In-range MCAD Newborn Screening Results

Primary Analyte	In-Range (µmol/L)
C8	< 0.40
Secondary Analytes[1]	**In-Range**
C8/C10 ratio	< 3.00
C6	< 0.25 µmol/L
C10	< 0.40 µmol/L
C10:1	< 0.30 µmol/L

[1] Elevations of the secondary analytes are reported as "not consistent with MCAD" if C8 is in-range.

What are the follow-up needs?
The follow-up program will provide detailed guidance on needed actions. The following metabolic specialists have approved all recommendations:

Integris Pediatric Specialty Clinic, Inborn Error of Metabolism (IEM) Clinic
Geneticist pager: (405) 630-3794

OU Children's Physicians – Genetics Clinic
Page Operator: (405) 271-3636

What is my role in screening?
If you are listed as the infant's planned health care provider on the filter paper requisition, you are required by the *Newborn Screening Program Regulations* to initiate follow-up activities.

Newborn Screening Program (405) 271-6617 opt 2 or 1-800-766-2223 opt 2 Metabolic
Nurse Specialist (405) 271-8001, ext. 42074
http://nsp.health.ok.gov