Fatty Acid Oxidation Disorder (FAOD) Screening Fact Sheet for Health Care Providers
Newborn Screening Program of the Oklahoma State Department of Health

What are the characteristics of FAOD?

- Autosomal recessive genetic conditions. Most infants are born to parents who are both unknowingly asymptomatic carriers and have NO known history of a fatty acid oxidation disorder in the family.
- The incidence of FAOD's ranges from 1:10,000 to 1:100,000. MCAD is one of the most common with an incidence of 1:10,000. VLCAD and LCHAD are more rare.
- Symptoms vary by disorder. These disorders can lead to metabolic crisis, especially in infants and children. This crisis can lead to seizures, respiratory failure, cardiac arrest and death. Crisis survivors may experience significant developmental disabilities. Some infants will present during the neonatal period with life threatening symptoms.
- Treatment involves a special diet managed by a metabolic specialist and a metabolic dietician, frequent feedings, and special care during times of illness or stress.

What is the screening methodology for FAOD disorders?

An acylcarnitine profile by Tandem Mass Spectrometry (MS/MS) is performed on each filter paper.
Primary and secondary analytes are simultaneously tested on each filter paper.
Primary analyte results are used to establish results requiring follow-up. All out-of-range primary analyte results require follow-up.
Secondary analyte are used in conjunction with primary analyte results to assign risk.
Elevations of the secondary analytes are reported as "not consistent with FAOD" if the primary analyte 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.

Summary of out of range FAO Analytes and follow-up recommendations.

Primary Marker (μmol/L)	Secondary Analyte (μmol/L)	Potential Disorder	Risk*	Recommendation
C0 > 2.00 & ≤ 5.5	NA	CUD	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C0 ≤ 2.00	NA	CUD	HR	EMERGENCY. Immediate referral to the metabolic specialist is required.
C4 ≥ 1.27	C4/C2 < 0.06	SCAD & GAII	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C4 ≥ 1.27	C4/C2 ≥ 0.06	SCAD & GAII	HR	Immediate referral to the metabolic specialist is required.
C4 ≥ 2.00	NA	SCAD & GAII	HR	Immediate referral to the metabolic specialist is required.
C8 ≥ 0.40	C8/C10 < 3.0	MCAD & MCAT	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C8 ≥ 0.40	C8/C10 ≥ 3.0	MCAD & MCAT	HR	Immediate referral to the metabolic specialist is required.
C14:1 ≥ 0.70	NA	VLCAD	HR	Immediate referral to the metabolic specialist is required.
C16 ≥ 7.46	C18:1 < 3.0	CACT & CPTII	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C18:1 ≥ 3.0	C16 < 7.46	CACT & CPTII	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C18:1 ≥ 3.0	C16 ≥ 7.46	CACT & CPTII	HR	Immediate referral to the metabolic specialist is required.
C0 ≥ 64	C0/(C16+C18)< 90	CPT1	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C0 ≥ 64	C0/(C16+C18)≥ 90	CPT1	HR	Immediate referral to the metabolic specialist is required.
C16OH ≥ 0.16	C18:1 OH < 0.15	LCHAD/TFP	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C18:1 OH ≥ 0.15	C16OH < 0.16	LCHAD/TFP	LR	Repeat filter paper. Consult with metabolic specialist as needed.
C18:1 OH ≥ 0.15	C16OH ≥ 0.16	LCHAD/TFP	HR	Immediate referral to the metabolic specialist is required.

Cut off values may change. Please contact the OK NBS Program for clarification if needed. More information is available on the OSDH website or call (405) 271-6617 opt 2.