NEBRASKA

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DEPT. OF HEALTH AND HUMAN SERVICES

MCAD Information

Medium Chain Acyl-CoA Dehydrogenase Deficiency is an autosomal recessive inherited disorder of fatty acid oxidation metabolism characterised by a defect in the oxidation of medium chain fatty acids. Metabolic crises can occur during periods of fasting and can cause episodes of hypoglycemia, vomiting and coma and potentially death. The phenotype is clinically identifiable in less than 25% of births. Approximately 20-25% of patients with unidentified MCAD have died as a result of the first metabolic crisis. (50% of older patients). It can present in the neonatal period, infancy, early childhood or later. It is a chronic disease with a life-long risk for episodes of hypoketotic hypoglycemia, often triggered by illness or prolonged fasting. Significant disability can result from severe, prolonged hyoglycemia episodes. Overall, MCAD occurs in about 1 in 15,000 US live births.

Clinical Features:

With appropriate treatment, the risk of any of these conditions is substantially reduced. The onset of symptoms typically occur between 6-24 months of age, but ranges from neonatal to adult. Recurrent episodes of hypoglycemia, vomiting, coma, sudden death and possible seizures. Hepatomegaly is usually present. Metabolic episodes or "crises" can cause significant developmental and physical delays.

Laboratory Test:

The laboratory test done to detect MCAD is analysis of acylcarnitines using tandem mass spectrometry to look concentrations and relative ratios of of octanoylcarnitine (C8), decanoylcarnitine (C10) and palmitoylcarnitine (C16).

TEST RESULTS	LIKELY CAUSES	ACTIONS TO TAKE WHEN POSITIVE
Substantially elevated C:8, May also have: Elevated C10 and C:8 to C:16 ratio elevated. *	MCAD or MADD (if multiple elevations) False positive	Laboratory immediately notifies via phone and fax (followed in writing) submitter, newborn's physician and NNSP. Newborn's physician should advise parent's to avoid fasting the newborn more than 4 hours, and obtain a repeat filter paper, and urine organic acid analysis from the baby. Consultation or referral to a Metabolic specialist is advised.

C8 slightly above normal or above normal	MCAD False positive	Laboratory immediately notifies via phone and fax (followed in writing) submitter, newborn's physician and NNSP. Newborn's physician should advise parents to avoid fasting the newborn more than 4 hours, and obtain a repeat filter paper specimen. Consultation with a metabolic specialist is advised.

Confirmation and Treatment:

Consultation with a pediatric metabolic specialist should be made for confirmation and diagnosis (See ACT Sheet). Upon notification of a positive screening result the physician should collect or cause to be collected a repeat filter paper specimen to be sent to the newborn screening laboratory, and a urine specimen for analysis of urine organic acids. (If notified of an inconclusive result or slight elevation, only a repeat specimen is indicated initially). Specific diagnosis is made by analysis of plasma acylacarnitines (and urine acylglycines) which reveal increased octanoylcarnitine (and increased octanoylglycine). It is strongly advised to consult with a pediatrician specializing in metabolic disorders. Confirmatorty testing following carnitine load may be appropriate.

MCAD has variable presentation. The newborn may be asymptomatic. However the clinical phenotype can rapidly progress to hypoketotic hypoglycemia causing lethargy, vomiting and sudden death. Parents should be advised to avoid fasting the newborn more than 4 hours, (which is not unusual with frequent feedings the first few weeks of life, but should be assured until diagnosis can be confirmed or ruled out). A metabolic specialist may recommend a carnitine supplement, following consultation or referral.