Alberta Health Services
Alberta Newborn Screening

Talking with Parents > When talking to a parent whose baby has received an abnormal screen result, let them know that an abnormal screen result doesn't mean that the infant has the condition. It means that they have a higher chance of having the condition, and they need more tests to find out for sure. A printable sheet for parents with more condition specific information can be used to support your conversation, visit ahs.ca/nms and under Quick Reference click on *What conditions are screened for?*

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

(metabolic condition: fatty acid oxidation disorder) *Information for Health Professionals*

Also known as:

- medium-chain acyl-coenzyme A dehydrogenase deficiency
- ACADM deficiency

What are fatty acid oxidation disorders?

Fatty acid oxidation disorders (FAOD) are a group of inherited conditions in which fatty acids cannot be broken down to provide the body with energy. Fatty acids are a key source of energy during fasting or stress, such as during an illness. An inability to metabolize fatty acids leads to an accumulation of toxic metabolites in the body along with cellular energy deficiency which in combination can cause serious health problems.

What is MCAD deficiency?

Medium chain acyl-CoA dehydrogenase (MCAD) is one of the mitochondrial enzymes required in the breakdown of fatty acids to produce energy. MCAD breaks down fatty acids between 6 and 12 carbon atoms in length. People with MCAD deficiency are unable to break down these medium chain fatty acids to produce energy.

What causes MCAD deficiency?

MCAD deficiency results from pathogenic variants in the MCAD gene resulting in absent or decreased activity of the enzyme.

How common is MCAD deficiency?

The incidence of MCAD deficiency is about 1 in every 12,000 infants born in Canada.

What are the clinical features of MCAD deficiency?

Although infants with MCAD deficiency may appear normal at birth, during a period of fasting (such as during an illness), an infant who was previously healthy may present with lethargy, vomiting, hypoketotic hypoglycemia and seizures. While the first episode usually presents between 2 months and 2 years of age, it is possible that clinical features may present at any age. Coma and death may occur if treatment is not initiated quickly. These children may also have an enlarged liver and acute liver disease.

What is the screening test for MCAD deficiency?

A specific pattern of fatty acid metabolites is detected on the newborn blood spot screen. Newborn blood spot screening will not detect all infants with MCAD. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of MCAD deficiency is confirmed by detecting specific metabolites in urine or on blood acylcarnitine analysis. Further testing may include enzyme analysis and/or molecular genetic analysis of the MCAD gene. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is MCAD deficiency treated?

MCAD deficiency is effectively treated by avoidance of fasting. A low fat diet and carnitine supplementation may be considered. Prompt treatment of a metabolic crisis with intravenous fluids and glucose is necessary. The treatment is lifelong.

Is MCAD deficiency inherited?

MCAD deficiency is inherited as an autosomal recessive disorder. Parents of a child with MCAD deficiency are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. MCAD deficiency carriers are healthy. Genetic counselling is available to families with MCAD deficiency.

Additional resources are available through:

Clinical & Metabolic Genetics Program (Edmonton)

8-53 Medical Sciences Building
8440 – 112 St. NW
Edmonton, AB T6G 2H7
Phone: 780-407-7333
Fax: 780-407-6845

Emergency consultations:
Phone 780-407-8822 and ask for the specialist on call for metabolic diseases.

Clinical & Metabolic Genetics Program (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7587
Fax: 403-955-3091

Emergency consultations:
Phone 403-955-7211 and ask for the specialist on call for metabolic diseases.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening