Alberta Health Services

Newborn Metabolic Screening

Information for Health Professionals

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency (metabolic condition: fatty acid oxidation disorder)

Also known as:

- long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency
- long-chain 3-OH acyl-CoA dehydrogenase deficiency
- type 1 trifunctional protein 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 LCHAD deficiency?

Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) is one of the mitochondrial enzymes required in the breakdown of fatty acids to produce energy. LCHAD breaks down fatty acids and hydroxy fatty acids between 12 and 18 carbon atoms in length. People with LCHAD deficiency are unable to break down these long chain fatty acids to produce energy.

What causes LCHAD deficiency?

LCHAD deficiency results from mutations in one part of the tri-functional protein (TFP) gene resulting in absent or decreased activity of the enzyme. TFP is a protein with several metabolic functions in fatty acid breakdown.

How common is LCHAD deficiency?

LCHAD deficiency is a rare condition; the incidence is unknown.

What are the clinical features of LCHAD deficiency?

Although children with LCHAD deficiency may appear normal at birth, during a period of fasting (such as during a common illness), a child who was previously healthy may present with lethargy, vomiting or diarrhea, hypoketotic hypoglycemia, hypotonia, and seizures. This can progress quickly to coma and death. These children may also have developmental delay, poor weight gain, neurological problems, liver disease and cardiomyopathy. The first episode usually occurs in infancy or early childhood.

What is the screening test for LCHAD deficiency?

A specific pattern of fatty acid metabolites is detected on the newborn blood spot screen. Newborn blood spot screening is not 100% perfect. 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 LCHAD deficiency is confirmed by detecting specific metabolites in urine or on blood acylcarnitine analysis. Further testing may include enzyme analysis and/or mutation analysis of the LCHAD gene. In order to distinguish LCHAD deficiency from tri-functional protein (TFP) deficiency, further diagnostic testing is required. Specialists at the clinics listed below will arrange diagnostic testing.

How is LCHAD deficiency treated?

LCHAD deficiency is treated by avoidance of fasting. Carnitine supplementation together with a diet which is low in long chain fatty acids may be considered. Parent education is an important component of treatment, including advising parents on actions to be taken when their child develops a minor illness. Prompt treatment of a metabolic crisis with intravenous fluids and glucose is necessary to prevent neurological problems. The treatment is lifelong.

What is the outcome of treatment for LCHAD deficiency?

Treatment can be effective in preventing metabolic crises and their sequelae.

Is LCHAD deficiency inherited?

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

For additional resources, please call:

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, detection and treatment – every infant, every time

For more information about the NMS Program, visit www.ahs.ca/newbornscreening

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