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?*

Maple Syrup Urine Disease (MSUD)

(metabolic condition: amino acid disorder)

Information for Health Professionals

Also known as:

- branched-chain alpha-keto acid dehydrogenase deficiency
- branched-chain ketoaciduria
- ketoacidemia

What are amino acid disorders?

Amino acid disorders are a group of inherited metabolic conditions in which certain amino acids cannot be broken down. This leads to an accumulation of toxic metabolites in the body which can cause serious health problems.

What is MSUD?

MSUD is an amino acid condition resulting from an enzyme defect in the breakdown of the branched chain amino acids leucine, isoleucine and valine. These amino acids are components of all proteins. The clinical features of MSUD result from accumulation of toxic metabolites.

What causes MSUD?

MSUD is caused by pathogenic variants in the BCKDHA, BCKDHB or DBT genes resulting in absent or decreased enzyme activity.

How common is MSUD?

MSUD is a rare condition with an incidence of about 1 in every 185,000 infants born in Canada. Although MSUD occurs in all ethnic groups, it is more common in Mennonites and people of French-Canadian ancestry.

What are the clinical features of MSUD?

MSUD usually presents acutely in the newborn period. Mild variants may present later. Initial clinical features include poor feeding, vomiting, lethargy, and failure to thrive. These can progress to an acute metabolic crisis with cerebral edema (swelling of the brain) which may lead to muscle rigidity, seizures, coma and death. Infants may have a characteristic smell of maple syrup. Milder forms of MSUD may present later.

What is the screening test for MSUD?

An increased concentration of branched chain amino acids is detected on the newborn blood spot screen. Newborn blood spot screening will not detect all infants with MSUD. 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 MSUD is confirmed by measurement of the branched chain amino acids in blood, and their corresponding organic acids in urine. The diagnosis can be confirmed by the enzyme analysis and molecular genetic analysis. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is MSUD treated?

Treatment consists of a low protein diet with restriction of the branched chain amino acids and a special formula. Some patients respond to high doses of thiamine, one of the B group of vitamins. 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 of glucose and lipids is necessary. Treatment improves the outcome of MSUD; however, some patient may have developmental delays even with treatment. The treatment is lifelong.

Is MSUD inherited?

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

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