Maple Syrup Urine Disease (MSUD) – Amino Acid Disorder

What are amino acid disorders?

The amino acid disorders are a class of inherited metabolic conditions that occur when certain amino acids either cannot be broken down or cannot be produced by the body, resulting in the toxic accumulation of some substances and the deficiency of other substances.

What is MSUD?

Maple Syrup Urine Disease (MSUD) occurs when the amino acids leucine, isoleucine, and valine cannot be broken down in the body. These are called “branched-chain” amino acids because of their structure. The symptoms of MSUD occur because of the toxic buildup of these amino acids in the blood.

What is the incidence of MSUD?

Maple Syrup Urine Disease is a rare disease that affects about 1 out of every 185,000 babies born in BC. Although MSUD occurs in all ethnic groups, it is more common in Mennonites and in people of French-Canadian ancestry.

What causes the disease?

In MSUD, the enzyme called “branched chain ketoacid dehydrogenase” (BCKAD) is deficient or not working well.

What are the clinical features of the disease?

Although babies with MSUD are normal at birth, without treatment they begin to have symptoms as soon as they are fed protein. Symptoms of MSUD include poor suck and poor feeding with weight loss, vomiting, lethargy, convulsions, coma, and death. During a metabolic crisis, children with MSUD may also have encephalopathy, muscle rigidity, and high levels of acidic substances in the blood (acidosis). The urine of an affected individual can have the odour of burnt sugar or maple syrup, giving the disorder its name.

There are also milder forms of MSUD with a later age of onset; the presentation is variable.

How is the diagnosis confirmed?

The diagnosis is confirmed by measuring the levels of amino acids in the blood and organic acids in the urine. The finding of alloisoleucine in the blood is characteristic. Enzyme testing and genetic testing of the BCKAD gene may also be used to confirm the diagnosis. Diagnostic testing is arranged by specialists at BC Children’s Hospital.

What is the treatment of the disease?

A low protein diet low in leucine, isoleucine, and valine is recommended in children with MSUD.

They should also avoid going long periods without food. Supplementation with thiamine may also be considered. In an acute symptomatic episode, IV fluids and glucose can be given to decrease the level of harmful substances in the blood. Treatment is coordinated by specialists at BC Children’s Hospital.

The BC Newborn Screening Program is a collaboration of BC Women’s Hospital & Health Centre, BC Children’s Hospital and the BC Perinatal Health Program, all part of the Provincial Health Services Authority.

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What is the outcome of treatment?

Treatment can prevent metabolic crises and their sequelae; however, response to treatment and therefore the outcome is variable.

Can a family have more than one child with MSUD?

Maple Syrup Urine Disease is inherited as an autosomal recessive disease. Parents of a child with MSUD are assumed to be carriers for the disorder and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with this condition. Prenatal testing for MSUD can be done as early as 10-12 weeks of pregnancy. Genetic counselling to discuss the benefits of prenatal testing options in more detail is recommended.

Unaffected siblings of a child with MSUD have a 2/3 chance of being carriers. MSUD carriers are healthy and do not experience symptoms of the disease.

Resources

http://www.newbornscreening.info/Parents/aminoaciddisorders/MSUD.html
http://www.msud-support.org/

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