Maple syrup urine disease (MSUD)

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. This leads to accumulation of these amino acids in the blood which causes the toxic symptoms of MSUD.

What causes the disease?

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

What are the clinical features of MSUD?

Although babies with MSUD are normal at birth, without treatment they begin to have symptoms as soon as they are given protein, usually in first few weeks of life. The symptoms of MSUD include poor suckling motion and lack of appetite with weight loss, vomiting, and lethargy which, if untreated, could progress encephalopathy and eventually a coma. Patients usually have maple syrup odor of cerumen and noticed to have finding and bicycling abnormal movements. Interestingly, these patients usually does not have hyperammonemia or metabolic acidosis. 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 achieved by measuring the levels of amino acids in the blood and organic acids in the urine. The finding of alloisoleucine in the blood is a characteristic of MSUD. Enzyme testing and genetic testing of the BCKAD gene may also be used to confirm the diagnosis.

What is the treatment of the disease?

A low protein diet that is low in leucine, isoleucine, and valine is recommended to children with MSUD. A Biochemical genetics specialist and a Metabolic Genetics dietitian should coordinate the treatment. Supplementation with thiamine may also be considered.