

Maple Syrup Urine Disease (MSUD)

General Overview

Q. What is MSUD?

A. Maple syrup urine disease (MSUD) is a treatable disorder that affects the way the body processes protein. Children with MSUD cannot use parts of the protein called branched-chain amino acids. If left untreated, branched-chain amino acids build up in the bloodstream and cause brain damage.

Q. Is there only one form of MSUD?

A. No, there are several forms of MSUD. This means that every child will not have the same degree of enzyme deficiency, so the level of treatment will vary.

Q. How does the body normally process branched-chain amino acids?

A. The body normally produces enzymes that convert branched-chain amino acids into smaller molecules. These are used by the body in other metabolic functions.

Q. What happens to branched-chain amino acids in a child with MSUD?

A. In a child with MSUD, branched-chain amino acids cannot be used by the body because the needed enzyme does not work properly. This results in large amounts of branched-chain amino acids, which are toxic to the brain and nervous system.

Q. What are the effects of having MSUD if it is not treated?

A. Untreated MSUD results in severe mental retardation and can cause death.

Q. What is the treatment for MSUD?

A. Maple syrup urine disease is treated with a special diet that is low in branched-chain amino acids. To prevent mental retardation, treatment must begin shortly after birth. Since protein is essential for normal growth and development, the child must continue on a special formula that provides protein and essential nutrients, but contains little or no branched-chain amino acids. People with MSUD require specialized treatment through a clinic with experience in treating this disorder.

Q. Why would a child have MSUD?

A. Maple syrup urine disease is an inherited disorder. It results when a baby receives a double-dose of a specific non-working gene involved in breaking down branched-chain amino acids (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is MSUD?

A. About one in every 200,000 babies in the United States is born with MSUD.

For more information about MSUD, please see the Disorders section of our website: www.doh.wa.gov/nbs.

