

# Maple Syrup Urine Disease (MSUD)

## General Overview

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### **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](http://www.doh.wa.gov/nbs).

