

Overview of Newborn Screening for Maple Syrup Urine Disease (MSUD) For Parents

What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions. One of these conditions is **maple syrup urine disease** (also called **MSUD**).

This name comes from the sweet “maple syrup” smell found in the urine of people with MSUD who do not receive treatment.

Babies who have a positive newborn screen need follow-up tests done to confirm they have MSUD.

Not all babies with a positive newborn screen will have MSUD.

What is MSUD?

When a person has MSUD, his/her body is not able to break down certain **amino acids** (“building blocks” that our bodies use to make proteins). People with MSUD are not able to break down **leucine, isoleucine and valine**. These three amino acids are also called “**branched-chain amino acids**” because they have a “tree-like” shape.

Most people with MSUD are missing one or more **enzymes** (proteins that helps our bodies work) called **branched-chain ketoacid dehydrogenases** (also called **BCKADs**). When these enzymes are missing, a person cannot break down the branched-chain amino acids. People with MSUD have high levels of these amino acids in their bodies.

What causes MSUD?

MSUD is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the genes for the BCKADs. We inherit one copy of each BCKAD gene from our fathers and one copy from our mothers. Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly. In order for a person to have MSUD, he or she must have two copies of a gene change for the same BCKAD. People with one BCKAD gene change do not have MSUD.

What are the symptoms of MSUD?

Every child with MSUD is different. Most babies with MSUD will look normal at birth. Most often, symptoms of MSUD appear shortly after birth or as soon as a baby starts feeding, but they may appear later in childhood. Some of the earliest symptoms of untreated MSUD include a poor appetite, vomiting, weight loss and urine that smells like maple syrup. Without treatment, people with MSUD can have a **metabolic crisis** (a severe illness caused by a build-up of branched-chain amino acids). Symptoms of a metabolic crisis include sleepiness, irritability, and vomiting. Other symptoms of untreated MSUD include a change in muscle tone (where the muscles may be floppy or rigid), seizures, developmental delay, and coma.

What is the treatment for MSUD?

There is no cure for MSUD. However, there are treatments that can help with the symptoms. People with MSUD may drink a special formula, take medicines or supplements, and/or need to follow a special diet that contains low levels of branched-chain amino acids. A person with MSUD will need treatment for his/her entire life.

What happens next?

Although there is no cure for MSUD, good medical care makes a difference. Children with MSUD should see a metabolic geneticist (a doctor who specializes in MSUD and other related conditions) as well as their pediatrician. Your child’s doctor will work with the metabolic geneticist to coordinate any treatment, tests, or appointments that your child needs.

Where are Indiana’s metabolic genetics clinics?

Indiana’s metabolic genetics clinics are located at Riley Hospital for Children at IU Health, Indianapolis, IN, (317) 274-3966 and The Community Health Clinic, Topeka, IN, (260) 593-0108.

Where can I get more information about MSUD?

- **STAR-G** - <http://newbornscreening.info/Parents/aminoaciddisorders/MSUD.html>
- **Region 4 Genetics Collaborative** – <https://www.region4genetics.org/education/families/>