



Newborn Screening FACT Sheet

Maple Syrup Urine Disease (MSUD)

What is MSUD?

MSUD is a type of amino acid disorder. It is named for the sweet maple syrup smell of the urine in untreated babies. People with MSUD have problems breaking down certain amino acids from protein in food.

What Causes MSUD?

Enzymes help start chemical reactions in the body. The most common form of MSUD is caused by the lack of a group of enzymes called “branched-chain ketoacid dehydrogenase” (BCKAD). This enzyme group breaks down three different amino acids. These are called “branched chain amino acids” (BCAA). When the body can’t break them down, they build up in the blood and cause problems.

What Symptoms or Problems Occur with MSUD?

[Symptoms are something out of the ordinary that parents notice.]

Symptoms start as soon as a baby is fed protein, usually right after birth. Some of the first are:

- poor appetite
- weak suck
- weight loss
- high-pitched cry
- urine that smells like maple syrup or burnt sugar

Babies with MSUD have periods of illness called Metabolic Crises. Some first symptoms are:

- too much sleepiness
- tiredness
- irritable mood
- vomiting

Brain damage can occur if untreated. This can cause mental disability. Some babies become blind. Most babies die within a few months if not treated. There are milder forms that may cause mental disability.

What is the Treatment for MSUD?

The following treatments are often used for children with MSUD:

1. Medical Formula – The doctor may prescribe a special medical formula with the right amount of protein. This will help keep your child’s BCAA levels in a safe range.

2. Diet low in BCCAs – The right diet is made up of foods that are very low in the BCAAs. This means your child will need to not eat foods such as cow’s milk, regular formula, meat, fish, cheese, and eggs. Regular flour, dried beans, nuts, and peanut butter also must be highly limited or not eaten. Many vegetables and fruits can be eaten in the right amounts. Your child should use this diet for life.

3. Checking BCAA levels – Your child will have regular blood tests to measure amino acid levels. The diet and formula may need to be changed based on blood test results.

Things to Remember

For children with MSUD, even minor illness such as a cold or flu can cause a Metabolic Crisis. Call your doctor right away when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- an infection or illness
- a fever
- behavior or personality changes (such as crying too much)
- difficulty walking or balance problems

Children with MSUD need to eat more starchy foods (such as rice, cereal, bread) and drink more fluids during any illness or they could have a Metabolic Crisis. Children who are sick may not want to eat. If they can’t eat, or if they show signs of a Metabolic Crisis, they may need to be treated in the hospital.