



# Maple Syrup Urine Disease (MSUD)

## Family Fact Sheet

### What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called MSUD.

**A positive newborn screen does not mean your baby has MSUD, but it does mean your baby needs more testing to know for sure.**

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

### What is MSUD?

MSUD affects an enzyme needed to break down proteins in the food we eat. In MSUD, the enzyme used to break down proteins is missing or not working properly.

A person with MSUD doesn't have enough enzyme to break down protein containing branched chain amino acids (BCAA). When the body can't break down BCAAs, they build up in the body and cause health problems.

MSUD is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause MSUD. Parents usually do not have signs or symptoms, or even know they carry the gene change.

### What problems can MSUD cause?

MSUD is different for each child. Some children with MSUD have few health problems, while other children may have very serious complications.

If MSUD is not treated, a child might develop:

- Feeding problems
- Weight loss
- High-pitched cry
- Urine that smells like maple syrup
- Sleepiness
- Vomiting
- Blindness (loss of eyesight)

It is very important to follow the doctor's instructions for testing and treatment.

### What is the treatment for MSUD?

MSUD can be treated. Treatment is life-long and can include:

- Diet low in BCAAs - a dietician will help you with the best diet for your child.
- Special formula low in BCAAs.

Children with MSUD should see their regular doctor, a doctor who specializes in MSUD, and a dietician.

Prompt and careful treatment helps children with MSUD live the healthiest lives possible.

### Michigan Resources and Support

**Michigan Newborn Screening**  
Nurse Consultant  
Toll-free: 1-866-673-9939  
[newbornscreening@michigan.gov](mailto:newbornscreening@michigan.gov)

**Children's Hospital of Michigan Metabolic Clinic**  
Toll-free: 1-866-442-4662

**Children's Special Health Care Services**  
Toll-free: 1-800-359-3722