

Maple Syrup Urine Disease (MSUD)

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests 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 (em-ess-you-dee).

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

Your baby's doctor will help arrange for more testing by specialists in disorders like MSUD.

What is MSUD?

MSUD affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. 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. The buildup of BCAAs can make the baby's urine smell like pancake syrup.

MSUD is a disorder that is passed on, or inherited, from a child's mother and father. Because MSUD is a genetic disease, family members are at risk of having MSUD too, even if no one in the family has had it before.

What problems can MSUD cause?

MSUD is different for each child. Some children with MSUD have fewer 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
- Developmental delay

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

What is the treatment for MSUD?

MSUD can be treated. The treatment is life-long. Treatment for children with MSUD can include:

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

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

Children with MSUD benefit from prompt and careful treatment.

Resources for parents

Genetics Home Reference

<http://ghr.nlm.nih.gov>

MSUD Family Support

www.msud-support.org

MN Children & Youth with Special Health Needs

(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program

www.health.state.mn.us/newbornscreening

