

Maple Syrup Urine Disease (MSUD)



What is newborn blood spot screening?

This is a blood test, from a heel poke, that is done a day or 2 after babies are born to test for treatable conditions including maple syrup urine disease (MSUD) that you, your doctor, or midwife can't see by just looking at your baby. This screening test is different from those done during pregnancy.



What does it mean if my baby needs more tests for MSUD?

A baby needs to have more testing for MSUD when their screening test result is abnormal or positive. This doesn't mean your baby has MSUD. It means your baby has a higher chance of having the condition and will need more tests to find out if they have MSUD or not.

It's normal to feel worried if your baby needs more testing. Your specialists and other healthcare providers are there to support you. They'll explain what will happen and answer your questions.

What is MSUD?

MSUD is a metabolic condition that affects how your body uses protein from the food you eat to make energy. This includes breastmilk and regular formula. With MSUD, your body can't break down 3 building blocks of protein called leucine, isoleucine, and valine. These proteins then build up in your body and can cause serious health problems. Babies with this condition often have urine that smells like maple syrup.

What are some early signs of MSUD?

Your baby may:

- find it hard to feed well and gain weight
- have urine that smells like maple syrup
- be very sleepy (hard to wake for feedings)

What causes MSUD?

MSUD is genetic. This means babies with MSUD are born with a gene from both parents, which has a change in it. This change stops the gene from working properly. MSUD isn't caused by anything that happened during pregnancy.

There may be no signs of MSUD at birth. It's important to remember that this condition can't be seen by looking at your baby.



What can I expect with more testing?

Everything that will happen with more testing will be explained to you. Your baby may need to have their blood and urine tested to check for MSUD.

This testing may be done at a clinic in Edmonton or Calgary that specializes in MSUD.

What if more testing shows my baby has MSUD?

If testing shows that your baby has MSUD a specialized healthcare team will work with you to make a care plan for your baby. You and your baby will get the best care and support possible.

How is MSUD treated?

Babies with MSUD usually go on a low-protein diet and drink a special formula. The specialized healthcare team will give you detailed instructions about how to manage your baby's health.



