

Citrullinemia (CIT)

(Sounds like SIT-ROO-LYN-E-MIA)



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 citrullinemia (CIT) 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 CIT?

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

CIT 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 CIT, your body can't safely break down certain building blocks of protein called amino acids. This causes a buildup of a chemical called ammonia in your body and can cause serious health problems.

What causes CIT?

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

There may be no signs of CIT 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 CIT.

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

What if more testing shows my baby has CIT?

If testing shows that your baby has CIT 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 CIT treated?

Babies with CIT usually go on a low-protein diet and drink a special formula. They may also need special medicine to help lower ammonia levels. The specialized healthcare team will give you detailed instructions about how to manage your baby's health.



