

Biotinidase (BIOT) Deficiency

(Sounds like BYE-OH-TIN-I-DAYS DE-FISH-IN-SEA)



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 biotinidase (BIOT) deficiency 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 BIOT deficiency?

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

BIOT deficiency is a metabolic condition that affects your body's ability to recycle a vitamin called biotin. Biotin plays many important roles in your body. For example, biotin helps break down protein. A deficiency, or not enough biotin, can cause serious health problems.

What causes BIOT deficiency?

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

There may be no signs of BIOT deficiency 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 BIOT deficiency.

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

What if more testing shows my baby has BIOT deficiency?

If testing shows that your baby has BIOT deficiency 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 BIOT deficiency treated?

Your doctor at the metabolic clinic may suggest biotin supplements. The specialized healthcare team will give you detailed instructions about how to manage your baby's health.

