



INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Biotinidase Deficiency Newborn Screening Result

What is newborn screening?

These are routine tests done soon after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including biotinidase deficiency.

What does it mean if my baby has a positive biotinidase deficiency newborn screening result?

This result does **not** mean that your baby has biotinidase deficiency. It means that more testing is needed because your baby **might** have biotinidase deficiency. Babies with biotinidase deficiency can grow and develop normally if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has biotinidase deficiency.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has biotinidase deficiency until follow up testing has been done.

What is biotinidase deficiency?

Biotinidase deficiency is an inherited (genetic) condition that happens when the enzyme biotinidase is not working properly. This enzyme's job is to recycle (reuse) a vitamin called biotin. Biotin is an important co-factor (helper) for many other enzymes in the body that make certain fats and carbohydrates and break down proteins. If the body is unable to recycle biotin, all of the enzymes that it helps are unable to do their jobs which can cause serious health problems.

Some people with biotinidase deficiency have a milder form of the disease. However, there is no way to know at birth who will develop mild problems caused by biotinidase deficiency or serious problems caused by biotinidase deficiency.

Biotinidase deficiency affects about 1 in every 60 000 babies born in Ontario.

What causes biotinidase deficiency?

Biotinidase deficiency happens when the enzyme biotinidase is either missing or not working properly. This enzyme's job is to recycle (reuse) the vitamin biotin. If biotin can not be recycled by the body, many other enzymes can not do their jobs which can cause serious health problems.

How do I find out if my baby has biotinidase deficiency?

Blood tests are done to find out if a baby who screened positive actually has biotinidase deficiency.

When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange follow up testing as soon as possible.

Why screen for biotinidase deficiency?

Babies who have biotinidase deficiency look normal when they are born but may develop seizures, low muscle tone, mental retardation, hearing loss and skin and hair problems. If treatment for biotinidase deficiency begins before symptoms start, babies with biotinidase deficiency can grow and develop normally.





How is biotinidase deficiency treated?

Babies with biotinidase deficiency are treated and monitored by a team of health care specialists including a metabolic doctor. Biotinidase deficiency is treated with large doses of biotin. The treatment is life long and is very good at preventing the problems that go along with biotinidase deficiency.

How does a baby get biotinidase deficiency?

Biotinidase deficiency is an inherited (genetic) disease. A baby with biotinidase deficiency inherited two non-working copies of the biotinidase enzyme gene (called BTB), one copy from each parent. People who have one non-working copy of the BTB gene are called "carriers." Carriers of biotinidase deficiency are healthy, do not have, and will never develop, symptoms of biotinidase deficiency.

Where can I get more information?

For more information on newborn screening, please visit the Parents section of our website at www.newbornscreening.on.ca or talk to your local health care provider.

For more information on biotinidase deficiency, please visit the Genetics Home Reference website at <http://ghr.nlm.nih.gov/condition=biotinidasedeficiency>.

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for biotinidase deficiency. Please remember that this fact sheet was written for information purposes only. The fact sheet does not replace professional medical advice, diagnosis or treatment.

