Information on a Positive Newborn Screening Result for

Biotinidase Deficiency (BD)

MARITIME Newborn Screening / Dépistage néonatal DES MARITIMES

What is newborn screening (NBS)?

These are routine tests done soon after birth. A few drops of blood from a baby's heel are put onto a piece of absorbent paper (blotter). The blood is tested for serious but treatable conditions. These tests are done because a newborn can look healthy but have one of these conditions and need treatment. We want to find babies with these conditions so that we can start treatment to keep them as healthy as possible.

Summary:

- This result means a baby may have biotinidase deficiency (BD). **More testing is needed** for a final answer.
- **There is treatment for BD.**
- Maritime Newborn Screening connects the baby with the IWK genetics metabolic team for follow-up testing.

What are the possible results from NBS?

Most babies have a "screen negative" (normal) result. This means a baby has a decreased chance of having the conditions on newborn screening, and no follow-up is needed. Some babies (~10%) require a repeat of the newborn screen and another sample to be collected and tested. A "screen positive" result means a baby has an increased chance to have a condition from newborn screening, but it does not mean the baby has this condition. More testing is needed to give a final answer.

What does it mean if a baby has a positive screen result for biotinidase deficiency (BD)?

This result does not mean that the baby has BD. It means that more testing is needed because the baby **may** have BD. Babies with BD grow and develop better if treatment begins as soon after birth as possible. Follow-up testing is done quickly to find out if a baby has BD. This is done for the baby's best possible outcome.

What happens when there is a positive newborn screen result for biotinidase deficiency (BD)?

A healthcare provider from Maritime Newborn Screening (MNBS) will contact the primary care provider and the family to discuss the result. MNBS will connect the baby to the clinical team providing the follow-up. This is the **Maritime Medical Genetics Service (MMGS) metabolic team at the IWK**. The MMGS metabolic team will arrange for follow-up appointments and testing as soon as possible. Families may feel worried about the baby's newborn screen result. Many families in this situation feel this way. The healthcare teams are here to support families during the next steps.

What is biotinidase deficiency (BD)?

BD is an **inherited** condition, meaning the baby is born with it. BD is also a **metabolic** condition where the body cannot properly recycle the vitamin **biotin**. Typically, the enzyme **biotinidase** processes biotin into a form the body can use. In BD, the biotinidase enzyme is missing or not working. BD can be milder (partial deficiency) or more severe (profound deficiency), depending on how well the biotinidase enzyme is working.

- Profound BD: Symptoms typically appear in the first few months of life. If *untreated*, symptoms can include seizures, weak muscles (hypotonia), progressive hearing and vision loss, skin rashes, hair loss, delayed development, and problems with breathing, movement, and balance.
- Partial BD: Symptoms typically appear later in childhood, or may only occur during illness or stress. If *untreated*, symptoms can include muscle weakness, skin rashes, and hair loss.

Treatment is given to prevent these symptoms, or improve them if they are already happening.

How many babies have biotinidase deficiency (BD)?

BD is a rare disease. BD affects about 1/60,000 babies, but partial BD may be more common.

Why screen for biotinidase deficiency (BD)?

Babies with BD usually look normal at birth and may not show any health problems right away. If they are not screened by newborn screening they might not be diagnosed right away. Treatment for BD is most effective when started before or soon after symptoms begin. Newborn screening allows us to find babies that need treatment as soon as possible.

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How is a baby diagnosed with biotinidase deficiency (BD)?

To find out if a baby has BD, they are seen by a genetics metabolic team for follow up testing. Blood tests measuring biotinidase enzyme activity and genetic changes to the *BTD* gene confirm a diagnosis of BD. It can take a few days/weeks to confirm a potential diagnosis. This waiting period can be hard for families. The healthcare teams are here to support families.

How is biotinidase deficiency (BD) treated?

BD is treated by taking the vitamin **biotin** (B7). Biotin can be taken as a pill, or made into a liquid to be given to babies and young children. Treatment for BD is coordinated by the MMGS metabolic team.

How does a baby get biotinidase deficiency (BD)?

BD is an **inherited condition**. This means the baby is born with it. When a baby has BD, we know that biological parents are likely carriers of the condition. Carriers do not have the condition but can have a baby with the condition if the baby inherits a non-working copy of the *BTD* gene from each parent. This is autosomal recessive inheritance (see Figure 1).

Could a family have another baby with biotinidase deficiency (BD)?

Yes, if biological parents are carriers there is a 25% (1 in 4) chance in each pregnancy to have a child with BD. We offer genetic counselling through **Maritime Medical Genetics Service at the IWK** to talk about the following:

- Inheritance of BD
- Chance of recurrence of BD in future pregnancies
- Testing options for other family members

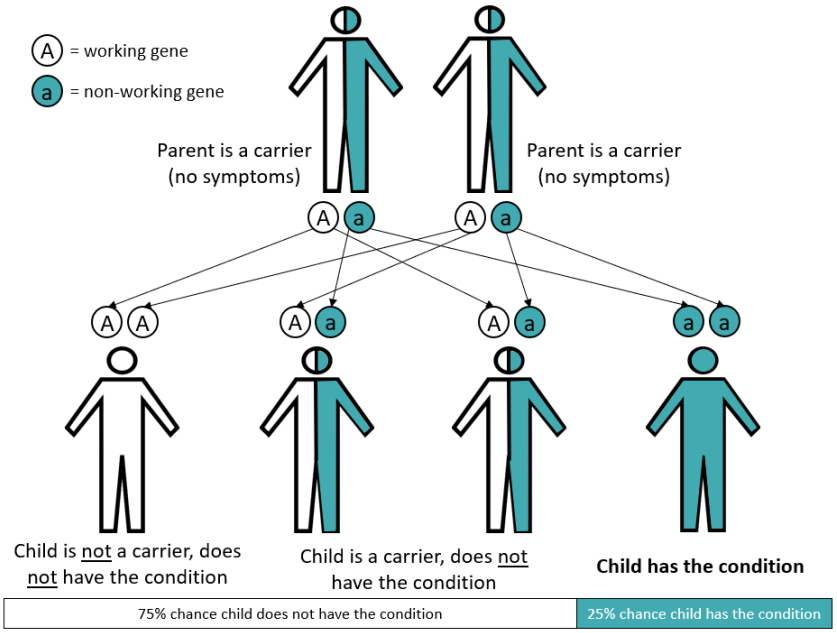


Figure 1. Autosomal recessive inheritance when both parents are carriers of biotinidase deficiency.

Where can I get more information?

- For more information on newborn screening, please visit our website at www.maritimenewbornscreening.ca or call the newborn screening genetic counsellor at 902-470-2783.

- Websites for families:
  - ThinkGenetic Foundation: https://thinkgenetic.org/diseases/biotinidase-deficiency/

- Websites for healthcare providers:
  - MedlinePlus: https://medlineplus.gov/genetics/condition/biotinidase-deficiency/
  - GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1322/

NOTE TO PARENTS/GUARDIANS: *This material is provided for informational purposes and provides basic information only. This material is not intended to be and does not take the place of medical advice, diagnosis, or treatment. Please talk to your health care provider if you have any questions or concerns.*

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