

Positive Result:

Blood Spot Screen Result Notification



Absent/Reduced Biotinidase (BTD)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has no or low levels of an enzyme called biotinidase.

What does this mean?

Absent or low levels of biotinidase can indicate that your child may have biotinidase deficiency. A positive result does not mean your baby has biotinidase deficiency, but more testing is needed to know for sure.

What happens next?

Your baby's doctor or a metabolic specialist familiar with biotinidase deficiency will help arrange for more testing and potentially start your baby on special vitamins. Your baby will also likely be scheduled to be seen by a metabolic specialist.

What is biotinidase deficiency?

Biotinidase deficiency is a condition present in a baby at birth where the body is not able to use and recycle the vitamin, biotin. Biotin comes from food and is needed for energy and growth. There are two forms of biotinidase deficiency: partial (mild) and profound (severe). Both forms may cause health problems.

What health problems can it cause?

Biotinidase deficiency is a lifelong condition. If untreated, it can cause:

- Muscle weakness
- Hearing and vision problems
- Hair loss
- Skin rashes
- Seizures
- Intellectual disabilities

Children with biotinidase deficiency can benefit from prompt and careful treatment.

What treatment options are available?

Although biotinidase deficiency cannot be cured, it can be treated. Children with biotinidase deficiency are treated with lifelong biotin supplements. If treated before symptoms develop, children can have healthy growth and development.

Children with biotinidase deficiency should see their regular doctor and a doctor who specializes in biotinidase deficiency.

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org