

Biotinidase Deficiency

(bye-o-tin-ah-daze de-fish-in-sea)

FAMILY FACT SHEET POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called biotinidase deficiency.

A positive newborn screen does not mean your baby has biotinidase deficiency, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in disorders like biotinidase deficiency.

What is biotinidase deficiency?

Biotinidase deficiency affects an enzyme needed to free biotin (one of the B vitamins) from the food we eat, so it can be used for energy and growth.

A person with biotinidase deficiency doesn't have enough enzyme to free biotin from foods so it can be used by the body.

Biotinidase deficiency is a disorder that is passed on, or inherited, from a child's mother and father. Because biotinidase deficiency is a genetic disease, family members are at risk of having biotinidase deficiency too, even if no one in the family has had it before.

What problems can biotinidase deficiency cause?

Biotinidase deficiency is different for each child. Some children have a mild, partial biotinidase deficiency with few health problems, while other children may have complete biotinidase deficiency with serious complications.

If biotinidase deficiency is not treated, a child might develop:

- Muscle weakness
- Hearing loss
- Vision (eye) problems
- Hair loss
- Skin rashes
- Seizures
- Developmental delay

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for biotinidase deficiency?

Biotinidase deficiency can be treated. The treatment is life-long. Treatment for children with biotinidase deficiency includes:

- Daily biotin vitamin pill(s) or liquid

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

With prompt and careful treatment, children with biotinidase deficiency have a good chance to live healthy lives with normal growth and development.

Resources for parents

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

GeneTests
www.genetests.org

MN Children & Youth with Special Health Needs
(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program
www.health.state.mn.us/newbornscreening

