BIOTINIDASE DEFICIENCY (BIOT)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested 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.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

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 genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause biotinidase deficiency. Parents usually do not have signs or symptoms, or even know they carry the gene change.

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. Treatment is life-long and includes:

- Daily biotin vitamin pill(s) or liquid.

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

Prompt and careful treatment helps children with biotinidase deficiency live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Michigan Metabolic Coordinating Center
C.S. Mott Children's Hospital, Michigan Medicine:
1-734-764-0579

Children's Special Health Care Services
Toll-free: 1-800-359-3722

MDHHS Michigan Department of Health & Human Services | Michigan Newborn Screening

Phone 1-866-673-9939
www.michigan.gov/newbornscreening
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