NORTH CAROLINA NEWBORN SCREENING PROGRAM

Biotinidase Deficiency Fact Sheet for Providers

What is Biotinidase Deficiency?

Biotinidase deficiency is an inherited disorder that affects a patient's energy metabolism. Specifically, a patient cannot reuse or recycle the vitamin biotin, which is required to metabolize many kinds of food correctly. There are two types of biotinidase deficiency, partial and profound, which differ in severity and treatment. Both forms can lead to serious health problems.

What are the symptoms of Biotinidase Deficiency?

Newborns with biotinidase deficiency are often asymptomatic.

Symptoms of biotinidase deficiency may include:

- Lethargy
- Hypotonia
- Poor feeding

Severe biotinidase deficiency may result in the following symptoms:

- Hypotonia
- Seizures
- Alopecia
- Skin rash
- Hyperventilation
- Apnea

If the baby is symptomatic, please contact the nearest genetics center or the UNC-CH operator at (984) 974-1000 and ask to speak to the pediatric genetics physician on call.

How is Biotinidase Deficiency identified and diagnosed?

All infants with results concerning for biotinidase deficiency on Newborn Screening will require additional evaluation and confirmatory testing.

- A clinical examination with a genetics team
- Confirmation with whole blood sample for biotinidase enzyme testing and often molecular DNA testing for the biotinidase gene.

How do I handle an abnormal screen for Biotinidase Deficiency?

Early diagnosis is very important for biotinidase deficiency. Please IMMEDIATELY take the following steps:

- Follow the instructions faxed by the North Carolina Newborn Screening (NBS) Coordinator regarding follow up testing, referrals and treatment. Reach out to the NBS Coordinator if you have additional questions.
- If the baby is displaying any of the symptoms listed, consult with a metabolic or genetic specialist and arrange to transfer the baby to a hospital for further treatment.

How is Biotinidase Deficiency treated?

- A patient with biotinidase deficiency is treated with lifelong biotin supplements.

Where do I go for more information?

Use your phone's camera to scan the QR codes below:

Medline Plus: https://medlineplus.gov/download/genetics/condition/biotinidase-deficiency.pdf

Gene Reviews: https://www.ncbi.nlm.nih.gov/books/NBK1322/

NC DEPARTMENT OF
HEALTH AND HUMAN SERVICES
Division of Child and Family Well-Being

State of North Carolina Department of Health and Human Services
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