Michigan Department of Community Health
MDCH
Jennifer M. Granholm, Governor
Janet Olszewski, Director

www.mi.gov/newbornscreening

Michigan Resources & Support

Children's Hospital of Michigan Metabolic Clinic
Toll-free: 1-866-44CHMMC

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

Early On® Michigan
Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Genetics Connection
www.migeneticsconnection.org

Michigan Newborn Screening
Follow-up Coordinator
Toll-free: 1-866-673-9939
E-mail: MDCH-newbornscreening @ michigan.gov

Michigan NBS Parent Liaison
Toll-free: 1-866-673-9939
E-mail: NBS-parent@michigan.gov

National Resources & Support

Biotinidase Family Support Group
www.biotinidasedeficiency.20m.com/

Family Village
www.familyvillage.wisc.edu

GeneReviews
www.genetests.org

Genetic Alliance
www.geneticalliance.org

What is Biotinidase Deficiency?

Biotinidase deficiency is an inherited disorder preventing babies from using biotin (a needed vitamin) in a normal manner. Biotin is required to break down certain foods. Biotinidase deficiency occurs in about 1 in 27,000 Michigan newborns. If untreated, profound biotinidase deficiency can result in seizures, mental retardation or coma. The milder form called partial biotinidase deficiency is not associated with serious complications.

How may Biotinidase Deficiency affect my child?

Timing of Signs and Symptoms

Usually signs of biotinidase deficiency occur in infancy or early childhood. Signs may occur from the first few weeks of life until 10 years of age or older.

Common Early Signs

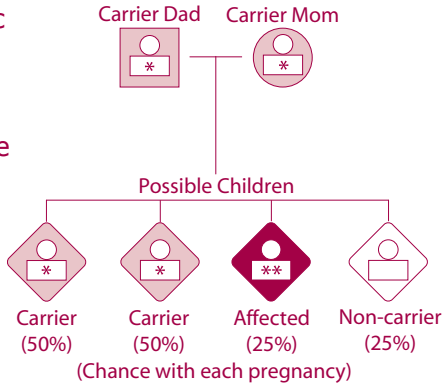
- Seizures
- Low muscle tone (hypotonia)
- Hair loss (alopecia)
- Skin rashes

Common Later Signs

- Developmental delay
- Speech problems
- Vision and hearing loss
- Abnormalities in movement and muscle control (Ataxia)

How does Biotinidase Deficiency occur?

Biotinidase deficiency is a genetic disorder. Parents of an affected child carry a genetic trait that can cause biotinidase deficiency. Both parents pass the trait to a child with biotinidase deficiency. There is a 1 in 4 chance that each child will have biotinidase deficiency when both parents carry the trait for the disorder.



How is Biotinidase Deficiency treated?

Newborns and children are given biotin (vitamin) supplements. No special diet is required. Regular monitoring and care through the Metabolic Clinic and your pediatrician are required to ensure proper health, growth and development.

For more information contact the Newborn Screening Program toll-free at 1-866-673-9939 or e-mail NBS-Parent@michigan.gov

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