



Biotinidase Deficiency

What is it?

Biotinidase deficiency is a condition that causes the body to have trouble using biotin, an important vitamin the body needs. Biotin is found in many foods. However, a person with biotinidase deficiency needs more biotin than the amount eaten in the normal diet. Biotinidase is an enzyme (a chemical that causes reactions in the body to take place). In a person with biotinidase deficiency, the biotinidase enzyme is not working well, and the body does not have enough free biotin to use.

What causes it?

Biotinidase deficiency is inherited when both parents pass an abnormal biotinidase gene to their child. This means both parents are carriers of biotinidase deficiency. Carriers do not experience any health problems related to biotinidase deficiency. When two carriers of biotinidase deficiency have children together, there is a 1 in 4 (25 percent) chance for each child to have biotinidase deficiency.

How is Biotinidase deficiency detected?

Newborn screening test is done on tiny samples of blood taken from the infant's heel 24-48 hours after birth. After a positive newborn screen, testing at special labs must be done to know for sure if a baby has biotinidase deficiency.

What problems can it cause?

Biotinidase deficiency is different for each child. Untreated biotinidase deficiency can cause seizures, low muscle tone (floppiness), hearing loss, eye problems, hair loss, skin rashes and possibly coma or death. It is very

important to follow the doctor's instructions about caring for a child with biotinidase deficiency.

How is it treated?

Biotinidase can be treated. A child needs to take extra biotin every day (more than what is normally eaten in the diet) in the form of a pill. The treatment is life-long.

Biotinidase Deficiency in Children?

The child should have a primary care doctor and a pediatric metabolic specialist, and a dietician. These health professionals give the child good medical care and educate the family about the condition.

Treatment for biotinidase deficiency is a life-long, and a child with biotinidase deficiency should see a doctor regularly.

Where can I find more information?

www.netnet.net/mums
877.336.5333

www.rarediseases.org
203.744.0100

Children's Special Health Services (CSHS)

State Capitol Judicial Wing
600 E. Boulevard Ave., Department 301
Bismarck, ND 58505-0269

Toll Free: 800.755.2714
701.328.2436

Relay TDD: 701.328.3975

CSHS website: www.ndhealth.gov/CSHS

North Dakota Department of Health website:
www.ndhealth.gov

Family resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

Family Resources

Family to Family Network
Center for Rural Health
University of North Dakota
School of Medicine and Health Sciences
P.O. Box 9037
Grand Forks, ND 58202-9037
Toll Free: 888.434.7436
701.777.2359
Fax: 701.777.2353
E-mail: NDF2F@medicine.nodak.edu
www.medicine.nodak.edu/crh

Pathfinder Services of ND
Pathfinder Family Center
1600 2nd Ave. SW, Ste. 19
Minot, ND 58701
Toll Free: 800.245.5840
701.837.7500
Relay TDD: 701.837.7501
E-mail: ndpath01@ndak.net
www.pathfinder.minot.com

Family Voices of North Dakota, Inc.
P.O. Box 163
Edgeley, ND 58433
Toll Free: 888.522.9654
701.493.2634
Fax: 701.493.2635
www.geocities.com/ndfv

**This fact sheet has general information.
Every child is different and some of these facts
may not apply to your child specifically.
Certain treatments may be recommended for
some children but not others. All children
should be followed by a metabolic doctor in
addition to their primary-care provider.

Acknowledgement

The North Dakota Department of Health
Newborn Screening Program thanks Star-G
Screening, Technology and Research in
Genetics for allowing us to utilize its material.

Disclaimer

THIS INFORMATION DOES NOT
PROVIDE MEDICAL ADVICE. All content
including text, graphics, images and
information is for general informational
purposes only. You are encouraged to talk with
your doctor or other health-care professional
about the information contained in this fact
sheet. You are encouraged to review the
information carefully with your doctor or other
health-care provider. The content is not
intended to be a substitute for professional
medical advice, diagnosis or treatment. Never
disregard professional medical advice, or delay
in seeking it, because of something you have
read on this information sheet.



North Dakota Department of Health
Newborn Screening Program
600 E. Boulevard Ave., Dept. 301
Bismarck, ND 58505-0200
800.472.2286 or 701.328.2493
www.ndhealth.gov/familyhealth