Kansas
Department of Health and Environment

Biotinidase Deficiency
Information for Parents

➢ Overview

Biotinidase deficiency is a rare condition that affects the way a person's body uses the vitamin biotin. Individuals with biotinidase deficiency can not use the biotin that is normally found in foods. Treatment is very effective for these individuals, and can prevent most symptoms from occurring.

➢ What is biotinidase deficiency?

Biotin is found in many foods and is important for proper growth and development. Normally, an enzyme in our body called biotinidase helps separate biotin from the food we eat so our body can use it to help other enzymes in the body do their job. In babies with biotinidase deficiency, the enzyme doesn't work very well so the baby's body doesn't get enough biotin.

➢ Why is newborn screening done for biotinidase deficiency?

Newborn screening is done for biotinidase deficiency so that babies with this condition can be diagnosed quickly. If babies are diagnosed quickly, treatment can begin before any health problems occur.

➢ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has biotinidase deficiency?

No, not necessarily. Newborn screening tests the baby's level of biotinidase enzyme, but additional tests will need to be done to determine if the baby has biotinidase deficiency or not.

➢ How common is biotinidase deficiency?

About 1 out of every 75,000 babies born has either a partial or complete absence of this enzyme.

➢ How is biotinidase deficiency inherited?

Biotinidase deficiency is inherited in an autosomal recessive pattern. Parents of a child diagnosed with biotinidase deficiency are unaffected. These individuals are carriers of the condition and have one normal BTD gene and one abnormal BTD gene. Each pregnancy between carrier parents has a 25% chance of producing a child affected with biotinidase deficiency, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

➢ What are the signs and symptoms of biotinidase deficiency?

Babies that have biotinidase deficiency will appear normal at birth. Within a few weeks or months after birth, symptoms will develop if the individual is untreated. The number of symptoms that a baby will develop, as well as how severe the symptoms will be, varies from baby to baby.

Some common early signs include:

- seizures
- low muscle tone (floppiness)
- hair loss
- skin rash

Some common later signs include:

- hearing loss
- vision loss
- delayed development
- ataxia (poor coordination)
- possibly coma and death

Once treatment begins, most of the symptoms will disappear. Some symptoms, such as hearing loss, developmental delay, and vision loss, may not be fully corrected with treatment, but usually won't get worse. If babies are diagnosed early and treatment is started before symptoms appear, they usually don't develop any symptoms.

➢ How is biotinidase deficiency diagnosed?

Confirmation of any initial abnormal newborn screening result requires an immediate referral to a metabolic disease specialist.

➢ Is there a cure for biotinidase deficiency?

No, there is no cure for biotinidase deficiency. Treatment can ensure that babies grow up healthy and prevent many symptoms of this condition.

➢ How is biotinidase deficiency treated?

Children with biotinidase deficiency will need to take extra biotin. Biotin supplements come in pill and liquid forms. They will have to take this supplement throughout their life. Babies should also have regular checkups with a metabolic specialist and their pediatrician.

➢ Where can I get additional information?

Parents may find beneficial support groups that give them the opportunity to talk with parents of other children with biotinidase deficiency. For more information about newborn screening in general and about biotinidase deficiency specifically, contact the National Newborn Screening and Genetics Resource Center, 1912 W. Anderson Lane, Suite 210, Austin, TX 78757; telephone 512-454-6419; fax 512-454-6509.

GeneTests at www.genetests.org

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

Biotinidase Deficiency: A Booklet for Families & Professionals
www.ccmckids.org/research/Biotinidase/Biotinidase_Deficiency_Booklet.pdf