Parents' Guide To BD

Biotinidase Deficiency

California
Department of
Public Health

Newborn Screening Branch
Genetic Disease Screening Program
www.dhs.ca.gov/nbs

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To Parents:

California State Law requires that all babies have the newborn screening test before leaving their hospital of birth. A few drops of blood were taken from your baby's heel. One of the tests was for biotinidase deficiency.

Your health care provider may have told you that your baby has biotinidase deficiency (BD). Babies with this disease can look healthy at birth. If left untreated, these babies often have serious and permanent health problems. With early and ongoing care, your baby should develop normally both physically and mentally.

This booklet was written to help parents learn more about this disorder. Use this booklet to learn more about how to care for your child.

For more information about the Newborn Screening Program or BD, visit our website at www.dhs.ca.gov/nbs.

Discuss this booklet with your doctor.

What Is BD?

BD stands for “biotinidase deficiency.” This results in problems using biotin, a member of the B vitamin complex family, which is needed for digestion or metabolism of food.

What Causes BD?

BD occurs when an enzyme called biotinidase is either missing or not enough of this enzyme is made. Biotinidase is an important enzyme. Enzymes are special proteins the body uses to process the foods we eat and make certain fats and sugars. Biotinidase helps our body process biotin from the foods we eat and reuse the biotin. Biotin is important to the health of skin, hair, bone marrow, and the nervous system.

What Causes The BD Enzyme To Be Missing Or Not Working?

Genes tell the body to make various enzymes. People with BD have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the BD enzyme either is not made at all or not enough is made.

What Types Of BD Are There?

There are two types of BD, partial and profound. Babies with partial BD have a certain level of the enzyme biotinidase in their blood. This level is between 10-30% of the normal biotinidase levels. These babies make some enzyme. This is a milder form of the disease. These babies do not have symptoms but may develop some symptoms at times of stress like when they are sick or have a poor diet.

Babies with profound BD have no activity or less than 10% of enzyme activity. Profound biotinidase deficiency is a more severe form of the disease. This booklet will only deal with profound BD.

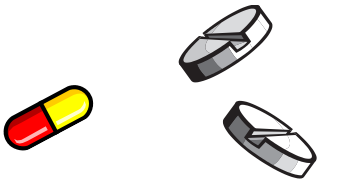
If BD Is Not treated, What Problems Occur?

If left untreated, babies will develop symptoms that can include skin rash, poor muscle tone, developmental delay (mental retardation), seizures, hearing loss, vision problems, hair loss and some life-threatening conditions. The severity and types of symptoms can vary from child to child. Symptoms can show up as early as one week of age but are usually seen between 3-6 months of age if untreated.

What Is The Treatment For BD?

Since people with this disease are unable to use the biotin from the food in their diet or recycle the biotin, a special biotin supplement must be taken every day shortly after birth. Treatment is life long. With proper treatment, BD is not a life-threatening disease. Babies treated before symptoms appear usually do not develop any symptoms of the disease later in life.

How To Give Biotin



Biotin comes in either a tablet, capsule or liquid form. Check with the metabolic specialist or nutritionist as to which form is best for your child. Give the biotin at the same time every day and make it a part of your routine so you do not forget.

With infants less than 12 months, the tablets need to be crushed or the capsules need to be opened and given carefully. When your infant is older, it will be easier to give. There are several ways you can give the biotin medicine.

For The Infant:

- Wash hands well. Crush tablet(s) between two spoons
  or
  open the capsule(s) then mix with a small amount of water, breast milk or formula. Give this mixture through a clean dropper on the side of the mouth between the cheek and gum. To be sure that all of the medicine is taken, draw up some water into the dropper after you have given the mixture and give this water to your baby.

Use room temperature water. DO NOT mix with hot water.

Or

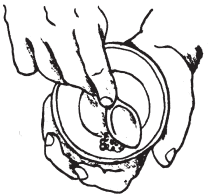
- Wash hands well. Finely crush the tablet(s) between two spoons until it is a fine powder. Moisten your fingertips with water and dip into crushed tablet(s) or opened capsule(s). Let your baby suck the powder off your finger and continue to dip your finger in until no powder is left. Offer the baby a small amount of water after giving the biotin.

For The Older Infant And Toddler:

When your baby is on solid foods, you may want to try one of these methods.

- Wash hands well. Place tablet(s) on a spoon and add a few drops of water with a dropper to soften them. After a few minutes, add a few drops of water on the spoon then feed this mixture to your baby.

- Wash hands well. Mix the crushed tablet(s) or opened capsule(s) in a small amount of baby cereal, applesauce, breast milk or formula. Always keep this separate from the rest of your baby's food. For example, if your baby is taking two tablespoons of cereal in the morning, mix the crushed tablet(s) or opened capsule(s) with a ½ teaspoon of cereal in a separate dish. Give this portion to your baby. Some doctors recommend waiting at least one half hour after giving the capsule/tablet(s) before feeding your baby the rest of the meal. That way, you will know that your baby has received all the biotin.



DO NOT put the tablet(s) into a full serving of the baby's food. DO NOT put the capsule/tablet(s) in the baby's bottle. The baby may not eat or drink the entire amount.

When your child is older, he or she may take the whole tablet(s) by mouth. Since they are so small, they will be swallowed whole. Keep a record of when you give the medicine. You can mark a calendar when the tablets have been given. If you forget to give a dose on one day, give it the next day. Missing a dose once should not cause a problem, but if not given for more than a few days, symptoms of BD may occur.

What Else Can I Do?

- Call your doctor or nurse if your child develops any of the following symptoms of BD:
  - seizures
  - poor muscle tone
  - hair loss
  - skin rash/infection
  - poor coordination or clumsiness
  - redness of the eye (conjunctivitis)
- Always keep an extra supply of the tablets for emergencies. Keep extra medicine in your diaper bag and with your emergency/earthquake supplies. Always check the expiration date.
- Always examine the tablets when the prescription is filled. Note the color and the number on the tablet to be sure it is the right dose.
- Be sure to bring the bottle of biotin tablets with you to each of your child's doctor visits.

Will My Child Grow Like Other Children?

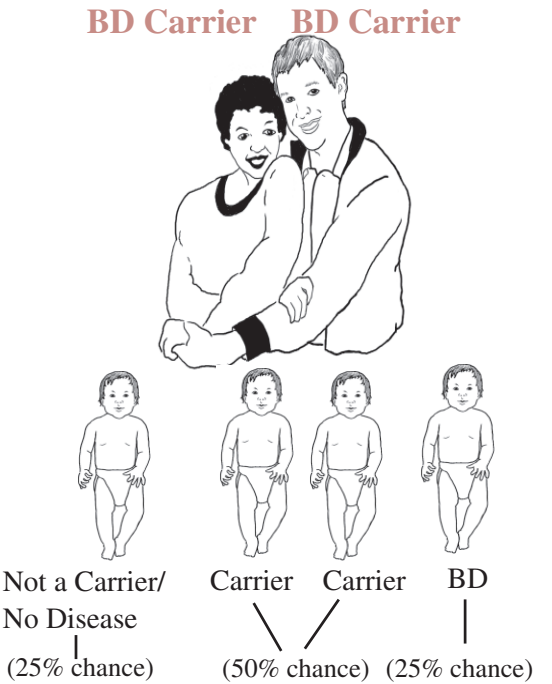
With early detection and treatment before symptoms appear, children with this disease do well. They are like any other children except they need to take daily doses of biotin. In addition, regular visits to your baby's doctor and scheduled visits to a special clinic called a metabolic center are needed to maintain normal health, growth and development.

Remember, your child has inherited BD and will never outgrow it. BD is not contagious. This means you can not "catch it" like a common cold. Your child can not "give" it to another child. Daily treatment with biotin and taking your baby for doctor visits to monitor his or her health is important for his or her well-being. Your baby will have blood tests to monitor the level of biotin on a regular basis. These blood tests help the doctor to adjust the biotin so your baby will get the correct amount. The dose may be adjusted as your baby grows.

Can I Have Other Future Children with BD?

Yes, if both parents have one non-working gene for BD, they can pass on this gene to their children. A child with BD inherited it from both parents just as he or she inherited hair, skin or eye color. Everyone has a pair of genes that make the biotinidase enzyme. In children with BD, neither of these genes work correctly. These children inherit one non-working gene for the condition from each parent. Parents of children with BD rarely have the disease. Instead, each parent has a single non-working gene for BD. They are called carriers. Carriers are not affected because their other gene of this pair is working correctly. The diagram on the right shows the chances of having a child with BD in a future pregnancy.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have two working genes. This means the child is not a carrier and does not have the disease. There is a 50% chance for the child to be a carrier, just like the parents. There is a 25% chance for the child to have BD.



CCS Metabolic Centers in California

Northern California

Children's Hospital Central California
Madera
(559) 353-6400

Children's Hospital & Research Center
Oakland
(510) 428-3550

Lucile Salter Packard Children's Hospital at
Palo Alto
(650) 723-6858

Kaiser Permanente, No. Cal
Oakland
(510) 752-7703

Sutter Memorial Hospital
Sacramento
(916) 733-6023

UC Davis Medical Center
Sacramento
(916) 734-3112

UCSF Medical Center
San Francisco
(415) 476-2757

CCS Metabolic Centers in California

Southern California

Cedars-Sinai Medical Center
Los Angeles
(310) 423-9914

Children's Hospital of LA
Los Angeles
(323) 660-2450

Children's Hospital of Orange County
Orange
(714) 532-8852

Harbor/UCLA Medical Center
Torrance
(310) 222-3756

Kaiser Permanente, So. Cal.
Los Angeles
(323) 783-6970

Los Angeles County/ USC Medical Center
Los Angeles
(323) 226-3816

UCLA Medical Center
Los Angeles
(310) 206-6581

UC Irvine Medical Center
Orange
(714) 456-8513

UCSD/Rady
Children's Hospital
San Diego
(619) 543-7800

Glossary

Biotin - This vitamin is a member of the B vitamin complex family and plays an important role in metabolism of sugars, fats, and proteins within the human body.

Biotinidase - This enzyme is responsible for obtaining biotin (one of the necessary B vitamins) from protein in the diet. The enzyme also recycles the vitamin after being used by the body.

Carrier - A person who has a gene mutation in one of their genes that cause a disease, but does not have any symptoms of the disease themself. The mutation is often recessive, which means that both copies of the gene have to be mutated in order for disease symptoms to develop. Carriers are able to pass the mutation onto their children and therefore have an increased chance of having a child with the disease.

Enzyme - A special protein that helps chemical reactions take place. For example, enzymes in the stomach speed up the process of breaking down food. Each enzyme can participate in many chemical reactions without changing or being used up.

Gene - A segment of DNA that contains the instructions to make a specific protein (or part of a protein). Genes are contained on chromosomes. Chromosomes, and the genes on those chromosomes, are passed on from parent to child. Errors in the DNA that make up a gene are called mutations and can lead to diseases.

Supplement - A product taken orally that contains one or more ingredients (as vitamins or amino acids) that is intended to complete or make an addition to one's diet.

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If you have any questions, please call the person listed below.

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