Parent Fact Sheet

Biotinidase Deficiency

(Bio tin a daze De fish en see)

What is Biotinidase Deficiency?

Biotinidase deficiency is a metabolic disorder. This means the body has a chemical imbalance. This is a condition in which there is a deficiency of Biotin or vitamin B. Biotin is needed to release free biotin so the body is able break down fats, proteins, and carbohydrates. Biotinidase is inherited. It is present at birth.

What type of problems occurs with Biotinidase?

A baby who has this condition may have skin rashes, breathing problems, and seizures. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have Biotinidase?

This condition occurs in less than 1 in every 75,000 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of Biotinidase?

The treatment for biotinidase is biotin therapy. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care.

Where in Virginia can I take my baby for care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is http://www.vahealth.org/gns.

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