State of CT
Genetics Newborn Screening Program
Family Fact Sheet

BIOTINIDASE DEFICIENCY

Vitamins are an important part of our diet. They are needed to maintain a healthy, strong body. Babies who have the disorder called biotinidase deficiency (BYE-o-TIN-I-days) do not have enough free biotin in their body. Free biotin is needed to breakdown and use fats, carbohydrates, and proteins. Without enough free biotin, mental retardation, eye and ear problems, skin infections, and other physical problems may result.
Connecticut began screening newborns for biotinidase deficiency in 1993. In Connecticut, about 1 in every 73,387 newborn babies has biotinidase deficiency.

How does a baby get biotinidase deficiency?

Biotinidase deficiency is an inherited disorder. Both the mother and the father have an altered gene for biotin processing. When a child inherits both altered genes, one from each parent, the child is affected with biotinidase deficiency.

How is a baby tested for biotinidase deficiency?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State laboratory to screen for biotinidase deficiency. Abnormal screening results are reported to the Department of Public Health, Genetic Newborn Screening Tracking Unit nurse consultant. The DPH nurse consultant will notify the baby's primary care provider of the need for a prompt referral to one of the Genetic Regional Treatment Centers for confirmation testing and follow-up treatment if necessary.

What is the treatment for biotinidase deficiency?

The Regional Treatment Center specialists will prescribe treatment for biotinidase deficiency if necessary, which may include supplements of free biotin. Early diagnosis of biotinidase deficiency leads to early treatment, which is important for the health of your baby.

CONNECTICUT DEPARTMENT OF PUBLIC HEALTH

Keeping Connecticut Healthy

860-509-8081

The information provided is offered for general informational and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such. Contact your physician if there are any concerns or questions.

1/19/2005