

Profound Biotinidase Deficiency

General Overview

Q. What is biotinidase deficiency?

A. Biotinidase deficiency is a treatable disorder that affects the way the body recycles biotin, one of the B12 vitamins. Because this system doesn't work properly, the body has very low levels of biotin. This vitamin is necessary for many functions in the body. If left untreated, biotinidase deficiency can cause brain damage and other complications.

Q. How does the body normally process biotin?

A. The body gets biotin from the food we eat and conserves it by recycling. Biotin attaches to proteins during metabolism. Biotinidase is the name of the enzyme that separates biotin from proteins and allows the body to reuse it.

Q. What happens to biotin in a child with biotinidase deficiency?

A. When there is not enough biotinidase enzyme or it does not work properly, biotin cannot be freed into the form that is used by the body. This results in very low levels of biotin.

Q. What are the effects of having biotinidase deficiency if it is not treated?

A. Untreated biotinidase deficiency can lead to brain damage and, in rare cases, death.

Q. What is the treatment for biotinidase deficiency?

A. Biotinidase deficiency is easily treated with daily supplements of the biotin vitamin. To prevent problems, treatment must begin shortly after birth.

Q. Why would a child have biotinidase deficiency?

A. Biotinidase deficiency is an inherited disorder. It results when a baby receives a non-working biotinidase gene from each parent. For more information about this, contact your health care provider or a genetic counselor.

Q. How common is biotinidase deficiency?

A. About one in every 60,000 babies in the United States is born with biotinidase deficiency.

For more information about biotinidase deficiency, please see the Disorders section of our website:
www.doh.wa.gov/nbs.

