

BIOTINIDASE DEFICIENCY

What is Biotinidase Deficiency?

Biotinidase Deficiency is due to deficiency of an enzyme called biotinidase. Biotinidase helps in the production of biotin, which is a vitamin required by carboxylases (enzymes needed to change the food into energy). Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting and a skin rash. They may have mental retardation and brain damage as a complication. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

What causes Biotinidase Deficiency?

Due to a lack of biotinidase, children cannot effectively breakdown certain carbohydrates and fats which in turn are needed to breakdown protein.

The gene for the biotinidase enzyme is contained in the genetic material that is inherited from parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with biotinidase deficiency have one working and one non-working gene coding for this enzyme. They do not manifest the disease but can pass them on to their children. They are known as carriers. If the child inherits the non-working gene from both parents, he or she will have biotinidase deficiency.

What are the signs and symptoms of Biotinidase Deficiency?

Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting and a skin rash. They may have mental retardation and brain damage as a complication.

How do you diagnose Biotinidase Deficiency?

Newborn screening can identify biotinidase deficiency. Confirmatory diagnosis encompasses enzymatic and molecular testing approaches.

What is the treatment of Biotinidase Deficiency?

The main treatment of biotinidase deficiency is through dietary supplementation with biotin.