



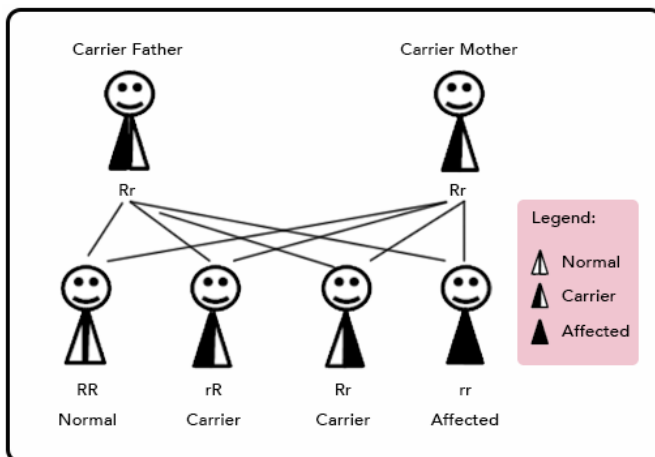
OTHERS: BIOTINIDASE DEFICIENCY

What is Biotinidase Deficiency?

Biotinidase Deficiency is due to a deficiency of an enzyme (or chemical scissors) called biotinidase. Biotinidase helps in the production of biotin. Biotin is a vitamin that is required by carboxylases which are other enzymes needed to change the food we eat 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?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition 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 we inherit from our 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. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Biotinidase Deficiency?

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BIOTINIDASE DEFICIENCY



WHAT IS THE TREATMENT OF BIOTINIDASE DEFICIENCY?

The main treatment of biotinidase deficiency is through dietary supplementation with biotin. Please consult with your attending physician prior to starting treatment.