Biotinidase Deficiency

What is Biotinidase Deficiency?

It is one of the vitamin responsive disorders due to a deficiency in one of the enzymes that are involved in recycling the biotin vitamin. This enzyme is called Biotinidase. Biotin, in turn, is important as an enzyme cofactor. Free biotin is needed to activate Carboxylase enzymes by binding at a specific site. Carboxylases are essential in the production of fats, carbohydrates and for the breakdown of proteins. This process is blocked if an individual has Biotinidase deficiency.

What causes the disease?

Mutations in *BTD* genes are a potential cause of the Biotinidase enzyme deficiency.

What are the clinical features of the disease?

The affected babies are usually normal at birth; however, they present symptoms in their first few months of life with poor feeding, vomiting, lethargy and intractable seizures. It is frequently accompanied by hearing loss, conjuctivites and visual problems, skin rash, alopecia. Later, ataxia and developmental delay became apparent if not treated.

How is the diagnosis confirmed?

The diagnosis of Biotinidase deficiency is confirmed by measuring the enzymatic activity in the blood sample and confirmed by finding specific urine organic acid and plasma acylcarnitine profiles. DNA molecular testing of *BTD* gene may help also in confirming the diagnosis.

What is the treatment of the disease?

Supplementation with biotin (10-20mg) can prevent symptoms of Biotinidase deficiency. A Biochemical genetics specialist should coordinate the treatment.