Climb™

Supporting those affected by Inherited Metabolic Disorders

# Biotinidase Deficiency

Biotinidase Deficiency is an inherited disorder in which the body is unable to process a vitamin called biotin. It is caused by a genetic fault in the BTD gene which provides instructions for the biotinidase enzyme. This enzyme is needed to separate biotin to allow it to be used by the body's metabolism, specifically for biotin-dependant carboxylase enzymes to break down fats, proteins and carbohydrates. Signs and symptoms typically appear within the first few months of life, although it can also become apparent later in childhood. There are two forms; partial Biotinidase Deficiency which without treatment causes low muscle tone, hair loss and rashes during periods of illness, infection or stress, and a more severe form called Profound Biotinidase Deficiency which also causes developmetal delay, seizures, breathing problems, hearing and sight loss, ataxia, and candidiasis. Symptoms can be improved or prevented with lifelong treatment.

## Synonyms

Alternative names for this condition are:

- BTD deficiency
- Infantile multiple carboxylase deficiency
- Juvenile multiple carboxylase deficiency
- Late-onset multiple carboxylase deficiency
- Multiple carboxylase deficiency, due to biotinidase deficiency

Further information about this condition is available from Climb.


## Disclaimer



Updated 20/07/17

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