

Climb National Information Centre for Metabolic Diseases



Biotinidase Deficiency

Other names used for this disorder are:

- BIOT
- BTD Deficiency
- Late Onset Biotin-Responsive Multiple Carboxylase Deficiency
- Late Onset Multiple Carboxylase Deficiency

Biotinidase deficiency is a rare inherited disorder which is characterised by the body's inability to recycle the vitamin biotin. Biotinidase recycles the biotin so the biotin can be used by various carboxylase enzymes that are important for the normal metabolism of fats, carbohydrates and proteins. The deficiency is caused by a defect in the biotinidase (BTD) gene. The role of this gene is to provide instructions to produce biotinidase, an enzyme needed for the body to reuse biotin. Biotin is found in certain foods, such as liver, egg yolks and milk, but these are in low quantities and may not be bioavailable. When there is a defect or mutation in the BTD gene, the activity of the biotinidase enzyme is reduced. When the activity is reduced to less than 10% of mean normal activity, the condition is known as profound biotinidase deficiency. When the activity is reduced to between 10% and 30% of mean normal activity, it is known as partial biotinidase deficiency. The gene that encodes biotinidase is located on the short arm of chromosome 3 (3p25).

This information has been kindly checked by Dr B. Wolf (USA) and is correct as of 02/05/13. This is a short internet summary only. For a full summary and further more detailed information please contact CLIMB by email, letter or telephone.

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This information about metabolic diseases is provided by Climb and is intended for educational purposes only. It should not be used for diagnostic or treatment purposes. Should you require more detailed information please contact Climb by email (info.svcs@climb.org.uk) or by telephone (0800 652 3181). For specific medical information regarding a particular disease or individual please contact your GP or Paediatrician.

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Climb is the only charity in the United Kingdom that provides support on all

Metabolic Diseases with links worldwide