Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Biotin-Responsive Basal Ganglia Disease

Biotin-Responsive Basal Ganglia Disease is caused by a defect in a gene which provides instructions for the thiamine transporter protein needed to carry the vitamin thiamine, obtained from food, into the cells. This disorder affects the nervous system and without lifelong management including biotin and thiamine treatment which can improve the condition it can lead to progressive neurological problems. Symptoms often occur in severe episodes and predominantly affect movement due to structures in the brain called the basal ganglia being affected. Episodes can be triggered by stress, illness, infection and other factors. Although the prevalence is unknown, this is a rare disorder often presenting in childhood. Seizures, confusion, movement problems and other neurological signs are often listed as presenting symptoms.

Synonyms

Alternative names for this condition are:

- BBGD
- Biotin-Thiamine-Responsive Basal Ganglia Disease
- BTBGD
- Thiamine Metabolism Dysfunction Syndrome 2
- Thiamine-Responsive Encephalopathy
- Thiamine Transporter-2 Deficiency
- THMD2

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

Updated 21/07/17

Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: www.climb.org.uk