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

Climb™

Supporting those affected by Inherited Metabolic Disorders

HSD10 Deficiency

In HSD10 Deficiency the body cannot process an amino acid called isoleucine, and certain fats. This is caused by a deficiency of an enzyme that is found in the mitochondria, the 'batteries' of the body's cells, responsible for producing energy. The enzyme is needed to break down isoleucine and a specific group of fats (BCFAs) and is also involved in chemical reactions involving certain hormones, and substances that regulate nervous system activity.

Synonyms

Alternative names for this condition are:

- 2-Methyl-3-Hydroxybutyric Aciduria
- 3H2MBD deficiency
- 3-hydroxy-2-methylbutyryl-CoA dehydrogenase deficiency
- Hydroxyacyl-CoA dehydrogenase II deficiency
- 2M3HBA
- 17 beta-hydroxysteroid dehydrogenase type 10 deficiency
- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- MHBD deficiency

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

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