Climb™

Supporting those affected by Inherited Metabolic Disorders

Beta-Ketothiolase Deficiency

Beta-Ketothiolase Deficiency is caused by a deficiency of an enzyme called ACAT1 which helps to process the amino acid isoleucine. The deficiency of ACAT1 means that the body is unable to properly break down fats and proteins causing a build up of compounds which become toxic and damage the tissues and organs. Onset usually begins between the ages of 5 months to 2 years. This disorder is characterised by episodes of vomiting, dehydration, lethargy, low muscle tone, breathing problems and sometimes seizures. Episodes may be caused by certain triggers including long periods without food, stress, acute illness, infection, or an increased intake of protein in the diet.

Alternative names for this condition are:

- 2-alpha-methyl-3-hydroxybutyricacidemia
- 3-alpha-ketothiolase deficiency
- 3-alpha-ktd deficiency
- 3-alpha-oxothiolase deficiency
- 3-Ketothiolase deficiency
- 3-Methylhydroxybutyric acidemia
- Alpha-Methylacetoacetic aciduria
- BKT
- MAT deficiency
- Mitochondrial 2-methylacetoacetyl-CoA thiolase deficiency - potassium stimulated
- Mitochondrial acetoacetyl-CoA thiolase deficiency
- T2 deficiency

Synonyms

Further information about this condition is available from Climb.

Disclaimer

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