Climb™

Supporting those affected by Inherited Metabolic Disorders

HMG-CoA Lyase Deficiency

Also known as **3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency**

HMG-CoA Lyase Deficiency belongs to a group known as the Organic Acidurias. In HMG-CoA Lyase Deficiency there is a defect in the HMGCL gene which provides instructions for producing the HMG-CoA Lyase enzyme. This enzyme is needed to break down a specific protein (amino acid) called leucine and in the production of ketones during the break down of fats. Certain organs and tissues, including the brain use ketones for energy when glucose is not available. Therefore, ketones are extremely important for when a person goes a long period of time without food.

When leucine is not broken down properly it causes a build-up of organic acids in the blood (metabolic acidosis) and the lack of ketones can cause low blood sugar levels. Both problems can result in severe illness. In some cases, this disorder can be misdiagnosed as Reye Syndrome.

Synonyms

Alternative names for this condition are:

- Hydroxymethylglutaric Aciduria
- 3-OH 3-CH3 Glutaric Aciduria

Further information about this condition is available from Climb.

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