Climb™

Supporting those affected by Inherited Metabolic Disorders

Carnitine Palmitoyltransferase I Deficiency

Carnitine Palmitoyltransferase 1A, which is found in the liver, is an enzyme that is essential in the process of breaking down certain fats and storing them as energy. It is needed to attach carnitine to long-chain fatty acids so they are able to enter the mitochondria which is the powerhouse of cells, producing energy. A deficiency of the enzyme means that these fatty acids cannot enter the mitochondria, leading to a reduction in cell energy and a build up of fatty acids in the cells causing damage to the liver and other organs. Symptoms usually begin in childhood and include hypoketotic hypoglycaemia which means there are low levels of ketones and sugar in the blood. Symptoms can be triggered by illness, infection or long periods without food. Other symptoms include an enlarged liver and liver dysfunction, and high levels of carnitine in the blood.

Synonyms

Alternative names for this condition are:

- CPT 1 Deficiency
- CPT I Deficiency
- CPT 1A Deficiency

Further information about this condition is available from Climb.

Disclaimer

Updated 28/07/17

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