



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



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

[Please read our disclaimer and information on data protection.](#)



Updated 28/07/17

Telephone: **0845 241 2173**  
Email: [contact@climb.org.uk](mailto:contact@climb.org.uk)

Freephone: **0800 652 3181**  
Website: [www.climb.org.uk](http://www.climb.org.uk)

Children Living with Inherited Metabolic Diseases is a charity registered in England and Wales (1089588) in Scotland (SC044634) and a Company Limited by Guarantee 4267454



ClimbHQ



ClimbHQ



TeamClimb