Climb™

Supporting those affected by Inherited Metabolic Disorders

Carnitine-Acylcarnitine Translocase Deficiency

Carnitine-Acylcarnitine Translocase (CACT) is an enzyme that is essential in the process of breaking down certain fats and storing them as energy. A deficiency of this enzyme means the body is unable to use these fats properly especially during long periods without food. Most cases have a severe form of CACT Deficiency which presents within the first 48 hours of life with hypoketotic hypoglycaemia which means there are low levels of ketones and sugar in the blood. Other symptoms include breathing difficulties, seizures, high blood ammonia levels, skeletal muscle damage, enlarged liver, weakened heart muscle, developmental delay, and an irregular heartbeat. The milder form is rare and is characterised by episodes of hypoketotic hyperglycaemia triggered by illness, infection and long periods without food.

Synonyms

Alternative names for this condition are:

- CACT Deficiency
- Carnitine-Acylcarnitine Carrier Deficiency

Further information about this condition is available from Climb.

Disclaimer

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

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