Climb™

Supporting those affected by Inherited Metabolic Disorders

Carnitine Palmitoyltransferase II Deficiency

Carnitine Palmitoyltransferase 2 (CPT2), 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. Carnitine needs to be attached to long-chain fatty acids so they are able to enter the mitochondria which is the powerhouse of cells, producing energy. Once inside the mitochondria the CPT2 enzyme removes the carnitine so they are ready to be converted. A deficiency of the enzyme means that these fatty acids cannot be used to provide energy. This leads to a reduction in cell energy and a build up of fatty acids in the mitochondria causing damage to the liver and other organs. There are 3 forms of CPT II deficiency: a myopathic form, a severe infantile form and a neonatal form. The neonatal form is extremely rare and is the most severe form. The myopathic is the least severe and the most common form with early diagnosis and good management can have a good prognosis.

Synonyms

Alternative names for this condition are:

- CPT 2 Deficiency
- CPT II Deficiency

Further information about this condition is available from Climb.

Disclaimer

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