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

Climb™

Supporting those affected by Inherited Metabolic Disorders

Carbamoyl Phosphate Synthetase I Deficiency

CPS I Deficiency is a Urea Cycle Disorder, in which the body's ability to manage dietary protein is impaired. When the body digests protein it is broken down into small molecules known as amino acids. Excess amounts of these amino acids are converted into a toxic substance known as ammonia. In the liver, the ammonia is converted to urea and excreted in the urine. In Urea Cycle Disorders the ammonia is not removed from the blood stream and accumulates (hyperammonaemia), becoming toxic. CPS I Deficiency is caused by a deficiency or absence of the enzyme carbamoyl phosphate synthetase which is involved in the first step of the Urea Cycle. Signs of hyperammonaemia include lethargy, poor breathing, irregular body temperature, refusal to feed, vomiting, unusual body movements and postures, seizures and, if left untreated, coma.

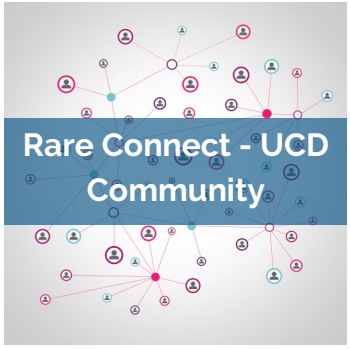
Synonyms

Alternative names for this condition are:

- CPS I Deficiency

Further information about this condition is available from Climb.

Disclaimer



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