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

Climb™

Supporting those affected by Inherited Metabolic Disorders

Ornithine Transcarbamylase Deficiency

This disorder belongs to a group of conditions known as the urea cycle disorders. If we eat an excess of proteins (amino acids), the body converts these amino acids into ammonia. In large amounts this is toxic to the body and so the liver, through a number of enzymes, converts it into urea, so the body can excrete it in the urine. In this disorder there is a deficiency or absence of the enzyme ornithine transcarbamylase which is found in the liver cells and this prevents the ammonia from being converted into urea. This leads to a build up of ammonia in the body and causes the symptoms of this disorder.

The symptoms, severity and age of onset varies from one individual to another. A high level of ammonia in the blood causes swelling of the cells in the brain which leads to a progressive loss of function. Early signs of high ammonia in newborns include poor feeding, lethargy, irritability, rapid breathing, vomiting, altered muscle tone, seizures and/or a temporary cessation in breathing. Early signs in infancy include failure to grow or gain weight, vomiting, intolerance of food containing protein, irritability, drowsiness and/or developmental delay. The disorder can also present in childhood with drowsiness, vomiting, lethargy, confusion, headache, unsteadiness, speech problems, behavioural problems, a fluctuating level of consciousness, stroke, seizures and if left untreated can progress to coma. Management of this condition is lifelong.

Synonyms

Alternative names for this condition are:

- Hyperammonaemia Type II
- Hyperammonaemia due to Ornithine Transcarbamylase Deficiency
- OTC Deficiency
- Ornithine Carbamoyl Transferase Deficiency

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

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