Ornithine Transcarbamylase Deficiency

Other names that this condition is known by are:
- Hyperammonaemia Type II
- Hyperammonaemia due to Ornithine Transcarbamylase Deficiency
- OTC Deficiency
- Ornithine Carbamoyl Transferase Deficiency

This disorder belongs to a group of conditions known as 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 and this prevents the ammonia from being converted into urea. This leads to a build up of ammonia in the body (hyperammonemia) and causes the symptoms of this disorder.

Treatment of an acute episode includes restricting any further intake of protein to prevent any additional increase in ammonia levels. Individuals are placed on a low protein, high calorie diet with amino acid supplements. This can be achieved by using special food formulas. Glucose is usually given to prevent the breakdown of any stored protein that would increase ammonia levels further. If levels are really high then medications and possibly dialysis may be needed to help reduce them. Prompt and aggressive treatment is needed during episodes of high levels of ammonia or increased lethargy and severe vomiting.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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