Urea Cycle Disorders

The Urea Cycle Disorders are a group of genetic disorders caused by a deficiency of one of the six enzymes in the urea cycle that is responsible for the removal of ammonia from the blood stream. When the body digests protein it is broken down into small molecules known as amino acids. These travel through the blood stream and are transported to the cells. Excess amounts of these amino acids are converted into a toxic substance known as ammonia. Once the ammonia is in the liver, the ammonia is converted to urea and excreted in the urine. In urea cycle disorders one of the six enzymes does not function properly and ammonia is not removed from the blood stream.

There are six urea cycle disorders:
- Arginase Deficiency (AG)
- Argininosuccinic Aciduria (ASA)
- Carbamyl Phosphate Synthetase Deficiency (CPS)
- Citrullinaemia
- N-Acetylglutamate Synthetase Deficiency (NAGS)
- Ornithine Transcarbamylase Deficiency (OTC)

Individuals are treated with an emergency regime when they are unwell and/or unable to tolerate a normal diet, this is usually given as a high glucose energy drink such as Maxijul. The emergency regimen is prescribed to suit the individual. If the individual continues to vomit and appears not to be recovering, it is necessary to contact the doctor, take them to casualty or contact the paediatric department. If this happens, a glucose solution is given directly through the vein; medications (sodium phenylbutyrate and sodium benzoate) may be used and in some cases Arginine. If this does not appear to be working dialysis may be needed to help reduce levels of ammonia. Long term treatment involves dietary restrictions of protein. Feeding tubes may be used to aid feeding in some individuals. Frequent blood tests are required to monitor the disorder. Amino acid formulas and multivitamins and calcium supplements may be used.

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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Climb is the only charity in the United Kingdom that provides support on all Metabolic Diseases with links worldwide

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