Hartnup Disease

Other names that may be used for this disorder are:

- Hartnup Disorder
- Hartnup Syndrome
- Hart Syndrome
- H Disease
- HND
- Pellagra-Cerebellar-Ataxia-Renal Aminoaciduria Syndrome
- Trypophan Pyrrolase Deficiency

Hartnup Disease is a rare disorder caused by an inborn error of amino acid metabolism. A defect in tryptophan, which is a coded amino acid essential for nutrition, impairs the body's ability to break down and transport amino acids through the intestines. Hartnup Disease is caused by a defective gene, which has been located on the long arm of chromosome 11. In some cases the defective gene may be found on the short arm of chromosome 5, this gene is known as the SLC9A19 gene, which encodes transport for sodium-dependant amino acids.

Diet plays a major role in the treatment of this condition, good nutrition must be maintained. Those affected should avoid sunlight and sulphonamide drugs and take supplements of nicotinamide or niacin. Treatment may also include supplements of an oral drug called L-tryptophan ethyl. Genetic counselling may be of benefit to those affected by this condition.

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

Disclaimer

This information about metabolic diseases is provided by Climb and is intended for educational purposes only. It should not be used for diagnostic or treatment purposes. Should you require more detailed information please contact Climb by email (ir.svcs@climb.org.uk) or by telephone (0800 652 3181). For specific medical information regarding a particular disease or individual please contact your GP or Paediatrician. Climb accepts no responsibility for any errors or omissions nor does Climb assume any liability of any kind for the content of any information contained within this summary or any use that you may make of it.