

Climb National Information Centre for Metabolic Diseases



Homocystinuria

This condition belongs to a group known as the amino acid disorders. This is where there is an absence or a deficiency in an enzyme that is needed to breakdown proteins (amino acids), which prevents the body from using them for growth and repair. The amino acid methionine is converted into homocysteine, which in turn is converted into cysteine. In this disease there is a deficiency or an absence in an enzyme called cystathionine beta-synthase (CBS) that is required to breakdown homocysteine into cysteine. This leads to a build up of homocysteine in the body, especially the blood (homocysteinaemia) and the urine (homocystinuria). In addition there are also increased levels of methionine.

In some individuals treatment of this disorder includes giving large doses of vitamin B6 (pyridoxine), which helps change homocysteine into cysteine. Other individuals do not respond to this supplement and the condition is best controlled by diet. This involves restricting the amino acid methionine and giving cysteine supplements. In addition vitamin B12, folic acid and betaine may be of benefit. Other medications can provide relief from symptoms. It is advised that individuals do not use oral contraceptives.

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
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