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Climb™

Supporting those affected by Inherited Metabolic Disorders

# 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 amino acids, which prevents the body from using them for growth and repair. Proteins are made of chains of amino acids. One of these amino acids is methionine which is broken down into homocysteine then cysteine from which sulphur is eliminated. In this condition there is a defect in an enzyme known as cystathionine beta-synthase (CBS) which stops homocysteine from being broken down.

Homocystinuria is now routinely tested for shortly after birth as part of the Newborn Bloodspot Screening Programme. If Homocystinuria is diagnosed, treatment is started immediately using diet and medication. Some individuals are responsive to vitamin B6 (Pyridoxine) - treatment is lifelong. Newborn screening and early management means that most children with Homocystinuria are now able to live normal, healthy lives. Without lifelong treatment symptoms such as sight problems, brain damage, skeletal and joint problems including osteoporosis, and an increased risk of blood clots and strokes can occur.

## Synonyms

Alternative names for this condition are:

- Classical HCU
- Cystathionine Beta-Synthase (CBS) Deficiency
- HCU


Further information about this condition is available from Climb.

## Disclaimer

Please read our disclaimer and information on data protection.

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