Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Cystinuria

Cystinuria is a metabolic disorder characterised by the abnormal reabsorption of certain amino acids in the kidneys and small intestines. These amino acids include cystine, lysine, arginine, and ornithine. At urine physiological pH, cystine is not soluble; this causes abnormally high amounts of undissolved cystine in the urine (cystinuria) to crystallise and cause the formation of stones within the kidneys, ureters or bladder. Cystine stones are amber in colour and can progress to larger crystals which take on the shape of the chambers within the kidney. The other three amino acids do not contribute to stones formation.

Cystinuria affects approximately 1:15,000 of the population and accounts for 10% of all kidney stones in children and 0.5-1.0% in adults. It has a varying severity; severe, where stones formed at an early age, moderate, which first presents in middle-age and mild which patients never form kidney stones at all.

Synonyms

Alternative names for this condition are:

- Cystathionase Deficiency
- Cystathione Gamma-Lyase Deficiency Syndrome
- Gamma-cystathionase deficiency

Further information about this condition is available from Climb.

Disclaimer

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