Cystinosis

In Cystinosis there is a problem transporting an amino acid called cystine. Amino acids are broken down in a part of the cells called the lysosome, which enables them to be recycled by the body. Once broken down, cystine is normally transported out of the lysosome. However, in Cystinosis a transporter protein is missing and the body is unable to move the cystine, causing it to accumulate and form crystals within the lysosomes. This primarily damages the kidneys and the eyes, but may also affect other organs and systems. There are three forms of Cystinosis. Infantile Nephropathic Cystinosis is the most severe form, characterised by poor growth, excessive thirst, and frequent urination. One of the major complications is renal tubular Fanconi Syndrome, in which the kidneys are unable to reabsorb nutrients and minerals and they are instead lost in the urine. Other forms include intermediate cystinosis, which has similar symptoms occurring later and non-nephropathic/ocular cystinosis which usually only affects the eyes. Cystinosis affects between 1:100,000 and 1:200,000 and only 2-3 babies are born in the UK per year with this condition.

Further information about this condition is available from Climb.

Disclaimer
Please read our disclaimer and information on data protection.