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Growth retardation in children with cystinosis.

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Abstract

Cystinosis is a rare autosomal recessive disorder characterized by the intralysosomal accumulation of cystine in all tissues due to mutations in the CTNS gene (17p13.3). Infantile nephropatic cystinosis is the most severe and the most frequent form of the disease. It causes renal Fanconi syndrome, leading to end stage renal failure around the age of 10 years if left untreated. Cystine accumulation also affects the eyes, muscles, central nervous system and various endocrine organs. Children with cystinosis often suffer from growth retardation, which is multifactorial in origin. It is not only caused by a decreased renal function, but is aggravated by a poor metabolic status due to renal Fanconi syndrome, pronounced feeding difficulties, often requiring tube feeding and possibly cystine accumulation in the bone. Longitudinal growth can be improved by the correction of metabolic and nutritional deficits and by the treatment of hypothyroidism. The cystine depleting drug cysteamine slows down the progression of renal disease, protects extra-renal organs, accelerates growth and therefore should be administered as early as possible. Despite these treatment strategies, growth retardation remains a severe complication in cystinosis patients and frequently requires the administration of recombinant human growth hormone.