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Nephropathic cystinosis: late complications of a multisystemic disease.

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Abstract

Cystinosis is a rare autosomal recessive disorder due to impaired transport of cystine out of cellular lysosomes. Its estimated incidence is 1 in 100,000 live births. End-stage renal disease (ESRD) is the most prominent feature of cystinosis and, along with dehydration and electrolyte imbalance due to renal tubular Fanconi syndrome, has accounted for the bulk of deaths from this disorder. Prior to renal transplantation and cystine-depleting therapy with cysteamine for children with nephropathic cystinosis, their lifespan was approximately 10 years. Now, cystinotic patients have survived through their fifth decade, but the unremitting accumulation of cystine has created significant non-renal morbidity and mortality. In this article we review the classic presentation of nephropathic cystinosis and the natural history, diagnosis, and treatment of the disorder's systemic involvement. We also emphasize the role of oral cysteamine therapy in preventing the late complications of cystinosis.

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