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Corneal crystals in nephropathic cystinosis: natural history and treatment with cysteamine eyedrops.

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Abstract

Although renal disease is the most prominent feature of the lysosomal storage disease cystinosis, corneal cystine crystal formation remains a major complication, leading to photophobia, corneal erosions, and keratopathies. Moreover, the extent of corneal crystal accumulation reflects the course and severity of the disease itself, and the cornea is accessible to direct examination. Therefore, we employed a scoring system, based on a library of slit-lamp photographs of corneas with increasing crystal densities (0.00-3.00), to assess the degree of crystal accumulation in 170 patients with nephropathic cystinosis examined at the National Institutes of Health between 1976 and 2000. None of the patients had received topical cystine-depleting therapy at the time of the evaluation. In this natural history study, infants in the first year of life had absent or minimal corneal crystals, i.e., a corneal cystine crystal score (CCCS) of 0 or 0.25. However, the CCCS increased linearly with age, such that every patient had visible crystals by 16 months of age, and plateaued at approximately 3.00 by early adolescence. Longitudinal studies in representative patients support the cross-sectional results. Individuals homozygous for the common 57-kb deletion involving the cystinosis gene (CTNS) displayed the same course of corneal crystal accumulation as did individuals not bearing the large deletion. Patients with ocular or nonnephropathic cystinosis had CCCSs that were, in general, half those expected for patients with nephropathic cystinosis of the same age. Administration of 0.55% cysteamine eyedrops, given 6 to 12 times per day, dissolved corneal cystine crystals in 10 representative patients with nephropathic cystinosis aged 1 to 32 years within 8 to 41 months.

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