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Clinical Practice: A Proposed Standardized Ophthalmological Assessment for Patients with Cystinosis.

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Abstract

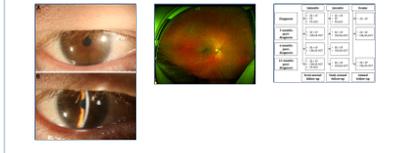
Cystinosis is a rare autosomal recessive disease with an incidence of approximately 1 case per 100,000-200,000 live births. Over the years, gaining in-depth knowledge of the disease has led to vast improvement in patient life expectancy. However, debilitating, extra-renal manifestations such as eye disease, in particular corneal crystal deposition and its associated photophobia, still occur frequently, regardless of patient age and notwithstanding the increased implementation of systemic therapy. Ophthalmological assessment has not yet been standardized. The aim of this article was to provide clear recommendations for ophthalmological assessment during follow-up of patients with cystinosis to improve quality and regularity of ophthalmological care and thereby minimize ophthalmological complications. A literature search was performed to assess previous and current recommendations on examinations to conduct during follow-up of patients with cystinosis. Multidisciplinary cystinosis clinics were set up in collaboration with the Department of Ophthalmology and the Department of Pediatric Nephrology to allow patients to be seen by a nephrologist, an ophthalmologist and other specialists on the same day. Based on the results of these multidisciplinary clinics the standardized clinical ophthalmological assessment was drafted. This is a protocol for follow-up, describing the approach taken regarding ophthalmological follow-up of patients with cystinosis, considering the different types of the disease and the time since diagnosis. Standard examination includes history, visual acuity, tonometry and slit-lamp examination, with fundus photography performed at diagnosis and annually thereafter. Confocal microscopy is the imaging modality of choice, while anterior segment optical coherence tomography (OCT) is a good alternative. Finally, posterior segment OCT for imaging of the macular region and optic nerve should be conducted on an annual basis.

KEYWORDS: Corneal crystal load; Cystinosis; Follow-up; Ophthalmological complications; Treatment

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