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Latest Clinical Approaches in the Ocular Management of Cystinosis: A Review of Current Practice and Opinion from the Ophthalmology Cystinosis Forum.

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Abstract

Cystinosis, a rare autosomal recessive disease caused by intracellular cystine accumulation, occurs in an estimated 1/100,000-200,000 live births. Ocular non-nephropathic cystinosis is typically diagnosed during adulthood, when patients present with corneal crystal deposition and no systemic involvement. Due to the rarity of the condition, diagnosis is often delayed and can have a significant impact on the overall prognosis of the disease. Early diagnosis is therefore imperative to ensure successful treatment and improve quality of life, as most of its clinical manifestations can be prevented or delayed. Early detection strategies and practical approaches for the ocular management of cystinosis were discussed during the Ophthalmology Cystinosis Forum, a 1-day meeting held in Berlin, Germany during June 2017. Recommendations for early detection comprise ophthalmic assessment, including self- and clinician-assessed recording of photophobia, and visual acuity, slit-lamp examination and tonometry ophthalmic examinations. In vivo confocal microscopy and anterior segment optical coherence tomography were highlighted as valuable techniques in evaluating cystine crystals in the cornea, in vivo and non-invasively. The mainstay of ocular cystinosis treatment is the cystine-depleting aminothiol cysteamine. Indeed, early treatment with and strict adherence to cysteamine therapy has a considerable impact on the long-term prognosis of ocular cystinosis. In rare diseases such as ocular cystinosis, standardised guidelines and recommendations for detection, patient care and follow-up assessments are essential. Such guidelines provide a support tool for healthcare professionals caring for ocular cystinosis patients. Multidisciplinary teams (MDTs) are essential for delivering gold standard care and improving quality of life for patients and their families. This review paper highlights current early detection policies, clinical treatment strategies and practical approaches for the ocular management of cystinosis, including implementing a cystinosis MDT. Additionally, discussions of the Ophthalmology Cystinosis Forum held in 2017 are summarised. FUNDING: Orphan Europe. Plain language summary available for this article.

KEYWORDS: Infantile nephropathic cystinosis; Juvenile nephropathic cystinosis; Multidisciplinary team; Ocular cystinosis

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