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Int Ophthalmol. 2018 Jun 18. doi: 10.1007/s10792-018-0954-7. [Epub ahead of print]

Ocular changes in nephropathic cystinosis: The course of the gold-dust.

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Abstract

PURPOSE: Cystinosis is an autosomal recessive inherited lysosomal storage disease with an incidence of 1:100.000 up to 1:200.000 caused by a gene mutation of a lysosomal transport protein resulting in deposition of cystine in lysosomes in all cells and tissues. In the cornea, crystalline, gold-dust deposition of cystine leads to visual impairment, recurrent erosions, photophobia, epiphora and blepharospasmus. Standard therapy is topical and systemic application of cysteamine which may resolve the accumulated cystine crystals.

PATIENT AND METHODS: This is a case report of a thirty-one-year-old patient who already underwent renal transplantation because of nephropathic cystinosis. Visual impairment by cystine crystal deposition was aggravated by a central avascular pannus formation in his right eye. Penetrating keratoplasty was performed in intention to improve the patient's visual acuity and life quality.

RESULTS: After penetrating keratoplasty in the right eye, there was only a slight visual improvement. OCT scans of the macula revealed intraretinal cystine crystals and a cystoid macular edema, which was treated with a bevacizumab injection. Transmission electron microscopy of the excised cornea revealed spiky intracorneal inclusions and confocal in vivo microscopy of the left eye allowed detailed visualization of the cystine crystal deposition.

CONCLUSIONS: There is a variability of ocular manifestations of nephropathic cystinosis. Ophthalmologists have a central role in the early diagnosis of cystinosis as mostly the first manifestation are cystine crystals in the cornea. Penetrating keratoplasty may be one of the therapeutical options. Nevertheless, the patient has to be informed about the limited prognosis because of the persisting underlying disease.

KEYWORDS: Cornea; Cystine crystals; Cystinosis; Keratoplasty; Nephropathic cystinosis

PMID: 29916124 DOI: 10.1007/s10792-018-0954-7