IMAGE

Crystalline keratopathy in nephropathic cystinosis

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A 3-year-old male child presented with complaints of poor weight gain, delayed motor milestones since 1 year of age and features suggestive of rickets (wrist widening, bowing of legs and Harrison's sulcus). He was third born of a third degree consanguineous marriage, had polyuria, with investigations revealing proximal renal tubular acidosis and generalised aminoaciduria (Fanconi syndrome). Eye evaluation revealed both the cornea to be studded with yellow-golden crystals throughout their surface. The crystals appeared to occupy the epithelial and superficial stromal layers of the cornea, and were distributed in a generalised fashion (Figure 1). Fundus evaluation of both the eyes too showed crystalline deposits within the retina (not cooperative for fundus imaging). Genetic studies confirmed the diagnosis of cystinosis with pathogenic *CTNS* gene mutations on intron 9 and exon 11. Younger sibling aged 2 years had a similar clinical presentation, and his genetic studies also determined the same pathogenic *CTNS* gene mutations.

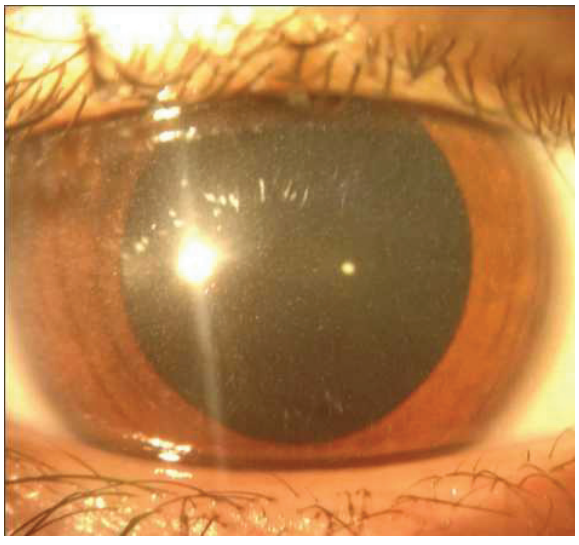

Figure 1. Crystalline keratopathy in the right eye. Cystine deposition can be visualized as superficial and shiny crystals involving the entire corneal surface.

Cystinosis is a rare autosomal recessive metabolic disorder characterised by accumulation of cystine in lysosomes. Kidneys and eyes are, especially, vulnerable though eventually the disease impacts all the organs including liver, muscles, white blood cells and central nervous system. *CTNS*, located on the short arm of the chromosome 17 (p13), is the responsible gene. It encodes the lysosomal cystine carrier cystinosin [1,2]. There are three clinical forms of cystinosis-infantile (nephropathic) cystinosis; late-onset cystinosis and benign cystinosis. Infantile cystinosis is the most severe and the most common type of cystinosis, but may be missed initially as all

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signs of renal Fanconi syndrome may not be present during the first months of life. Children with nephropathic cystinosis develop symptoms of polyuria, failure to thrive and short stature with features of rickets (due to high loss of phosphorous) by first year of life. The corner stone of diagnosis is an elevated leucocyte cystine content. The final confirmation is by molecular analysis of the *CTNS* gene. Lifelong treatment with the cysteamine, a cystine depleting drug, should be initiated as early as possible to prolong renal function and survival, and protect extra-renal organs.

Presence of corneal cystine crystals in patients with cystinosis after the age of 1 year is also considered pathognomic for cystinosis. Corneal involvement is asymptomatic initially, but an unabated natural course of cystinosis leads to blepharospasm, corneal erosions and superficial punctate keratopathy. Intense photophobia and lacrimation are the rule in advanced cases. Involvement of the retina causes irreversible loss of vision later in life [1,2]. While systemic therapy with cysteamine may help in keeping crystalline retinopathy under check, it has suboptimal influence on the crystalline deposits within the avascular cornea. Topical cysteamine eye drops 6–12 times per day have been used, but remain a challenging treatment because of frequency of instillation, low shelf life, poor drug availability and cost related issues [3].

CONFLICT OF INTEREST

None of the authors has any conflict of interest to disclose.

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None.

ETHICAL APPROVAL

The authors declare that ethics approval was not required for this case report. Signed informed consent for publication of medical details has been obtained from the parents of the child. Confidentiality was ensured at all stages.

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