ISSN: 2577 - 8005

Case Report

Medical & Clinical Research

Cystinosis in a routine ophthalmologic evaluation

Jorge Alberto Amorim Oliveira[1], Rodrigo Jorge Amorim[1], Mariana Xavier Araujo de Oliveira[1], Juliane de Carvalho Rocha Moura[1], Mateus Conciani[1], Thassia Kauany Feitosa da Silva[1], Thales Antonio Abra de Paula[1,2*]

[1]University of Santo Amaro (UNISA), Ophthalmology, Sao Paulo, Brazil.

[2]University of São Paulo (USP), Ophthalmology, Medical School, Sao Paulo, Brazil

[*]Corresponding Author
Thales AA. de Paula, University of Santo Amaro UNISA, Ophthalmology, São Paulo, Brazil.

Submitted:23 Feb 2023;**Accepted**: 28 Feb 2023;**Published**:12 Mar 2023

Citation: *Jorge Alberto Amorim Oliveira, Rodrigo Jorge Amorim, Mariana Xavier Araujo de Oliveira, Juliane de Carvalho Rocha Moura, Mateus Conciani, et al. (2023) Cystinosis in a routine ophthalmologic evaluation. Medical & Clinical Research 8(3):01-04.*

Abstract

Introduction: *Cystinosis is a rare autosomal recessive metabolic disease. It occurs due to a chromosomal mutation that allows the accumulation of this amino acid in lysosomes. Cystine can form crystals in various tissues such as the kidneys, pancreas, thyroid, testicles, bone marrow, brain, eye, and muscles. In the eyes, crystals can cause deposits on the cornea and conjunctiva. In the cornea, deposits occur in the periphery and anterior stroma. As the disease progresses, the crystals move centripetally and posteriorly. Oral and topical treatments with cysteamine have been described. The ophthalmologic solution of cysteamine 0.55% used continuously every 2 hours has shown good results.*

Clinical Case: *Patient in a routine ophthalmologic exam, with findings of cystine crystals in the cornea and past kidney transplantation without diagnosis.*

Discussion: *The patient's past history is relevant and corroborates with the ophthalmological findings. The patient had severe kidney disease, requiring kidney transplantation, but the disease was not elucidated. Therefore, the ophthalmological examination was fundamental for the diagnosis of the case*

Keywords: Cystinosis, Cornea, Crystals, Densitometry, Kidney disease

Introduction

Cystinosis is a rare autosomal recessive metabolic disease, with an incidence ranging from 1:100,000 to 1:200,000. It occurs by chromosomal mutation, in which a defect in the active transport of cystine across the lysosomal membrane allows the accumulation of this amino acid in lysosomes [1]. As a result, cystine can form crystals in various tissues such as the kidneys, pancreas, thyroid, testicles, bone marrow, brain, eye, and muscles [2].

Three phenotypic forms of cystinosis are described: Infantile nephropathic form-more common and more severe, usually associated with Fanconi syndrome and need for kidney transplantation in the first decade of life; Juvenile nephropathic form-later onset and less severe; Non-nephropathic form-more currently called ocular cystinosis, benign in character, usually with isolated, asymptomatic ophthalmologic findings, and may present from adolescence to the fifth decade of life [3].

The ocular symptoms of cystinosis result from the deposition of cystine crystals. The crystals are deposited mainly on the cornea and conjunctiva. In the cornea, the deposits are located in the periphery and anterior stroma, and as the disease progresses, the crystals move centripetally posteriorly [2]. The deposits may also affect the corneal epithelium, which may be related to the foreign body sensation reported by some patients [6]. The deposits become increasingly dense without proper treatment, causing photophobia and worsening of visual acuity [3,5].

Cystinosis may present with other ocular manifestations such as retinitis pigmentosa, macula ridge deposits, posterior syncytia, glaucoma, and hemorrhagic retinopathy. In all cases, there is a risk of worsening visual acuity [1]. Ocular complications may be associated with the progression of the systemic disease, so regular and multidisciplinary follow-up is important.

Treatment should be done with oral and topical cysteamine. The ocular treatment can be done with cysteamine solution 0.55% (used continuously 02/02h). Currently, the use of this solution has shown good results, and therefore it should be prescribed in all cases of cystinosis with ocular involvement [1].

Clinical Case

A 26-year-old female patient went for a routine ophthalmological evaluation, wishing to change her glasses. In the refractometry

exam she presented a visual acuity of 20/20, with the following correction OD: -0.50 DE; OE: -1.25-0.25 90°. The biomicroscopic examination observed in both eyes (BE): mild blepharitis, diffuse stromal crystal deposits throughout the corneal stroma (Figures 1A,B) Retinal mapping without alterations in both eyes. Complementary exam: corneal densitometry, performed by pentacam, showing higher density mainly in the anterior third of the cornea (up to 120 microns) BE. The graph also shows decreasing density in the anterior to posterior direction in relation to corneal thickness (Figures 2 A,B).

About 7 years ago the patient had constitutional symptoms with weight loss and anorexia, reason why she sought medical help where she was found to have difficult to control anemia and deteriorated renal function. At the same time she underwent dialysis for 4 months and was put on the kidney transplant waiting list. One year after the onset of the condition, she underwent a kidney transplant, with her sister as a compatible donor. No renal biopsy or further investigations about the type of pathology were performed.

After ophthalmological examination and oriented to seek a new evaluation with the nephrologist to report ophthalmological findings compatible with cystinosis.

The patient is currently awaiting an appointment for further clinical evaluation because she has lost the follow-up with the nephrologist who was following her case.

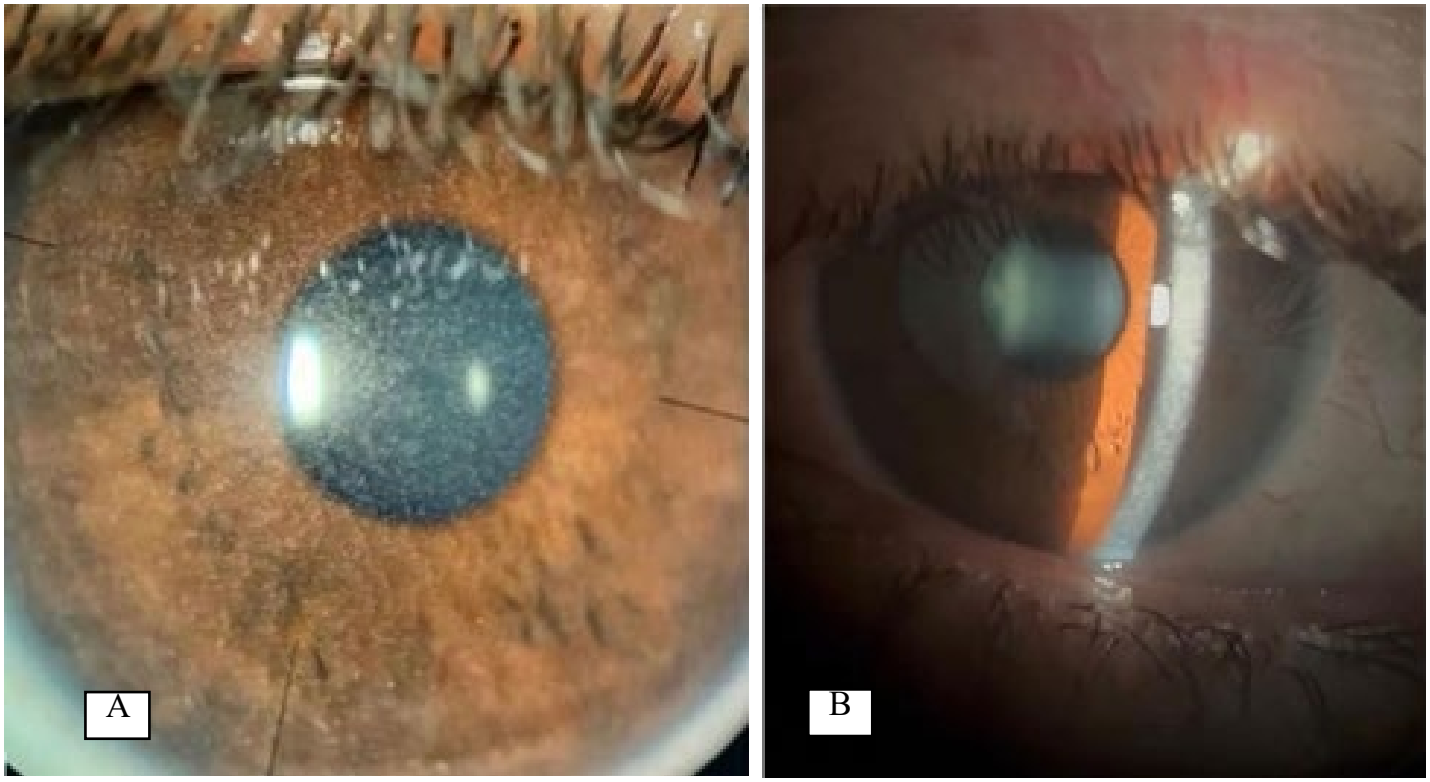

Figure 1 A,B: Biomicroscopy showing crystal deposits on the cornea.

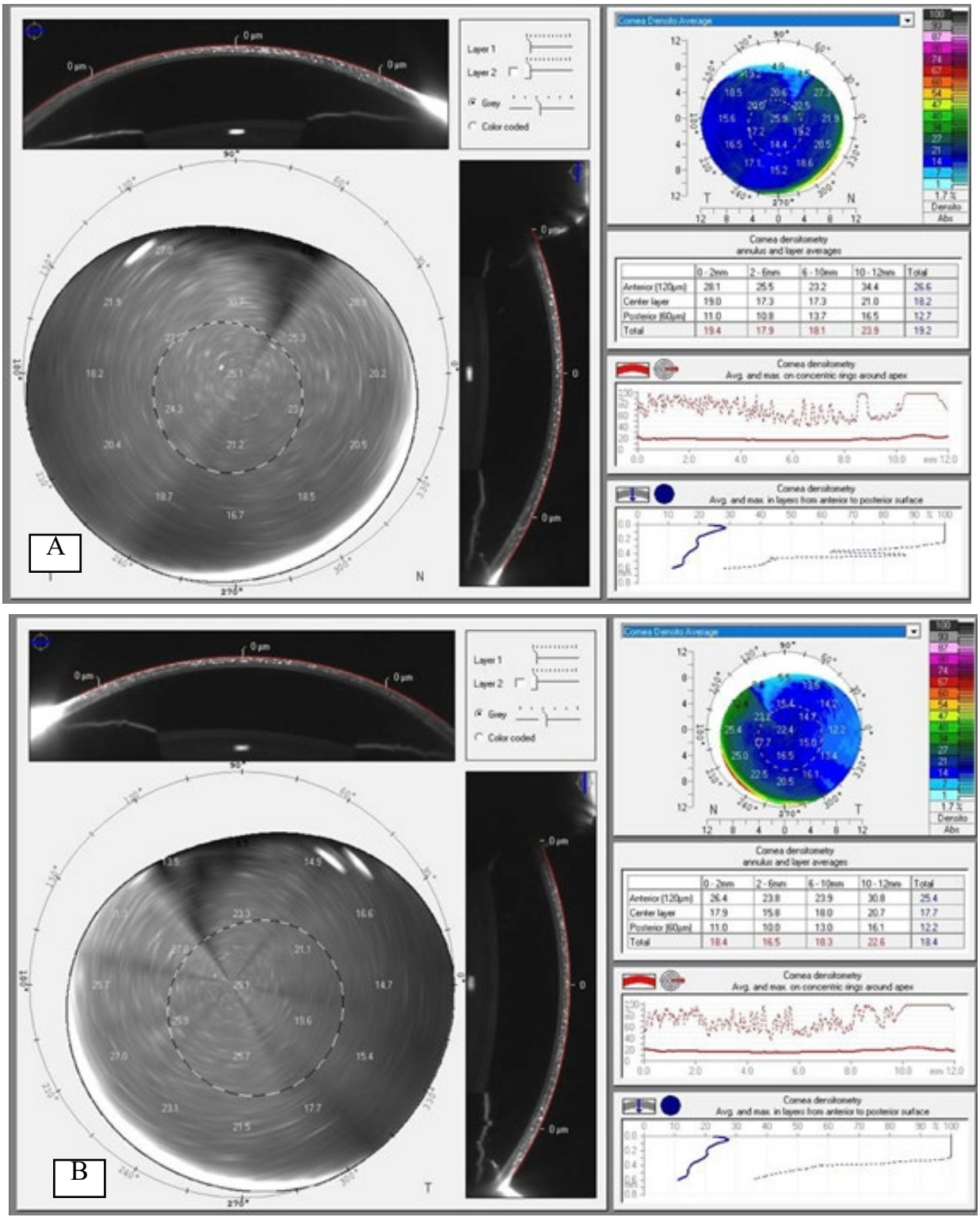

Figure 2A,B: Corneal densintometry of the right and left eyes showing increased density in the anterior third (120 µm), and decreasing density in the anterior to posterior direction.

Discussion

Cystinosis is a rare disease characterized by the accumulation of cystine crystals in various tissues of the body. Its spectrum ranges from milder forms with only ocular involvement to severe forms involving several organs, including the kidney.

The patient in question went for a routine consultation where she was asymptomatic. On ophthalmologic examination, the anterior biomicroscopy is compatible with a diagnosis of ocular cystinosis. In addition, she had corneal densitometry showing changes due to crystal deposits.

The patient's past history is relevant and corroborates with the ophthalmological findings. The patient had severe kidney disease, requiring kidney transplantation, but the disease was not elucidated. Therefore, the ophthalmological examination was fundamental for the diagnosis of the case.

References

1. Diogenes MB, Ana Carolina P. de Freitas, Nayara Queiroz Cardoso Pinto, Islane Maria Castro Vercosa, Dacio Carvalho Costa, et al. (2020) Cistinose ocular: um relato que enaltece a importância do tratamento tópico nos sintomas oculares. Fortaleza, CE. EOftalmo 6(2):46-49.
2. Shams F, Livingstone I, Oladiwura D, Ramaesh K (2014) Treatment of corneal cystine crystal accumulation in patients with cystinosis. Clinical Ophthalmology 8: 2077-2084.
3. Bunya VY, Maria A. Woodward, Matthew L. Haynie, S. Grace Prakalapakorn, Priyanka Kumar, et al. (2022) Cystinosis. A Academy Opthamol June 30, 2022.
4. Gustavo Victor, Paloma Juni Godinho Campos, Milton Ruiz Alves, Walton Nose (2004) Microscopia confocal in vivo na cistinose-Relato de caso. Sao Paulo (SP). Arq Bras Oftalmol 67:553-557.
5. Vaisbich MH, et al. (2019) Abordagem multi disciplinar para pacientes com cistinose nefropatica: modelo para atendimento em uma doença renal rara e crônica. Sao Paulo, SP, Brasil. Braz J Nephrol 41(1):131-141.
6. Liang H, Baudouin C, Hassani RTJ, Brignole-Baudouin F, Labbe A (2015) Photophobia and corneal crystal density in nephropathic cystinosis: an in vivo confocal microscopy and anterior-segment optical coherence tomography study. Invest Ophthalmol Vis Sci 56(5):3218-3225.