

Posterior polychromatic corneal dystrophy

Distrofia policromática posterior da córnea

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ABSTRACT

The authors describe two cases of posterior polychromatic corneal dystrophy, a pre-Descemet dystrophy, poorly described in national and world literatures, characterized by diffuse polychromatic points on the posterior corneal stroma, without visual impairment.

Keywords: Corneal dystrophies; Cornea/pathology; Descemet membrane/abnormalities; Corneal stroma/abnormalities; Case reports

RESUMO

Os autores descrevem dois casos de distrofia policromática posterior da córnea, uma distrofia pré-Descemet, pouco descrita nas literaturas nacional e mundial, em que se observam pontos policromáticos difusos no estroma posterior da córnea, sem aparente comprometimento da visão.

Descritores: *Distrofias hereditárias da córnea; Córnea/patologia; Lâmina limitante posterior/anormalidades; Substância própria/anormalidades; Relatos de casos*

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The authors declare no conflicts of interest

Received for publication 16/09/2014 - Aceito for publication 27/11/2014

INTRODUCTION

The term corneal dystrophy is used to describe hereditary, bilateral, symmetrical and progressive corneal diseases not related to environmental or systemic conditions¹.

Polychromatic dystrophy was described as a posterior stroma corneal dystrophy, pre-Descemet, characterized by punctate, polychromatic, uniform, diffusely distributed opacities with no apparent impairment of visual acuity².

Despite the probable dominant autosomal inheritance^{1,2} it has been described few times until today: only once in international² literature and once in national literature³, remaining still unknown by many ophthalmologists.

The aim of this article is to describe two cases of posterior polychromatic corneal dystrophy considering their features, their recognition in the ophthalmic practice and their clinical management.

CASE REPORT

CASE 1: DMS, 30 years old, female, leucoderma, married, economist, born and living in Belo Horizonte/MG, searched routine ophthalmologic evaluation due to ametropia. She wore glasses and soft contact lenses. She complained of recent discomfort from contact lenses. She reported that an ophthalmologist had already contraindicated the use of contact lenses a few years ago due to a “problem in the cornea” which she could not specify, but she decided to wear them again without medical supervision a few months ago. She denied having other previous eye diseases. She mentioned vitiligo without clinical manifestations and without the administration of medications at the time, and fibromyalgia with the administration of Cyclobenzaprine. She had no history of consanguinity and/or eye diseases in the family. The exam revealed static refraction in the right eye (RE) of -2.50 (VA = 20/25) and in the left eye (LE) of -2.50 (VA = 20/25). The biomicroscopy revealed mild hyperemia, mild papillary reaction, transparent corneas, with reduced BUT (*break up time*), diffuse punctatas and fine, uniform, polychromatic, diffusely distributed opacities in the posterior stroma (Figure 1). The intraocular pressure (IOP) was 11/10 mmHg at 5pm. The funduscopy showed no abnormalities. The non-contact specular microscopy (Konan® NONCON ROBO) showed cell count in RE = 2655 cells/mm² and LE = 2510 cells/mm², with discrete pleomorphism / polymegathism and numerous bright spots corresponding to the posterior stromal opacities seen at the slit lamp. The central ultrasonic pachymetry (Alcon® OcuScan RxP) was equal to 495 mm in the RE and 486 mm in the LE. Computerized corneal topography (Tomey® TMS-4) showed normal keratometric values, with a mild asymmetrical irregular astigmatism, suggestive of tear film changes.

The tomography exam of optical coherence (Visante® OCT) showed hyperreflective images in the posterior stroma, without other changes (Figure 1). The patient was advised to discontinue the use of contact lenses to restore the ocular surface, using lubricating eyedrops without preservatives and anti-allergy eyedrops. After that, use silicone hydrogel contact lenses with high oxygen permeability for a reduced time, with more frequent disposal, proper eye lubrication and periodic medical follow-up.

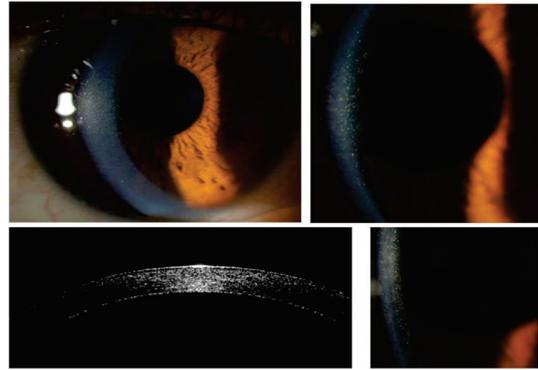


Figure 1 (patient 1): biomicroscopic images of punctate, fine, diffuse, polychromatic opacities in the posterior corneal stroma (above and below on the right) and optical coherence tomography showing hyper-reflective points in the posterior stroma (below on the left).

CASE 2: MOG, 23 years old, female, leucoderma, single, administrative assistant, born and living in Belo Horizonte / MG, searched routine ophthalmologic assessment due to ametropia. She wore glasses. She denied previous eye and/or systemic diseases. She had no history of consanguinity, and reported a grandfather with glaucoma. The exam revealed static refraction in the right eye (RE) of -1.50 -0.50 x 125° (VA = 20/20) and in the left eye (LE) of -2.00 -1.00 x 80° (VA = 20/20). The biomicroscopy presented mild meibomitis, mild hyperemia, mild papillary reaction, transparent corneas, with a little reduced BUT, discrete inferior punctatas and fine, uniform, polychromatic, diffusely distributed opacities in the posterior stroma (Figure 2). The IOP was 14/14 mmHg at 8pm. The funduscopy showed no abnormalities. The non-contact specular microscopy (Konan® NONCON ROBO) showed cell count in the RE = 3243 cells/mm² and LE = 2930 cells/mm², without pleomorphism / polymegathism and with numerous bright spots corresponding to the posterior stromal opacities (Figure 2). The central ultrasonic pachymetry (Alcon® OcuScan RxP) was equal to 561 mm in the RE and 570 mm in the LE. Computerized corneal topography (Tomey® TMS-4) showed normal keratometric values, with negligible astigmatism in both eyes (BE). The optical correction was prescribed, and the patient was advised to maintain regular medical follow-up (Figure 2).

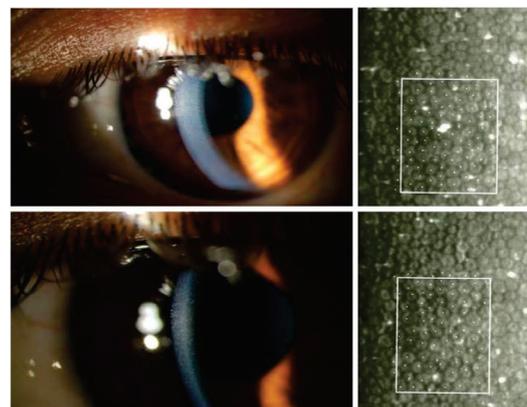


Figure 2 (patient 2): biomicroscopic images of punctate, fine, diffuse, polychromatic opacities in the posterior corneal stroma (on the left) and specular corneal microscopy showing hyper-reflective points corresponding to the opacities seen in the slit lamp (on the right).

DISCUSSION

The pre-Descemet dystrophies are not a well-defined clinical entity yet, and do not have a clarified pattern of genetic inheritance. For this reason, they are included in category 4 of the classification of the International Committee of Corneal Dystrophies Classification (IC3D), which includes new or previously documented corneal dystrophies with evidence that they are separate entities, although they are not well proven¹.

They usually manifest after 30 years old, but they have already been observed in children up to 3 years. Patients are usually asymptomatic, without vision impairment. And the clinical appearance of these dystrophies is thin, focal opacities located on the deep corneal stroma, prior to the Descemet's membrane, central, annular or diffuse, with different shapes and sizes¹.

Histological studies conducted showed only keratocytes increased in the posterior stroma containing cytoplasmic inclusions and vacuoles of lipid-like material, and the studies with electron microscopy showed vacuoles with electron-dense material suggestive of secondary lysosomes and lipofuscin¹. For this reason, some of them were related to a degenerative process and aging^{1,2}.

The studies with confocal microscopy showed only hiperrefletivas particles in the posterior corneal stroma, immediately prior to the Descemet membrane^{1,5,6}.

The differential diagnosis of these dystrophies should be done with the granular opacities of the Bowman's membrane (Reis-Bucklers), the stromal deposits of macular dystrophy, the guttatas of endothelial dystrophy⁷, the deposits of mucopolysaccharidosis^{7,8} and mucopolipidoses⁷ and deposits in the monoclonal gammopathy^{7,8-11}.

The first author to describe the corneal dystrophies of the pre-Descemet type was Vogt in 1923, and he called them farinata dystrophy⁴. Since then, several morphologies of pre-Descemet opacities have been described in the literature.

The first description of posterior polychromatic corneal dystrophy was made by Fernandez-Sasso et al in 1979 in Argentina, from the observation of 8 patients of the same family who had punctate, polychromatic, uniform, diffusely distributed opacities in the posterior corneal stroma. It was considered a new type of pre-Descemet corneal dystrophy, with probable dominant autosomal genetic inheritance².

Since then, only four extra clinical cases of two different families have been described by Tzelikis et al in 2007 in Brazil³.

These two present reports seem to be the third description in the literature to date.

The main features of posterior polychromatic corneal dystrophy, and which differs it from other pre-Descemet corneal dystrophies, are the variety of opacity colors both in direct lighting and in indirect lighting of the slit lamp, and the uniform and diffuse distribution of opacities of the same size, from limbo to limbo of the cornea, without forming aggregate areas, leaving transparent spaces between the opacities greater than their own opacities^{2,3}.

The diagnosis of this dystrophy is made by ophthalmologic exam in slit lamp, by means of observation of their morphological characteristics. It does not seem to interfere with the quality of vision of the patients, nor worsen with time^{2,3}.

There are few cases reported in the literature so far, and it is believed that this is due to lack of symptoms reported by the patients and the lack of knowledge about this entity by most

ophthalmologists. Therefore, many patients must not have been diagnosed yet.

Although it does not seem to require treatment over time, it is important to recognize this dystrophy, avoiding confusion with other clinical conditions which require specific care and treatment.

Studies to elucidate the gene responsible for this condition and the way of genetic inheritance are still needed so that the posterior polychromatic corneal dystrophy can be recognized as a well defined corneal dystrophy.

CONCLUSION

The posterior polychromatic corneal dystrophy is still not considered a well-defined clinical entity, and was described a few times, although it is believed to be more prevalent. The diagnosis is made primarily by eye examination at slit lamp. Although patients do not seem to have symptoms and/or progression of the disease, they should be correctly diagnosed for proper ophthalmologic advice and follow-up which allows greater knowledge about this condition.

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