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Case Report
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Unilateral PPMD with Keratoconus: An Atypical Presentation

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Abstract

Posterior polymorphous corneal dystrophy (PPMD) is a posterior corneal dystrophy with varied clinical presentation. We report a case of unilateral posterior polymorphous corneal dystrophy with advanced keratoconus in the fellow eye. The clinical manifestations and associations of this disease described in the literature have been discussed.

Keywords: PPMD, Keratoconus, Endothelial Abnormality

Introduction

Posterior polymorphous corneal dystrophy (PPMD) is a posterior corneal dystrophy with autosomal dominant mode of inheritance.The clinical manifestation can range from asymptomatic to endothelial decompensation and glaucoma in rare cases.Patients may be diagnosed with PPMD as part of routine ophthalmologic examination. Ocular associations of PPMD described in literature include keratoconus and secondary glaucoma.We report a case of a patient with changes consistent with unilateral PPMD with advanced keratoconus in the contralateral eye.

Case Report

A 39-year-old male patient presented to the outpatient department of a tertiary eye care hospital with progressive diminution of vision in his left eye. There was no history of redness or trauma. There was no history of use of glasses or any ocular surgery. On examination the unaided visual acuity was 6/6 in the right eye and counting fingers at 2 meters in the left eye which was improving to 3 meters with -2 dioptre cylinder at 180°. Slit lamp examination was normal except for the presence of scattered nodular lesions at the level of endothelium with normal corneal thickness, no stromal edema, quite anterior chamber and a clear lens in right eye. The left eye examination showed a steep thin cornea with apical scarring and Fleischer's ring, quite anterior chamber and clear lens suggestive of Keratoconus. The fundus findings were normal in both eyes with a cup disc ratio of 0.3:1 and a sharp foveal reflex. Intraocular pressure was recorded as 10 mmHg and 11 mmHg in right and left eye respectively with I- care tonometer [1-4].

Corneal topography (CSO Sirius) and Specular microscopy (Topcon) were performed for both the eyes. The patient had features suggestive of forme fruste keratonus in the right eye and advanced keratoconus in the left eye (figure 3). Anterior segment OCT (Casia 2) was done to assess the lesions on the endothelium and hypereflective lesions at the level of endothelium leading to irregular posterior surface were observed (figure 4A). ASOCT of left eye showed steep and thin cornea with apical scarring (figure 4B). The left eye specular microscopy couldn't be captured [4-6]. Right eye specular microscopy showed drop out zones of endothelium with cell density of 2358 and polymorphism in areas of endothelial abnormality (figure 5). The patient was given a trial for fitting of scleral contact lens for the left eye and with a prescription of 6.75/-4.5/18.9/8.5/5.13, he improved to 6/9. We prescribed scleral contact lens for the left eye and since right eye didn't have any symptoms or signs of endothelial decompensation the patient was advised regular follow-up.

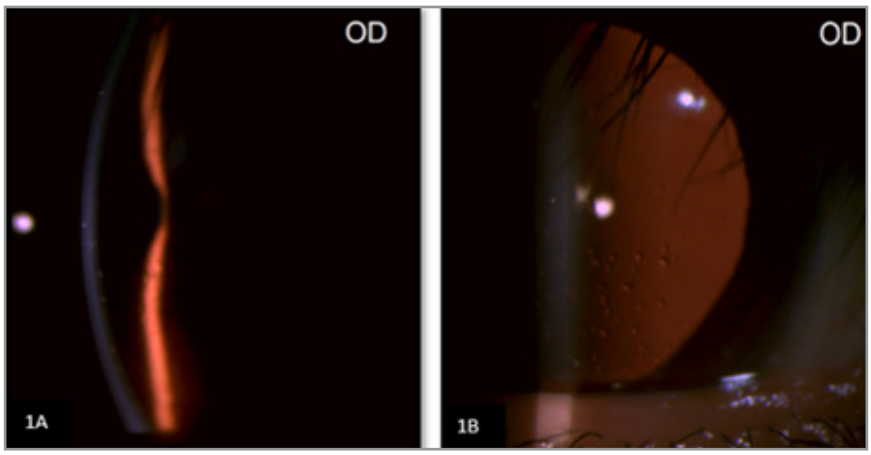

Figure 1: 1A- Slit lamp examination of right eye of patient showing lesions on the endothelium; 1B- examination with retro illumination of the same eye showing non-confluent lesions.

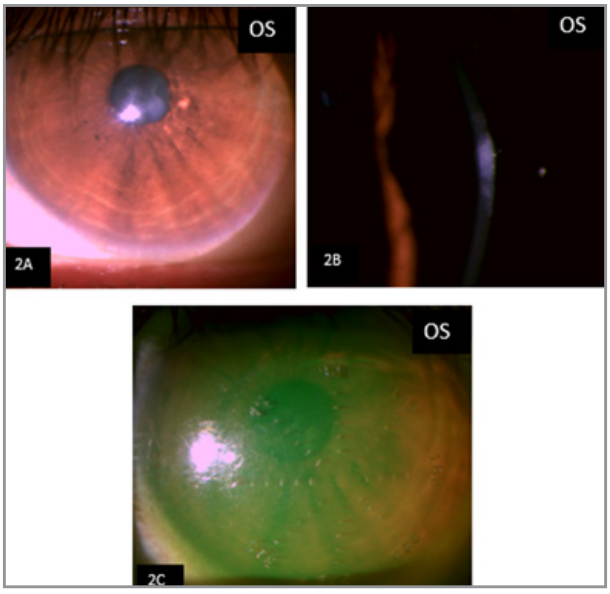

Figure 2: 2A- Left eye of the patient showing central scarring; 2B- Slit lamp exaination showing thinning and apical scarring in same eye; 2C- Scleral contact lens fit of the left eye of the patient

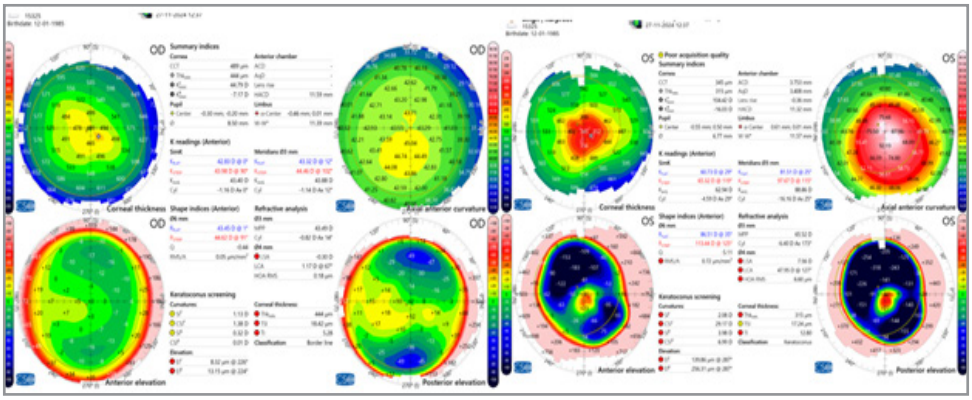

Figure 3: OD image showing right eye topography of patient with minimum corneal thickness 444µm; OS image showing advanced keratoconus with minimum thickness of 310 µm.

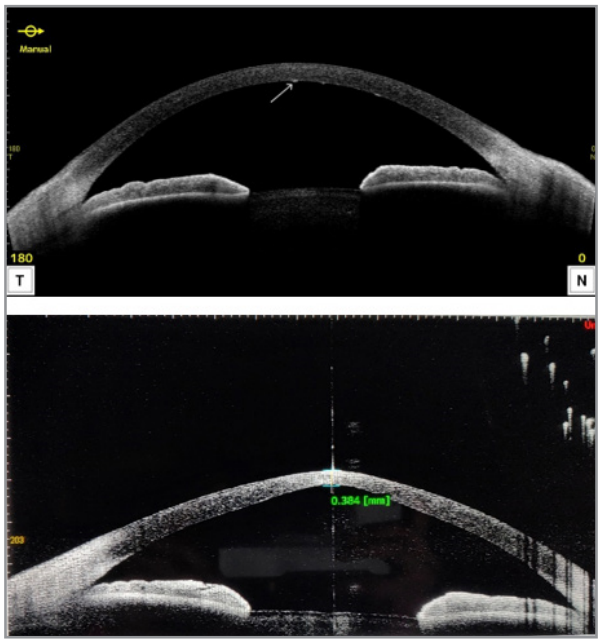

Figure 4: 4A- ASOCT of right eye showing hypereflective lesions at the level of endothelium (arrowmark) leading to irregular posterior surface; 4B- ASOCT of left eye showing steep and thin cornea with apical scarring.

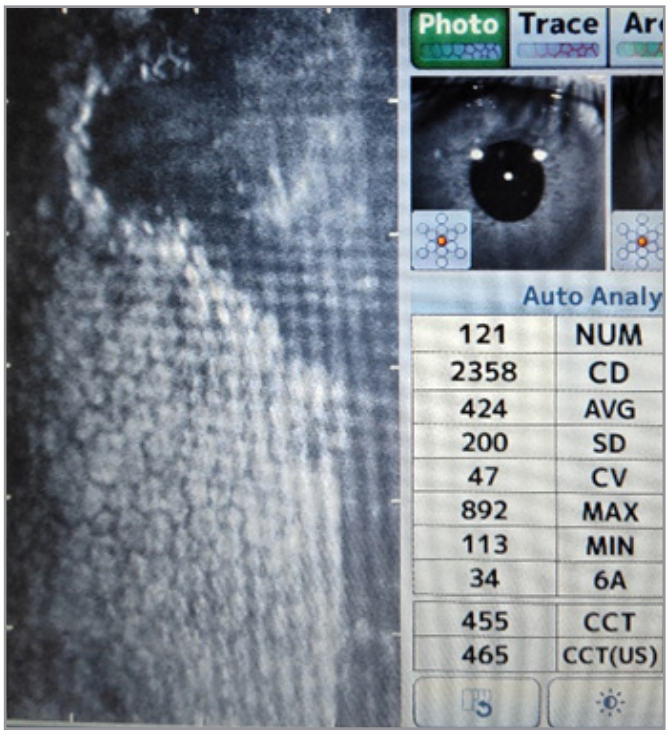

Figure 5: Right eye specular microscopy showing drop out zones of endothelium with cell density of 2358 and polymorphism in areas of endothelial abnormality.

Discussion

Posterior polymorphous corneal dystrophy (PPMD), also known as Schlichting dystrophy, is an autosomal dominant disorder of the corneal endothelium and Descemet membrane. The condition is characterized by its broad variability in clinical presentation, ranging from subtle corneal changes that may not affect vision to more severe cases leading to significant visual impairment.

The pathogenesis of PPMD is still not clear, but endothelial abnormality during embryonic development is proposed as a pos-

sible mechanism. Usually it manifests between the 3rd and 5th decade and affects females more often than males. In our case, the patient was a male which is different from the classical presentation [7, 8].

Differential diagnosis for PPMD includes other endothelial dystrophies like Fuchs endothelial dystrophy (FECD) and Congenital Hereditary endothelial dystrophy (CHED). FECD was unlikely because of absence of guttae. CHED was ruled out as it generally presents with increased early in life with decreased vision and edematous corneas.

Association of PPMD with other ocular diseases like keratoconus, ICE syndrome, secondary glaucoma has been described in literature. Some authors have also suggested to consider PPMD as part of other ectatic disorders of cornea like keratoconus, keratoglobus and pellucid marginal degeneration [9].

Reported two cases of keratoconus with PPMD. He proposed that the tears in Descemet membrane in keratoconus may have precipitated this dystrophy. However, in our case, the patient didn't have any findings suggestive of PPMD in the eye with advanced keratoconus.Described four cases of keratoconus which also showed findings consistent with PPMD. Out of the 4 cases, 2 patients had posterior corneal changes in one eye only. This finding suggested that though bilateral involvement is common in PPMD it can occur unilaterally as well. Our patient has unilateral PPMD, which may be considered as an unusual presentation. Reported a case of bilateral PPMD and keratoconus confirmed by histopathology. Keratoconus in that patient was not evident on clinical examination but the computer assisted keratography showed changes consistent with keratoconus.Investigated the anterior and posterior corneal topographic characteristics of three patients with PPMD. They concluded that PPMD causes changes both in anterior and posterior corneal curvatures and, in some patients, it might even be associated with keratoconus. They advised corneal topographic evaluation in every patient with PPMD and screening for refractive amblyopia in children with unilateral or highly asymmetric PPMD. Evidence exists in literature suggesting a common gene mutation shared by PPMD and keratoconus. Identified mutations in the VSX1 homeobox gene in patients of both diseases [10]. Our case may represent an atypical presentation of PPMD, given that the patient was male, exhibited unilateral PPMD, and did not show PPMD-related changes in the eye with keratoconus. Additional testing may be required to identify any gene mutations, such as ZEB1 or VSX1, linked to these observations. Further research is necessary to determine whether keratoconus and PPMD are interconnected or occur independently.

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