

## CASE REPORT

# Coincident Pellucid Marginal Degeneration and Fuchs' Endothelial Dystrophy

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## ABSTRACT

**Aim:** We report a rare case of coincident pellucid marginal degeneration and Fuchs' endothelial dystrophy.

**Background:** As far as the authors are aware there have been no previous reports of this combination of corneal disorders in the same patient.

**Case description:** A 45-year-old woman presented with progressive pellucid marginal degeneration and Fuchs' endothelial dystrophy. Progressive changes in corneal topography and specular microscopy imaging have been documented over 13 years since presentation. Declining vision has been successfully managed in this patient, to date, with serial crescentic corneal wedge excision biopsies to maintain acceptable spectacle-corrected visual acuity.

**Conclusion:** This rare combination of corneal disorders presents an interesting and unique challenge for surgical management in the future, where corneal decompensation and cataract are likely to become limiting factors for visual acuity.

**Clinical significance:** This is the first report of coincident pellucid marginal degeneration and Fuchs' endothelial dystrophy.

**Keywords:** Ectasia, Fuchs' endothelial dystrophy, Pellucid marginal degeneration.

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**Conflict of interest:** None

## AIM

To report a rare case of coincident pellucid marginal degeneration and Fuchs' endothelial dystrophy.

## BACKGROUND

Pellucid marginal degeneration (PMD) is a rare, bilateral, non-inflammatory corneal ectatic disorder characterized by a band of inferior corneal thinning.<sup>1</sup> PMD has a 3:1 male predominance and typically presents between the ages of 20–50 years with declining vision, irregular astigmatism, and inferior steepening and thinning noted on corneal tomography.<sup>2–4</sup> Fuchs' endothelial corneal dystrophy (FECD) is the most common corneal dystrophy and the most common dystrophy-related indication for corneal transplantation.<sup>5</sup> FECD is characterized by progressive loss of corneal endothelial cells, a formation of guttae and decreased vision typically associated with corneal edema.<sup>6</sup> In contrast to PMD, FECD has a 2.5–3:1 female predominance and presentation is typically either as an early onset subtype in the third decade, or a late onset subtype in the fifth decade of life.<sup>5</sup>

## CASE DESCRIPTION

We report the case of a 45-year-old woman who was referred in 2005 for management of suspected progressive keratoconus that had been present for many years. The patient had a history of asthma and hay fever, but no previous history of eye rubbing or contact lens use, and no family history of keratoconus or other ectatic disorders. The progressively declining vision had previously been corrected with spectacles, however, at the time of referral refractive correction in the right eye was becoming increasingly problematic.

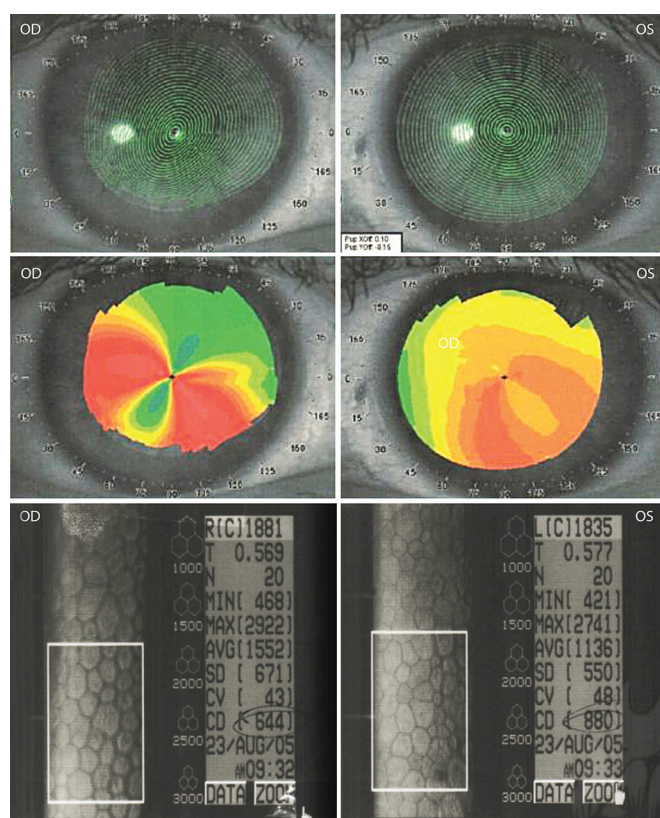
Unaided and spectacle-corrected visual acuity at presentation was 6/60-2 and 6/30 (right), 6/30 and 6/6 (left) respectively. Updated refraction at the first review demonstrated correction to 6/7.5 (+3/-12 x 65) in the right eye, and 6/6 (-0.5/-2 x 115) in the left eye. On slit lamp examination, a characteristic clear band of crescentic thinning of the inferior cornea in both eyes was noted between 4 o'clock and 8 o'clock. Significant central and paracentral endothelial guttata with regions of confluence were noted in both eyes. Pachymetry demonstrated central corneal thickness of 0.589 mm and 0.577 mm with endothelial cell densities measuring 644 and 880 cells/mm<sup>2</sup> on specular microscopy in the right and left eyes respectively. Corneal topography demonstrated a high magnitude of oblique irregular astigmatism in a 'crab claw' formation in the right eye and to a lesser degree in the left (Fig. 1). With the characteristic clinical signs and imaging findings, a diagnosis of PMD with coincident FECD was made.

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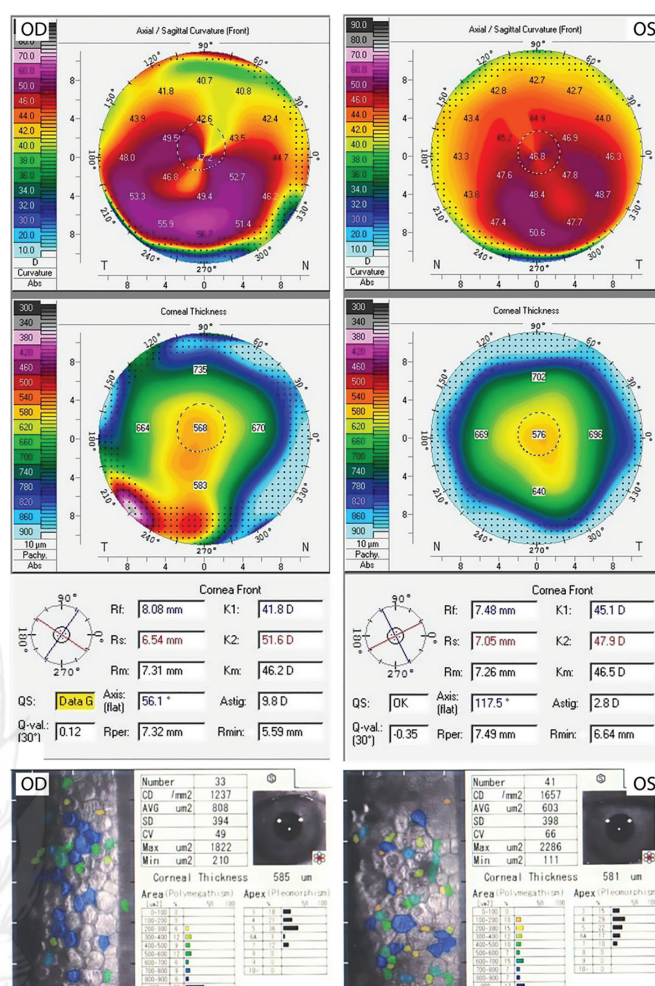
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**Fig. 1:** Topography (Tomey TMS 2N) and endothelial specular microscopy (Topcon SP2000) at presentation in 2005. Sim Ks: (OD) 56.96Dx139deg, 39.12Dx49deg; (OS) 47.64x39deg, 45.64Dx131deg

Contact lens fitting, intracorneal ring segments and other surgical options to correct the vision were declined by the patient at this time. Initial treatment was conservative with updated spectacle correction achieving 6/12 in the right eye and 6/4.8 in the left eye. Within three years of presentation, the best-corrected visual acuity had decreased to 6/60 in the right eye with associated corneal astigmatism of 18.5 D. An inferior lamellar crescentic corneal wedge excision of the right eye was completed. Following the removal of sutures, there was a significant reduction in corneal astigmatism to 2.7 D and a corresponding improvement in uncorrected right visual acuity to 6/12 (improving to 6/9 with pinhole).

Despite the success of the initial corneal wedge excision biopsy, by November 2017 the unaided visual acuity in the right eye had declined to 6/24 only improving to 6/9 with a pinhole. Corneal tomography demonstrated an increase in oblique astigmatism to 9.8 D in the right eye (Fig. 2). The left eye remained relatively stable with the uncorrected visual acuity of 6/12 improving to 6/4.8 with spectacle correction. Specular microscopy demonstrated cell density, the coefficient of variation (CV) and pachymetry of 1,237 cells/mm<sup>2</sup>, 49,585 µm (right), and 1,657 cells/mm<sup>2</sup>, 66,581 µm (left), respectively (Fig. 2). Corneal optical coherence tomography (OCT) demonstrated progressive thinning with 'beer-belly' protrusion of the inferior right cornea (Fig. 3).



**Fig. 2:** Tomography (Oculus Pentacam® HD) demonstrating progressive right astigmatism with inferior thinning and steepening. Specular microscopy (Tomey EM-3000) highlights low endothelial cell count and high coefficient of variation characteristic

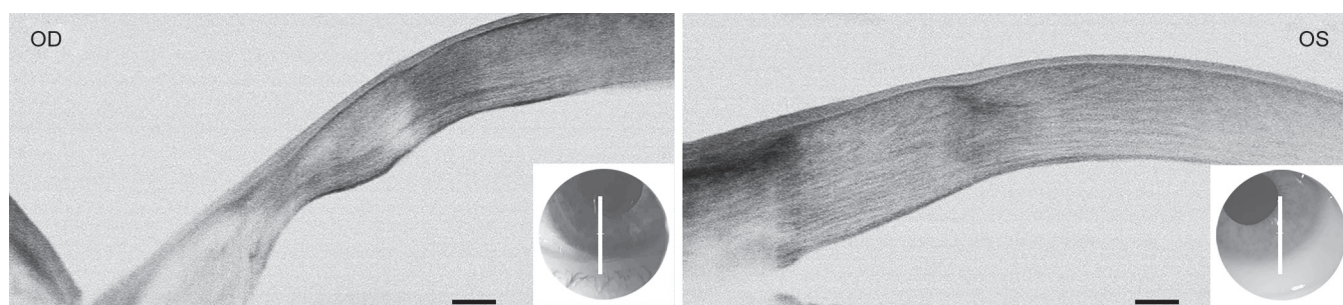
With the decreased vision noted in the right eye, a second inferior corneal wedge excision was completed. With significant inferior thinning and the low endothelial cell count, a decision not to crosslink the right eye was made at this time despite clear evidence of progressive tomographic changes. One week following surgery, the unaided visual acuity in the right eye had improved to 6/19.

## DISCUSSION

This is the first report in the literature of coincident FECD and PMD, as far as the authors are aware, and is likely to represent a very rare clinical entity. Many studies have reported the prevalence of FECD in the region of 4–10% of adults over 40 years old.<sup>7</sup> The prevalence of PMD is less well defined. A Japanese national study conducted in 2008–2010 identified 108 subjects with confirmed PMD in a country of 130 million.<sup>4</sup> The prevalence of coincident PMD and FECD, using the above prevalence data and assuming independence, could be as low as one in 10–20 million.

The coexistence of an ectatic corneal disorder with an endothelial disorder that is associated with increased corneal thickness and edema poses some interesting





**Fig. 3:** Vertical OCT images of both corneas demonstrating inferior ectasia prior to the second right corneal wedge excision biopsy. Evidence of the previous excision biopsy can be seen in the inferior right cornea appearing as a deep lamella scar

challenges for surgical management. Corneal cross-linking has previously been described in PMD;<sup>8-10</sup> however, in the context of endothelial dystrophy and significant ectasia, the risk of endothelial damage during cross-linking is likely to be high. Likewise, any decision regarding the timing of cataract surgery and endothelial keratoplasty is complicated with unstable and progressive corneal astigmatism.

Although there has been at least one case of PMD with positive family history reported,<sup>11</sup> there are no known genetic associations that have been identified for this ectatic disorder, and none that suggest any association with FCED. A nonsense mutation in the *KERA* gene has been described in a single case with superior PMD and cornea plana; however, no other family members homozygous for the mutation were noted to have PMD despite the presence of cornea plana in these individuals.<sup>12</sup> In contrast to PMD, FCED has been previously described in several rare cases of keratoconus and the association may have a genetic basis via mutation in the *ZEB1* gene.<sup>13</sup>

Central corneal thickness in this patient has remained relatively unchanged in the 13 years since presentation despite significant guttata and a relatively low endothelial cell count. The apparent increase in endothelial cell count since presentation is most likely due to a difference in specular microscopes used in each investigation. The most recently used Tomey EM-3000 specular microscope includes central and paracentral assessment of the corneal endothelium as opposed to the Topcon SP2000 microscope, used at the initial presentation, that evaluated central endothelium in isolation. It is very unlikely that there has been a true increase in endothelial cell density over time as suggested by the cell counts when comparing Figures 1 and 3.

## CONCLUSION

In summary, we present a rare and interesting case of coincident PMD and FCED that raises questions about the nature and timing of the most appropriate surgical management to optimize vision for this patient in the future. Due to asymmetric PMD, only one eye has demonstrated significant progressive visual decline and, to date, has been well managed with serial wedge excision biopsies. Interestingly, progression has been predominantly due to inferior steepening and a lesser degree, thinning.

## Clinical significance

This is the first report of coincident pellucid marginal degeneration and Fuchs' endothelial dystrophy.

## REFERENCES

1. Krachmer JH, Feder RS, Belin MW. Keratoconus and related noninflammatory corneal thinning disorders. *Surv Ophthalmol* 1984;28(4):293-322.
2. Moshirfar M, Edmonds JN, Behunin NL, et al. Current options in the management of pellucid marginal degeneration. *J Refract Surg* 2014;30(7):474-485.
3. Farrag AN, Hussein AA, Ummar S. Treatment of Pellucid Marginal Degeneration. *International Journal of Keratoconus and Ectatic Corneal Diseases* 2017;6(2):87-91.
4. Shimazaki J, Maeda N, Hieda O, Ohashi Y, Murakami A, Nishida K, Tsubota K. National survey of pellucid marginal corneal degeneration in Japan. *Jpn J Ophthalmol* 2016;60(5):341-348.
5. Elhalis H, Azizi B, Jurkunas UV. Fuchs endothelial corneal dystrophy. *Ocul Surf* 2010;8(4):173.
6. Vedana G, Villarreal G Jr, Jun AS. Fuchs endothelial corneal dystrophy: current perspectives. *Clin Ophthalmol* 2016;10:321-330.
7. Eghrari AO, Gottsch JD. Fuchs' corneal dystrophy. *Expert Rev Ophthalmol* 2010;5(2):147-159.
8. Bayraktar S, Cebeci Z, Oray M, et al. Corneal Collagen Cross-Linking in Pellucid Marginal Degeneration: 2 Patients, 4 Eyes. *Case Rep Ophthalmol Med* 2015;2015:840687.
9. Spadea L. Corneal collagen cross-linking with riboflavin and UVA irradiation in pellucid marginal degeneration. *J Refract Surg* 2010;26(5):375-377.
10. Hassan Z, Nemeth G, Modis L, et al. Collagen cross-linking in the treatment of pellucid marginal degeneration. *Indian J Ophthalmol* 2014;62(3):367-370.
11. Saeed HN, Bouchard CS, Garcia C. Case of Positive Family History for Pellucid Marginal Degeneration. *Invest Ophthalmol Vis Sci* 2012;53(14):4210-4210.
12. Khan AO, Aldahmesh M, Al-Saif A, et al. Pellucid marginal degeneration coexistent with cornea plana in one member of a family exhibiting a novel *KERA* mutation. *Br J Ophthalmol* 2005;89(11):1538-1540.
13. Mazzotta C, Traversi C, Raikup F, et al. First identification of a triple corneal dystrophy association: keratoconus, epithelial basement membrane corneal dystrophy and fuchs' endothelial corneal dystrophy. *Case Rep Ophthalmol* 2014;5(3): 281-288.