Coincident keratoconus and Fuchs' endothelial dystrophy: Dual dystrophies mask progression

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We present an interesting case of a 58-year-old woman who presented coincident keratoconus and Fuchs' endothelial dystrophy. During the 7months following initial presentation, a decline in visual acuity was associated with an apparent improvement in keratoconus, as characterized by corneal tomography parameters, while an increase in corneal pachymetry was also observed. The patient has been managed with left endothelial keratoplasty. This case poses the interesting question of how to classify coincident corneal disease, as traditional parameters for grading the severity of KC are altered by the presence of Fuchs' Endothelial Corneal Dystrophy and vice versa. In such cases, it is important to be aware of coincident disease to more accurately assess progression when considering treatment options and timing.

Key words: Ectasia, Fuchs endothelial dystrophy, keratoconus

Access this article online	
Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_1783_20

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Received: 02-Jun-2020 Revision: 18-Aug-2020
Accepted: 19-Oct-2020 Published: 23-Nov-2020

Keratoconus (KC) is a corneal ectatic disorder present in at least 1:2000 individuals that typically manifests in young adulthood with decreasing visual acuity associated with progressive corneal ectasia, myopia, and irregular astigmatism.[1-3] Disease progression often slows down in the third or fourth decade of life but can continue at a slower rate in older patients. Progression can be established and quantified using the ABCD or other similar grading systems that summarize complex changes in corneal shape and thickness.[4]

Fuchs' endothelial corneal dystrophy (FECD) is characterized by a decrease in endothelial cell density and regularity and eventually, in advanced cases, leads to progressive corneal edema.[5,6] Decreased visual acuity (VA) is largely due to the formation of corneal guttae and edema and in some cases, FECD may be hereditary.[5,7] Corneal edema and the associated visual changes may initially improve over the day but eventually persist as endothelial function progressively declines. FECD has a female preponderance of 3:1 with onset typically in the fourth decade of life and a prevalence of 4–10% in adults over 40 years of age.[6,8]

Case Report

We present the case of a 58-year-old woman who was initially diagnosed with progressive KC by her optometrist at the age of 43. She had no history of asthma, eczema, hay fever or eye rubbing, and no family history of KC. Several family members had been previously diagnosed with FECD, including her mother, maternal grandfather, and several other maternal family members. The patient had no previous history of eye surgery or treatment for her KC and reported progressively declining vision and glare, worse over the last 3 years,

Cite this article as: Wang N, McKelvie J. Coincident keratoconus and Fuchs' endothelial dystrophy: Dual dystrophies mask progression. Indian J Ophthalmol 2020;68:3074-6.

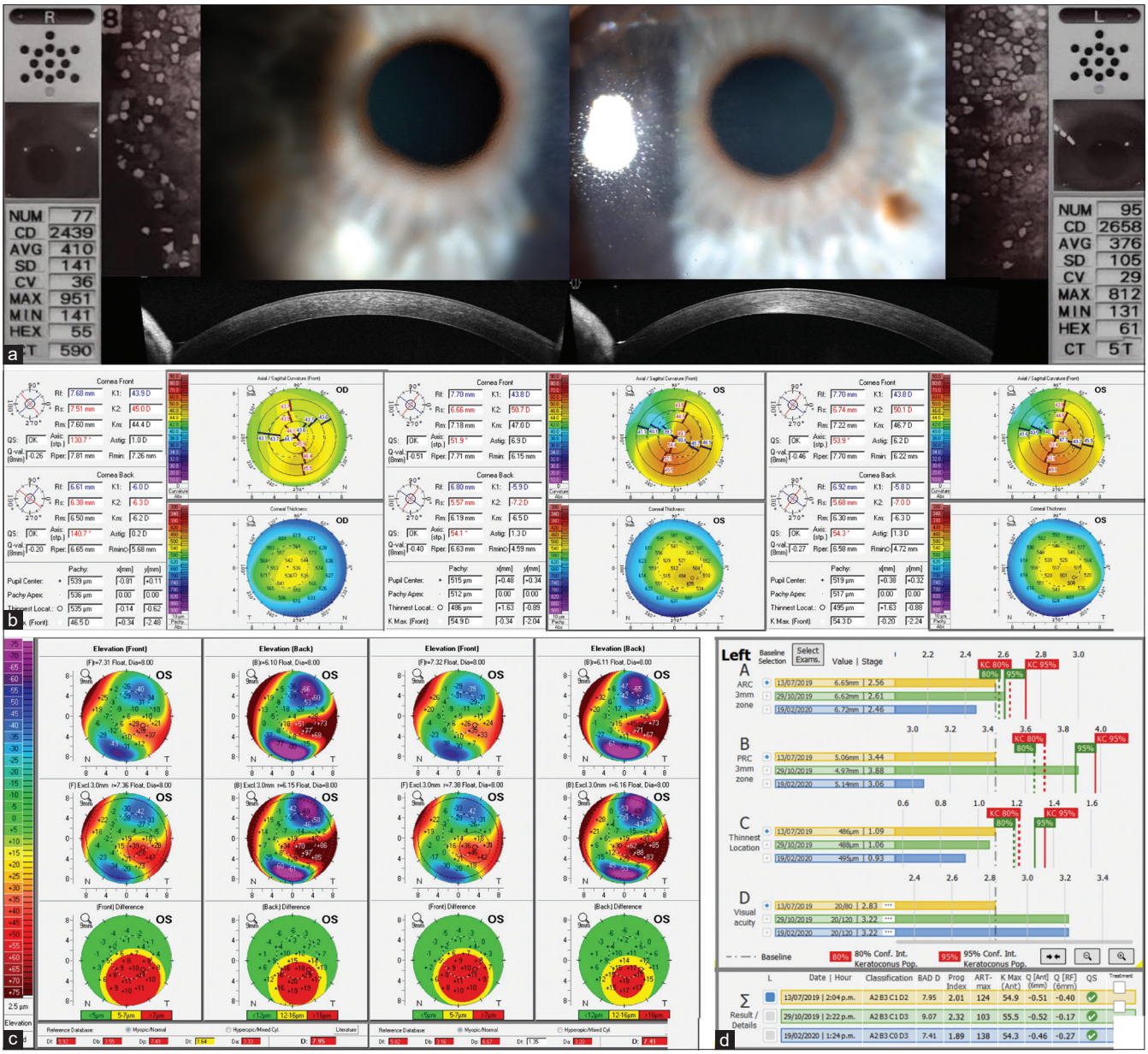

Figure 1: (a) Top row: specular microscopy and corneal photographs showing corneal guttae bilaterally. Bottom row: HD cornea OCT showing hyper-reflective endothelium bilaterally. (b) Pentacam tomography of the right eye and left eye at presentation (left) and the left eye 7 months later. (c) Pentacam Berlin/Ambrosio Display showing left eye at presentation (left) and after 7 months (right). (d) ABCD progression analysis of keratoconus showing improvement in all metrics, other than visual acuity (D), at 7 months

unbeknown to the patient which eye was affected. Visual changes were most noticeable in the mornings and triggered referral for assessment and treatment.

On examination, the best spectacle-corrected VA was 6/9 and 6/18 in the right and left eye, respectively. The slit-lamp examination demonstrated significant bilateral central guttata and paracentral thinning in her left cornea [Fig. 1a]. Pentacam tomography demonstrated irregular astigmatism with inferior steepening consistent with KC in the left and forme fruste KC in the right eye [Fig. 1b]. Corneal thickness at the thinnest location was measured at 535 µm on the right and 486 µm on the left, which increased to 495 µm over 7 months [Fig. 1b]. Severe central endothelial cell loss precluded accurate assessment of central endothelial cell counts; peripheral counts demonstrated cell densities of 2439 cell/mm^2 for the right eye and 2658 cell/mm^2 on the left eye [Fig. 1a]. Besides, high-definition (HD) cornea Optical coherence tomography (OCT) showed hyper-reflective endothelium bilaterally, a feature of FECD[9] These findings were consistent with a diagnosis of coincident KC and FECD. Optical coherence tomography of the right eye demonstrated granularity and thickening consistent with mild stromal edema.

The patient declined treatment and was monitored for further visual decline or progression of KC and FECD. During the 7 months following the presentation, VA deteriorated from 6/18 to 6/36 in the left eye and remained stable at 6/9 in the right eye Fig. 1c. depicts Pentacam the Berlin/Ambrosio Display (BAD)

showing left eye at presentation (left) and after 7 months (right). Front and back elevation maps demonstrate elevation patterns consistent with advanced KC. Note the change in the relative size of the red "elevation islands" in the difference maps (bottom row) with a reduction in the red exclusion zone observed at 7 months (right) compared with baseline (left). Simultaneously, a progressive increase in corneal thickness over the same period was noted [Fig. 1b]. Paradoxically, ABCD progression analysis of KC showed improved D indices (sum of standard deviations away from the mean) [Fig. 1d]. These changes and the improvement in the BAD-D value (from 7.95 to 7.41) are likely due to increased corneal edema rather than a decrease in the severity of the KC [Fig. 1d].

The deterioration in vision was primarily attributed to increasing corneal edema associated with FECD. After a discussion outlining conservative and surgical management options, the patient ultimately decided to proceed with left endothelial keratoplasty.

Discussion

This rare case of coincident KC and FECD highlights unique diagnostic monitoring and treatment challenges. Although the onset of KC may likely have been at a younger age than when first diagnosed in this patient, progressive KC was first noted at the age of 43 years with no significant associated risk factors. Late-onset progressive KC is atypical with most cases presenting in the second or third decade of life. KC and FECD have opposite effects on corneal thickness causing reciprocal masking of the severity and progression of one dystrophy by the other.

Despite the decline in VA, progression analysis using the Belin ABCD grading system demonstrated improvement at the 7 months rather than progression in all but VA. The apparent decrease in the severity of KC noted at 7 months in the left eye is largely due to increases in corneal thickness (Belin C grade) and decreased VA (Belin D grade) most likely associated with progressive FECD. This case highlights the need to carefully evaluate the progression of KC in the context of other factors that may influence corneal parameters typically used to quantify KC severity and progression. In this case, the KC "appears" to be mildly improving, masked by corneal thickening associated with worsening FECD. Meanwhile, the vision deteriorates from the combination of progressive KC and FECD.

Treatment options, in this case, include endothelial or full-thickness corneal transplantation to rehabilitate vision. With no significant lens opacity, after the relative risk and benefits were explained, this patient requested Descemet's membrane endothelial keratoplasty. If further progression is noted then corneal collagen cross-linking may also be required at a later date.

Conclusion

In conclusion, it would be prudent when assessing the progression of KC to consider other coincident diseases and factors that can also affect vision and corneal thickness, such as FECD.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Romero-Jiménez M, Santodomingo-Rubido J, Wolffsohn JS. Keratoconus: A review. Cont Lens Anterior Eye 2010;33:157-66.
2. Mas Tur V, MacGregor C, Jayaswal R, O'Brart D, Maycock N. A review of keratoconus: Diagnosis, pathophysiology, and genetics. Surv Ophthalmol 2017;62:770-83.
3. Rabinowitz YS. Major review. Surv Ophthalmol 1993;42:297-319.
4. Belin MW, Duncan JK. Keratoconus: The ABCD grading system. Klin Monbl Augenheilkd 2016;233:701-7.
5. Wilson SE, Bourne WM. Fuchs' dystrophy. Cornea 1988;7:2-18.
6. Nanda GG, Alone DP. REVIEW: Current understanding of the pathogenesis of Fuchs' endothelial corneal dystrophy. Mol Vis 2019;25:295-310.
7. Magovern M, Beauchamp GR, McTigue JW, Fine BS, Baumiller RC. Inheritance of Fuchs' combined dystrophy. Ophthalmology 1979;86:1897-923.
8. Vedana G, Villarreal G Jr, Jun AS. Fuchs endothelial corneal dystrophy: Current perspectives. Clin Ophthalmol 2016;10:321-30.
9. Iovino C, Fossarello M, Giannaccare G, Pellegrini M, Braghiroli M, Demarinis G, *et al*. Corneal endothelium features in Fuchs' endothelial corneal dystrophy: A preliminary 3D anterior segment optical coherence tomography study. PLoS One 2018;13:e0207891.