Keratoconus associated with Fuchs' Corneal Endothelial Dystrophy in a Young Woman: A Case Report

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ABSTRACT

Introduction: Keratoconus has association with various ocular diseases. Here a case of the co-occurrence of keratoconus and Fuchs' corneal endothelial dystrophy (FCED), a rare entity has been reported.

Case: A 26-year-old woman presented with a sudden onset of blurring of vision of right eye (RE) three weeks back.

Observation: The RE showed features of advanced keratoconus with apical scaring due to acute hydrops. Her left eye (LE) had myopic astigmatism with Vogt's striae. Careful examination revealed corneal guttate in both eyes. The co-existence of keratoconus and FCED was confirmed by tomographic scans and specular microscopy. The patient underwent penetrating keratoplasty in RE. The LE was planned for cross-linking therapy if keratoconus would progress.

Conclusion: Keratoconus and FCED have opposing effects on corneal thickness, resulting in reciprocal masking of the severity and progression of either condition. Corneal scans must be interpreted with caution as keratoconus and FCED differ completely in terms of their treatment options.

Key words: Fuchs'dystrophy; keratoconus; ocular disease.

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INTRODUCTION

In keratoconus, central part of the cornea becomes progressively thin and conical, resulting in irregular astigmatism. It begins at puberty and is prevalent in all races. Descemet's membrane may rupture leading to acute hydrops. Collagen cross-linking (CXL) can halt its progression. Advanced keratoconus requires keratoplasty. Keratoconus may be associated with ocular and systemic disorders (Gordon-Shaag et al., 2015; Feder et al., 2005). Fuchs' corneal endothelial dystrophy (FCED) is characterised by progressive decrease in endothelial cells resulting in corneal oedema and requiring endothelial keratoplasty. The FCED predominates in middle-aged females (Mylona et al., 2020). Co-occurrence of keratoconus and FCED is rare and may cause dilemma in diagnosis and management.

CASE REPORT

A 26-year-old woman came to Tilganga Institute of Ophthalmology (TIO) with history of blurred vision in both eyes that had been progressive, especially in the right eye (RE) over the last two years. She had been given glasses at local eye clinics and had changed glasses frequently. She said she had sudden blurred vision in her RE for last three weeks. She was then referred to TIO for further treatment. In her medical history, she had been treated for allergic conjunctivitis. She did not have other systemic disease. On examination, visual acuity in RE was 1/60 and did not improve with glasses. In left eye (LE) it was 6/24 and improved to 6/18 with -1 DS /-200 DC at 130 degrees. Retinoscopy showed scissoring reflex in both eyes. On slit lamp examination (Figures 1 A, B), central cornea of the RE was thin and bulging with central scarring. In the LE also, central thinning, bulging and also fine vertical lines (Vogt's striae) were seen in the corneal stroma. Munson's sign was present in RE, whereas Rizutti's sign and Fleischer ring were seen in both eyes. On specular reflection, the endothelium in both eyes showed corneal guttate with a beaten metal appearance (Figure 1B). Anterior chamber, lens, and posterior segment were normal in both eyes. Intraocular pressure could not be measured in the RE because of central cone formation and scarring, but digital tonometry was normal. The intraocular pressure of the LE was 8 mmHg on air puff tonometry.

The patient was evaluated for keratoconus and FCED. Anterior segment optical coherence topography (ASOCT) was observed (Figure 2). Patient underwent Pentacam scans (Figures 3, 4). These scans showed features of keratoconus that were advanced in RE.

Her corneal specular microscopy showed a low endothelial cell count, a high coefficient of variation, and a large average cell size noted in both eyes were suggestive of FCED (Figure 5).

The rigid, gas-permeable contact lens was intolerable in RE. After a trial of a scleral lens, vision improved to 6/60 in RE and 6/6 in LE. The patient underwent penetrating keratoplasty in the RE with a donor size of 8 mm and a recipient size of 7.75 mm. Post-operatively, The patient received topical steroids (prednisolone acetate 1%) and topical antibiotics (ofloxacin 0.3%), both six times daily for the first 10 days. Prednisolone drops were then reduced to four times daily for the next two months and thrice a day for another two months. Topical antibiotics

prophylaxis four times a day was given for a month and stopped thereafter. Her post-operative course was unremarkable. Four months after surgery, graft in the right eye was clear with unaided vision of 6/60 which improved to 6/18 with +2.50 DS /-6.00 DC at 70 degrees (Figure 6). For her left eye, close follow-up was advised to observe any progression of keratoconus. If progression occurs, CXL therapy is planned.

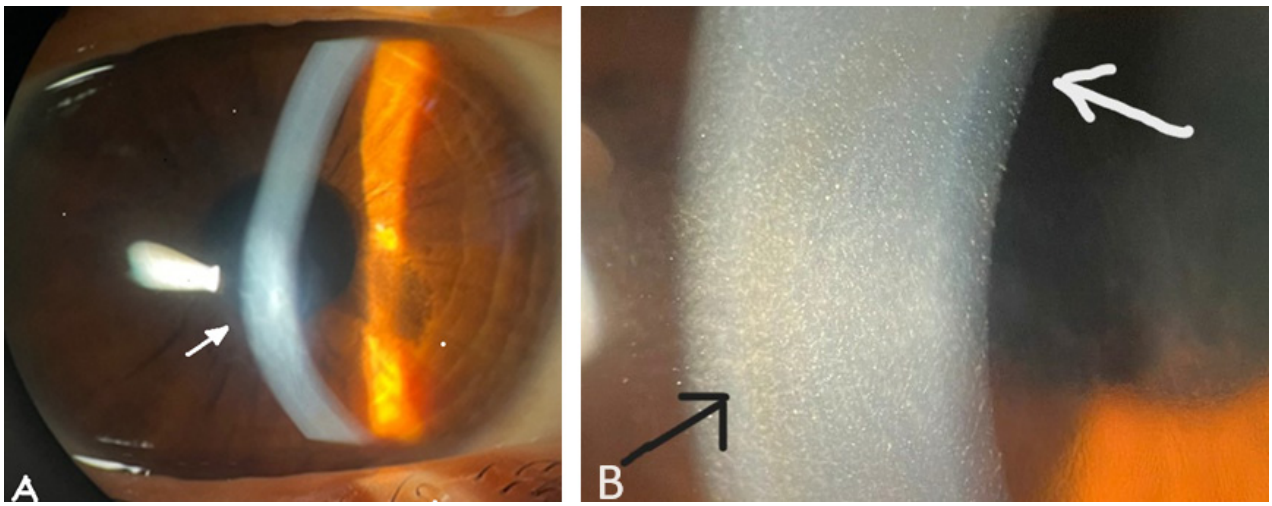

Figure 1: Slit lamp examination: A, Right eye showing cone formation (white arrow) of the cornea and hydrops which is resolving; B, Left eye (B) showing faint brownish line which is the part of Fleischer ring (black arrow) and guttae (white arrow).

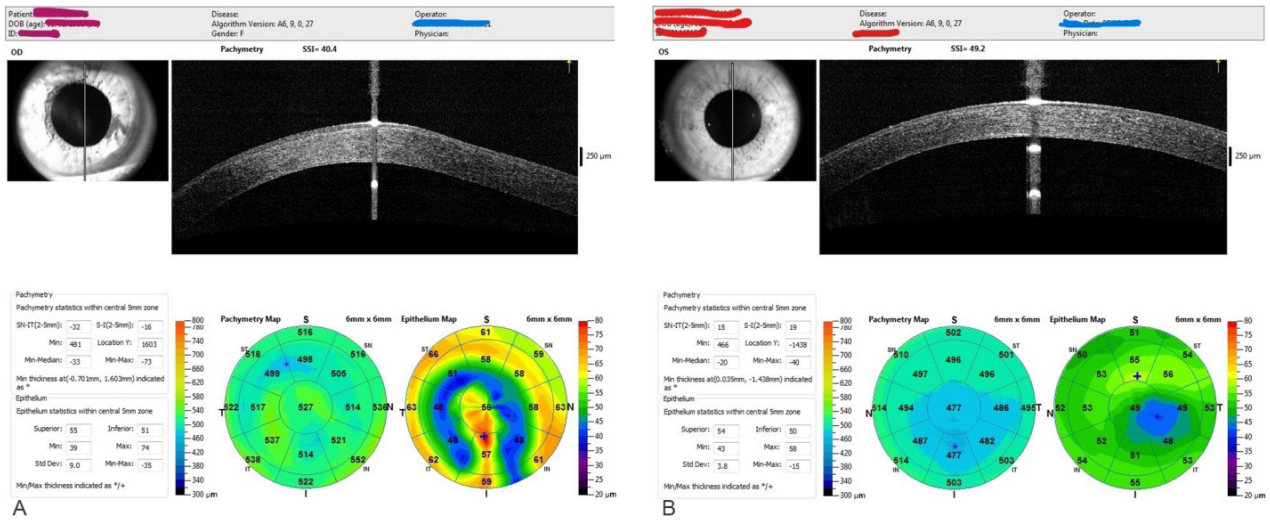

Figure 2: Anterior segment optical coherence topography (ASOCT) of right eye (A) and left eye (B). In both eyes, there is epithelial thinning in the central cornea; A, In the right eye, typical pattern of keratoconus is masked due to hydrops; B, In the left eye the minimal thickness of the cornea and the epithelium are corresponding to the same location, which is consistent with the keratoconus.

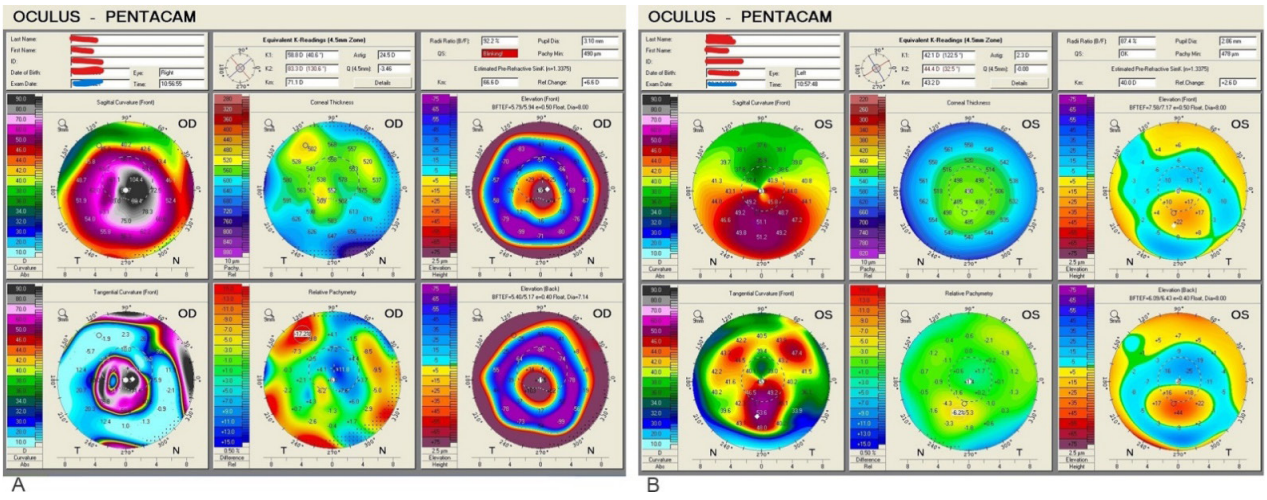

Figure 3: Pentacam image of right eye (A) and left eye (B); A, Steepening of the cornea of up to 83.3D, a posterior elevation of up to +137, but with a relatively thick cornea of 490 µm is observed; B, Central and inferior steepening of cornea with keratometry reading upto 44.4 D, posterior elevation of +44, and minimal pachymetry of 478 is seen.

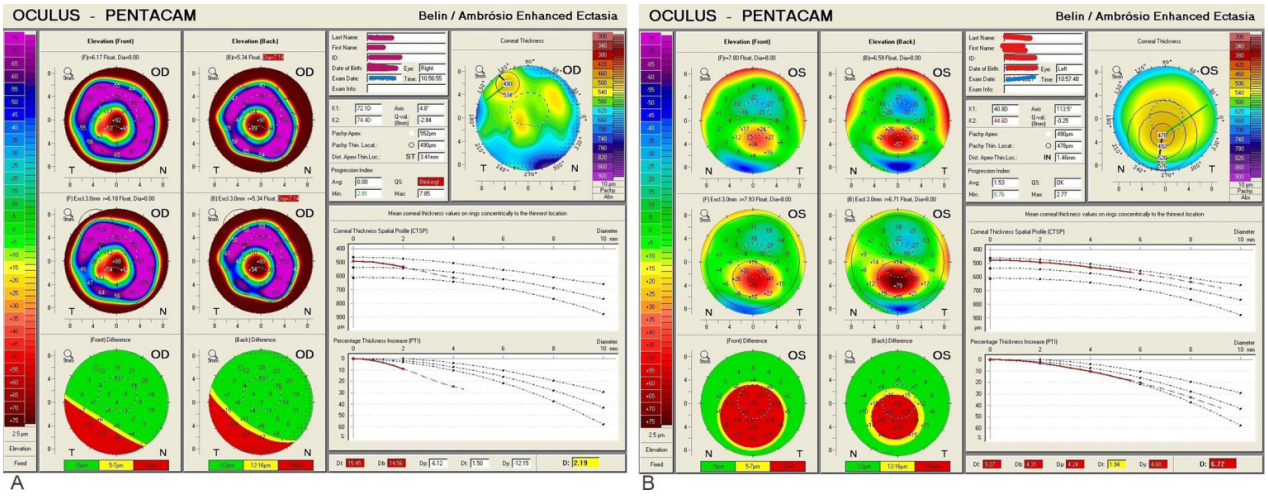

Figure 4: Berlin/Ambrosio Display Front and back elevation maps of right eye (A) and left eye (B). Both 'A' and 'B' show features of keratoconus.

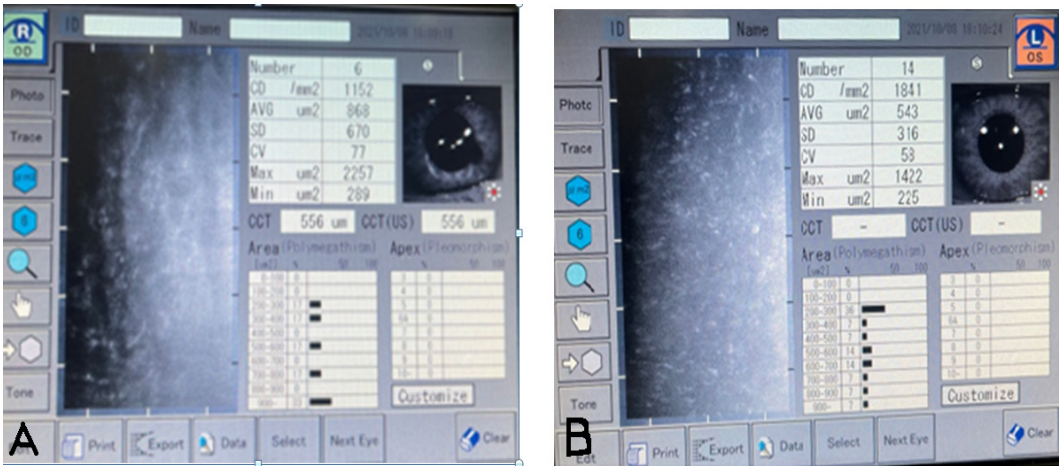

Figure 5: Specular microscopy of the right eye (A) and (B) showing loss of the honeycomb pattern of endothelial cells. Image 'A' showed cell count of 1152 /mm^2, average cell size of 868 µm^2, and a coefficient of variation of 77. Image 'B' showed cell count of 1841 /mm^2, average cell size of 543 µm^2, and coefficient of variation of 53.

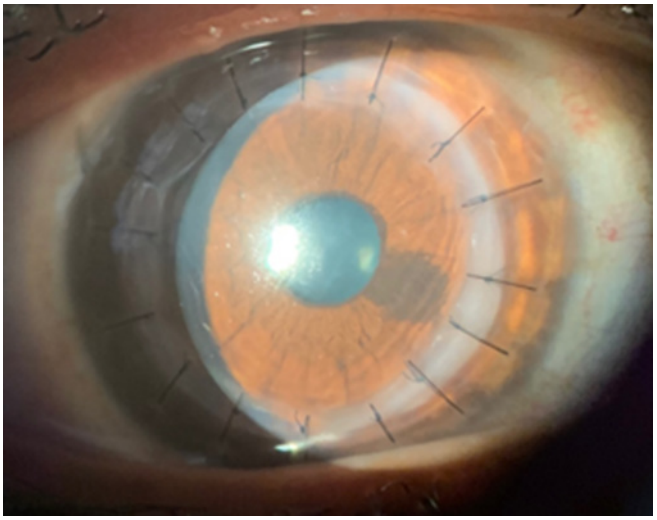

Figure 6: Four months after penetrating keratoplasty of right eye showing clear graft, (coincidental finding- Iris nevus near the pupil).

DISCUSSION

There have been few reported cases of keratoconus presenting together with FCED from different parts of the world. As per authors' knowledge current case is the first to be reported from Nepal.

Possible pathways for the co-occurrence of keratoconus and FCED have been studied in a literature review (Mylona et al., 2020) but so far there is no conclusive and proven genetic and histopathological explanation. Corneal thinning caused by keratoconus and corneal thickening caused by FCED may combine to normalise

corneal pachymetric values (Cremona et al., 2009). In our case, corneal thickness measured by ASOCT in the RE was 481 μm (Figure 2A). This value does not correlate with the advanced stage of disease, as one would normally expect the central corneal thickness (CCT) to be much lower when it is up to occurrence of hydrops. The relatively high CCT value in RE could be due to the residual corneal oedema of the acute hydrops or due to co-existing FCED. Ramos et al., (2012), Cremona et al., (2009), and Jurkunas et al., (2006) had reported CCT values of 482, 557, and 543 μm respectively in cases with keratoconus and FCED existing together. These values are normal or near normal, like in this case. The results of specular microscopy (Figure 5) of our patient with a low number of endothelial cells count, an abnormal endothelial mosaic with corneal guttate are in accordance with the study of Cremona et al., (2009) which showed similar findings associated with keratoconus.

Because of their different pathophysiology, keratoconus and FCED differ completely in terms of their treatment options. Different surgical approaches have evolved for the treatment of coexisting cases of keratoconus and FCED, depending on the initial presentation. Full-thickness corneal transplantation is the appropriate surgical option because both the anterior and posterior cornea is involved in the disease (Jurkunas et al., 2006). Vira et al., (2014), in his cases series had performed Descemet stripping endothelial keratoplasty (DSEK) in combined keratoconus and FCED and concluded that if DSEK is performed for FCED prior to visually significant apical corneal scarring from keratoconus, good visual outcome may be achieved. As per case report of Cooper et al., (2017), Descemet membrane endothelial keratoplasty (DMEK) may also be an effective alternative in eyes with coexisting stable keratoconus and FCED. But keratoconus corneas possess an abnormal curvature so there is some concern that DMEK grafts would be predisposed to detachment (Cooper et al., 2017). In our patient, as the RE had advanced keratoconus with central corneal scarring penetrating keratoplasty was the only option of management. Post-operatively, graft was clear and unaided vision improved to 6/60 and pin hole vision of 6/12 at four months.

Effect of CXL in coexisting cases of keratoconus and FCED is scarce in literature, owing to rarity of occurrence of the condition. The CXL can have endothelial adverse effect due to irradiation by ultraviolet light. The effect may manifest more in case of FCED due to already compromised endothelium, so early surgical intervention (endothelial keratoplasty) will likely be required if the CXL procedure is performed. Ono et al., (2018) in his case series mentioned therapeutic benefit of CXL in decreasing pain due to bullous keratopathy without any effect in Best Corrected Visual Acuity (BCVA) and corneal thickness. In current case, in the left eye, repeat-Pentacam scans at four months showed no progressive changes in keratoconus or FECD. The plan for the LE is that, in course of follow up, if LE shows progressive changes of keratoconus, CXL will be advised and if endothelial decompensation would occur, Descemet stripping automated endothelial keratoplasty (DSAEK) will be planned.

CONCLUSION

Due to rarity of co-existent keratoconus and FCED, strong suspicion is required for diagnosis. Because of counter-effect in corneal thickness, there is high possibility of missing one or both diagnoses or underestimating the disease severity which may predispose to unexpected surgical complications. Therefore, proper investigation and interpretation of the reports are important in such situations. Management of such corneal co-morbidity has to be customised according to the stage and course of the disease.

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