Atypical variant of Fuchs’ endothelial dystrophy mimicking iridocorneal endothelial syndrome: A case report

Mona Bhargava, Jyotirmay Biswas1, Shweta Patil1

To report an unusual presentation of Fuch’s endothelial dystrophy (FED) and its management. A 53-year-old male patient presented with unilateral progressive painless diminution of vision. Best-corrected visual acuity of the right and left eyes were 20/20 and 20/400, respectively. Slit lamp examination of the right eye was unremarkable, on left eye examination there was corneal edema, shallow anterior chamber, areas of 360 degrees iridocorneal touch with few synechiae on gonioscopy and age-related nuclear sclerosis. Pachymetry and specular microscopy imaging were performed before and after the surgical procedure. Ultrasound biomicroscopy (UBM) was performed preoperatively to rule out the presence of any angle anomalies. Combined procedure of Phacoemulsification with intraocular lens implantation and Descemet’s membrane endothelial keratoplasty (DMEK) with peripheral iridectomy were performed. At 6-weeks follow-up, best-corrected visual acuity improved from 20/400 to 20/20. Slit lamp examination of the left eye showed clear DMEK graft with patent inferior peripheral iridectomy. Specular microscopy showed a cell count of 1761 cells/mm². In a patient presenting with unilateral corneal edema, shallow anterior chamber depth, and iridocorneal adhesions, one needs to rule out the diagnosis of atypical variant of FED.

Key words: Atypical Fuch’s endothelial dystrophy, DMEK

Fuch’s endothelial dystrophy (FED) is a hereditary, progressive disease of the posterior cornea which results in excrescences of Descemet membrane termed as “guttae,” accompanied by endothelial cell loss, corneal edema, and, in late stages, bullous keratopathy. It usually presents in the fifth or sixth decade. Symptoms are related to the corneal edema, which causes decrease in vision.

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Microscopically, the endothelial cells are noted to be larger (polymegathism) and more polymorphic (pleomorphism). Primary dysfunction of the endothelial cells manifests as increased corneal swelling and deposition of collagen and extracellular matrix in Descemet’s membrane. As the endothelium further decompensates, the central cornea thickens.

**Case Report**

A 53-year-old male presented with painless, gradual diminution of vision in left eye from 1 year. The patient had undergone uneventful right eye phacoemulsification with intraocular lens implantation.

On examination, best-corrected visual acuity (BCVA) was 20/20 in the right eye and 20/400 in the left eye. Anterior segment examination of the right eye showed clear cornea, deep anterior chamber with pseudophakia, and left eye examination showed corneal edema, descemet folds, and guttae [Fig. 1a]. Anterior chamber was shallow (Von Herick grade I) with 360 degrees iridocorneal touch and lens had grade II nuclear sclerosis [Fig. 1b]. Applanation tonometry in right and left was 17 mm hg and 15 mm hg, respectively. Gonioscopy of the right eye showed open angles up to scleral spur, left eye showed iridocorneal touch with peripheral synechiae from 11 to 12 o’clock. Central corneal thickness (CCT) was 525 microns in right eye and 698 microns in left eye. Specular microscopy of right eye showed polymegathism with cell density of 876 cells per mm$^2$, coefficient of variation was 30.6 and hexagonality was 64% and left eye the specular image did not freeze. Ultrasound biomicroscopy of the left eye showed 360 degrees closed anterior chamber angle with peripheral iridocorneal touch, anteriorly pushed lens-iris diaphragm with convex configuration of iris [Fig. 2].

Uneventful DMEK was performed along with phacoemulsification. Phacoemulsification was done first. Anterior chamber deepened after phacoemulsification so the iridocorneal touch was resolved. Peripheral anterior synechiae was minimal so no surgical intervention was done for the same. DMEK graft of 8mm was prepared. The host Descemet’s membrane (DM) was stripped from the central 8 mm, and the donor graft inserted, unfolded, and attached to the recipient stroma with air in anterior chamber. An inferior peripheral iridectomy was performed at 6 o’clock position, to prevent pupillary block.

Post-operatively, slit lamp examination of the left eye showed clear and well apposed DMEK graft, with patent inferior peripheral iridectomy [Fig. 3]. No graft displacement was noted in the post-operative period.

At 6 weeks follow-up, the best corrected visual acuity was 20/20. Slit lamp examination showed a clear and well attached DMEK graft. Anterior chamber was of normal depth with intraocular pressure of 12 mm hg. Following surgery, left eye pachymetry was 482 microns, as compared to preoperative pachymetry of 698 microns, specular microscopy showed cell density 1761 cells per mm$^2$, as compared to preoperative image not freezing.

Histopathological analysis of the host DM showed wart like lesion with thickening of the DM, suggestive of FED [Fig. 4].

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**Figure 1:** Pre-operative slit lamp photo of left eye showing corneal edema (a) and shallow anterior chamber (b)

**Figure 2:** Ultrasound biomicroscopy showing area of iridocorneal touch (red arrow)

**Figure 3:** Post-operative slit lamp photo of left eye showing attached DMEK graft with patent inferior peripheral iridectomy

**Figure 4:** The photograph showing Descemetæs membrane (Haematoxylin and Eosin stain x100) with wart like projections (arrows) with insert haematoxylin and eosin stain x400
Discussion

FED exhibits dominant inheritance but the degree to which the condition is expressed is variable, so some can be more severely affected than others. Fuchs’ dystrophy comes in a wide variety of intensities from extremely dense, pigmented, central corneal guttata to a diffusely mildly depressed endothelial cell count with scattered guttata, to non-pigmented guttata. Accordingly, the presentation can vary from asymptomatic presentation to stromal edema, sub epithelial fibrosis and epithelial bulla.

Chandler’s syndrome is a variety of Iridocorneal endothelial (ICE) syndrome which presents unilaterally with a hammered silver appearance of the corneal endothelium and shallow anterior chamber with peripheral anterior synecchie, findings similar to our case. However, absence of iris abnormalities, corectopia and elevated intraocular pressure differentiates our case from Chandler’s syndrome.

Abbott et al. [1] observed six phakic patients with non-guttate corneal endothelial degeneration who had unilateral edema, with only polymorphic corneal endothelial cells in the contralateral eye, had a resemblance to our case with no evidence of corneal guttata in either eye clinically where endothelial cell counts were decreased, may be a different of FED as suggested by Abbott et al.

Pitts et al. [2] showed that Fuch’s dystrophy is associated with axial hypermetropia and shallow anterior chamber, which provides a link with the increased risk of angle closure glaucoma. They showed out of 25 eyes (76%) have an axial length below 22.3 mm. In our patient axial length was 24 mm. They also have calculated a mean spherical equivalent of +2.45 D in the study but in our case there was only cylinder of – 1.75D. It is interesting to speculate on the nature of the relationship between Fuchs’s dystrophy and axial hypermetropia. It is unlikely that the endothelial dystrophy leads to axial hypermetropia, as hypermetropia is known to develop in early life while Fuchs’s dystrophy tends to become manifest over the age of 50.

Tomlinson and Leighton [3] found a mean anterior chamber depth of 2.31 mm in patients with primary angle closure glaucoma and the corresponding mean axial length was 22.6 mm. In our case anterior chamber depth was 1.23 mm and axial length was 24 mm.

In FED, histopathologically DM appears as a thickened band of 2 opaque lines, the anterior line as smooth whereas the posterior line has a wavy and irregular appearance with areas of localized thickenings. [4] In our case we reported wart like lesion of DM.

Conclusion

Diagnosis of FED can be challenging when it presents with unilateral atypical characteristics. A careful evaluation is mandatory, to rule out the diagnosis of atypical variant of FED in a patient presenting with unilateral corneal edema, shallow anterior chamber depth, and iridocorneal adhesions. DMEK is the procedure of choice in managing cases of atypical FED.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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