CASE REPORT

Bilateral big-bubble deep anterior lamellar keratoplasty for vascularised corneal opacity in a patient with xeroderma pigmentosum

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SUMMARY

A 16-year-old male patient diagnosed with xeroderma pigmentosum developed bilateral vascularised corneal opacities after treatment for ocular surface squamous neoplasia. The patient underwent an uneventful big-bubble deep anterior lamellar keratoplasty in both eyes. The grafts remained clear and the corrected distance visual acuity was 20/30 in both the eyes at the final follow-up visit.

BACKGROUND

Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disorder which starts in early childhood and is characterised clinically by cutaneous photosensitivity, pigmentary changes, photophobia and propensity for early development of malignancy in sun-exposed mucocutaneous and ocular structures.^{1–3} The ocular changes include severe photophobia, conjunctivitis, corneal opacification, conjunctival naevi, epibulbar and lid neoplasms. As this condition is rare, only isolated case reports of XP are found in the literature. Corneal involvement is seen in 17% of cases.^{2,3} It is seen in the form of dryness, haze, oedema, exposure keratitis, vascularisation, pterygium-like growths, epithelial neoplasms and opacification.^{2,4} The literature on the histopathology of corneal specimens has shown changes in all layers of the cornea. In spite of the outcomes of penetrating keratoplasty (PKP) being poor, it remained the treatment of choice for visual rehabilitation of these patients with vascularised corneal scars.^{2,4} Chaurasia and Ramappa⁵ have shown that patients with XP can present with only corneal oedema and there can be a good outcome with endothelial keratoplasty. They have also shown that performing anterior lamellar keratoplasty in these patients may not be a good option because of poor endothelial cells noted on histopathology.⁶ We report a case of XP with vascularised corneal scars in both eyes that was successfully treated by deep anterior lamellar keratoplasty (DALK) with an uneventful postoperative course. To the best of our knowledge, this is the first case of bilateral DALK to be reported in a patient with XP.

CASE PRESENTATION

A 16-year-old male patient was referred to the cornea service for visual rehabilitation due to the presence of a vascularised corneal scar. He was diagnosed with XP and has been treated for

bilateral ocular surface squamous neoplasia (OSSN) by excision biopsy, alcohol-assisted keratoepitheliectomy and double-freeze cryotherapy to the resected margins and base of the lesion followed by amniotic membrane transplantation once in the right eye (OD) and twice in the left eye (OS). The child had difficulty in reading at school due to poor vision in both eyes (OU).

On examination, his corrected distance visual acuity (CDVA) was 20/600 in the right eye (OD) and 20/200 in the left eye (OS). The lid position, congruity and tear meniscus height were normal in OU. Slit-lamp examination of the cornea showed a vascularised corneal scar located in the anterior one-third to half of the corneal thickness in OU with the right eye being severe (figure 1A, B). The superior one-third of the cornea remained clear and there was no oedema. In view of the location of the scar, there were no clinical signs of endothelial decompensation and poor prognosis for PKP, and we decided to perform DALK in OU.

TREATMENT

The DALK procedures were performed under general anaesthesia in both the eyes. The surgery was performed first in the right eye, as the visual acuity was poor, and later in the left eye.

Recipient preparation

Partial thickness trephination (7–8 mm) up to two-third depths of the host cornea was carried out. A 26-gauge disposable needle bent at 90° and mounted over a 2 cc syringe was advanced into the central corneal stroma and air was injected. Big bubble was achieved and the anterior two-third part of the stroma was debulked using a crescent blade (Alcon Surgical, Fort Worth, Texas, USA) leaving behind only a thin layer of the stroma. The anterior chamber was decompressed via a paracentesis at 10-o'clock using a 15° blade (Alcon Surgical, Fort Worth, Texas, USA), prior to debulking. The big bubble in the predescemet space was reached by making an incision in the thin layer of the remnant posterior corneal stroma using a No.11 Bard-Parker blade. Sodium hyaluronate (Healon, Abbott Medical Optics, Inc, Illinois USA) was injected into the cleavage plane to maintain separation between the stroma and Descemet's membrane (DM). The cleaved stroma was divided into four quadrants, and the DM was completely bared.

CrossMark

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Figure 1 Clinical pictures showing (A) right eye with calcific plaque-like degeneration and vascularised corneal opacification; (B) left eye with vascularised corneal opacification, and (C and D) showing clear graft in the right eye and left eye, respectively, at the most recent follow-up.

Donor preparation

Corneas stored in the McCarey-Kaufman medium were used for transplantation. The donor cornea was oversized by 0.25 mm in DALK and 0.5 mm in PKP. In DALK, after staining the endothelial side with 0.06% Trypan blue dye (Blue Rhesis, Contacare Ophthalmics & Diagnostics, Vadodara, Gujarat, India), the DM was peeled off with a pair of fine non-toothed forceps. The donor lenticule was secured with 10-0 monofilament nylon by 16 interrupted sutures.

OUTCOME AND FOLLOW-UP

On postoperative day 1, he had an uncorrected visual acuity of 20/80, with mild graft oedema, well-apposed graft host junction and sutures in place. The patient was started on moxifloxacin (0.5%) eye drops four times a day, which were stopped at 1 week, and prednisolone acetate (1%) eye drops six times a day, which were tapered slowly. On subsequent visits, he maintained a clear graft (figure 1C). Corneal topography-guided suture removal was carried out at follow-up visits. At 53 months of follow-up after DALK in OD, his CDVA was 20/30 with -4.00 DSph/ -1.00 Dcyl @ 50° . With the success of DALK in OD, an uneventful big-bubble DALK procedure was performed in OS. Postoperative recovery was uneventful (figure 1D). At 14 months after the DALK procedure, his CDVA in OS was 20/30 with -3.5 DSph. The average endothelial cell density measured using a specular microscope (EM-3000; Tomey, Tennenlohe, Germany) at the last follow-up was 1327 and 1578 cells/mm² in OD and OS, respectively.

DISCUSSION

XP is characterised by hypersensitivity to ultraviolet (UV) radiation, with defective repair of DNA damage caused by short-wavelength radiation. Ocular manifestations involve the eyelids, conjunctiva, sclera and cornea. Corneal opacification may result from epithelial/stromal changes or may be secondary to endothelial decompensation as a result of damage owing to chronic sun exposure.^{4 7} Anterior lamellar keratoplasty has been opted

for as an alternative to PKP for various diseases not involving the endothelium due to its advantage of lack of endothelial rejection, which is one of the major causes of graft failure.⁸

The literature has shown that in a majority of cases all the layers of the cornea are involved. The chronology of involvement of these layers is difficult to predict. The decision about the type of keratoplasty (full thickness or lamellar) depends on the level of corneal involvement. Only small case series or case reports have been reported with variable success following PKP.^{4 9} The long-term success of PKP in patients with XP is questionable because they belong to a younger age group and have a compromised ocular surface and high probability of future UV exposure and its consequences.

Chaurasia and Ramappa⁵ have shown good success with endothelial keratoplasty in a patient who presented with corneal oedema and graft failure in case of anterior lamellar keratoplasty for a corneal scar. Our patient would have developed a vascularised corneal scar probably because of interventions performed to treat OSSN and hence had a good outcome. He had a corneal scar involving the anterior half of the cornea and no signs of endothelial compromise, and hence we considered DALK. In such situations, performing optical coherence tomography may aid in delineating the depth of the corneal scar and in identifying subtle corneal oedema if present. We agree with the fact that the endothelium is affected in patients with chronic exposure to UV rays. We have also seen low average cell density on specular microscopy in patients with XP (1994 cells/mm²) having a clear cornea compared with age-matched controls (2847 cells/mm², unpublished data). Although the endothelial counts were low, the corneal graft remained clear during the follow-up period but long-term follow-up is needed.

In patients with XP, all layers may be affected at different stages of the disease. Depending on the layers involved, selective corneal transplantation can be planned. Anterior lamellar keratoplasty may be considered in these patients with opacity located in anterior corneal layers and no signs of endothelial decompensation, especially those in the younger age group who are more prone to graft rejection and complications associated with long-term topical steroid use. Full thickness PKP can be reserved for patients with a full thickness corneal scar and corneal oedema associated with opacity.

Learning points

- ▶ Xeroderma pigmentosum (XP) is a rare disease that affects the cornea in various forms.
- ▶ In spite of the fact that all the layers of the cornea are affected histopathologically due to ultraviolet exposure, the severity may vary.
- ▶ Deep anterior lamellar keratoplasty may be considered for corneal scarring in patients with XP.

Contributors JCR and PKV participated in the design and conduct of the study. JCR, PKV and VK were involved in the collection, management, analysis and interpretation of the data. JCR and VK prepared the manuscript. PKV, JCR, and VK reviewed or approved the manuscript.

Competing interests None declared.

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