CASE REPORT

Type I keratoprosthesis for visual rehabilitation of patients with xeroderma pigmentosum

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SUMMARY

A 6-year-old girl, a case of xeroderma pigmentosum (XP), presented with a visual acuity (VA) of 20/20 and 20/400 in the right and left eye, respectively. For a diagnosis of vascularised corneal scar, penetrating keratoplasty was performed in the left eye twice and ultimately graft failed following multiple episodes of rejection. Type I keratoprosthesis was performed in the left eye. She continues to maintain VA of 20/40 in the left eye for more than a year. Similarly, a 24-year-old man, a case of XP, presented with VA of finger counting at 1 metre in both eyes. Ocular examination showed bilateral vascularised corneal scar and conjunctivalisation. Type I keratoprosthesis was performed as primary procedure in the left eye. He maintains a VA of 20/30 for more than a year. Type I keratoprosthesis could be a primary procedure for visual rehabilitation in patients with XP with severe ocular surface disease.

BACKGROUND

Xeroderma pigmentosum (XP) is an autosomal recessive disorder characterised by inability to repair DNA damage caused by ultraviolet light due to defective nucleotide excision repair enzymes. Ocular manifestations associated with XP are corneal xerosis and haze, exposure keratitis, limbal stem cell deficiency (LSCD), corneal vascularisation and opacification, epibulbar and lid neoplasms.¹ The presence of LSCD as well as corneal neovascularisation puts these patients at high risk for allograft rejection as well as surface breakdown.² Boston type I keratoprosthesis is a viable option in patients with repeated graft failure and/or with severe ocular surface disease.³ These cases report good visual outcome in patients with XP with corneal involvement using type I keratoprosthesis after repeated graft failure as well as when used as a primary procedure.

CASE PRESENTATION**Case 1**

A 6-year-old girl, known case of XP, presented in March 1995 with photophobia and watering in both eyes and decreased vision in the left eye since 5 years. She had a visual acuity (VA) of 20/20 and 20/400 in the right and left eye, respectively. Ocular examination revealed superficial corneal vascularisation and stromal scarring in both eyes. In the right eye scarring was more in the periphery of the cornea (figure 1A) and in the left eye it was central with conjunctivalisation of the nasal cornea (figure 1D). The patient was diagnosed with

vascularised corneal scar in both eyes with partial LSCD in the left eye, and she was advised penetrating keratoplasty (PK) in the left eye. PK was performed in the left eye in 1999. Postoperatively, she maintained a VA of 20/60 in the left eye for 1 year, when she developed acute allograft rejection, and subsequently corneal graft failed despite intense steroid therapy. Meanwhile, the right eye developed progressive conjunctivalisation approaching to visual axis and VA dropped to 20/160. The right eye pannus resection along with conjunctivolimbus autograft and amniotic membrane grafting was performed in 2001 and postoperatively VA improved to 20/40. Ten months postoperatively, impression cytology was taken for both eyes, which revealed partial LSCD in the left eye. A repeat PK was performed in the left eye in 2001 and the patient was kept on oral cyclosporine 50 mg twice a day considering a high risk of graft rejection. A VA of 20/50 was maintained in the left eye for 10 months following PK. She again sought an opinion in 2002 and was diagnosed to have acute allograft rejection despite being on oral cyclosporine. Intense topical and systemic steroid therapy was given and the graft survived this episode of rejection. She had an uneventful period until 2004 when the left eye developed rejection and graft failed subsequently (figure 1E). Over a period of 4 years, the right eye developed recurrence of conjunctivalisation and corneal scarring and vascularisation progressed to a stage limiting VA to finger counting (FC). She underwent deep anterior lamellar keratoplasty in the right eye in 2005. Postoperatively she had a VA of 20/30, which was maintained for a period of 6 years and slowly started declining as graft developed progressive vascularisation and opacification from the periphery of the cornea; however, central corneal graft remained relatively clear (figure 1B). As the left eye experienced multiple episodes of rejection, keratoprosthesis was considered as an option for visual rehabilitation. An extracapsular cataract extraction with posterior chamber intraocular lens implantation and type I Boston keratoprosthesis was performed in the left eye in 2011 and postoperatively she had a VA of 20/40 which was stable for more than a year.

In 2013, she presented to emergency services with a 2-day history of pain and redness in the right eye. Her VA declined to FC at 1 metre in the right eye and ocular examination revealed graft infiltrates (figure 1C). The left eye had keratoprosthesis in place with early posterior capsular opacification and maintaining a VA of 20/40. The right eye corneal scrapings grew *Pseudomonas aeruginosa* sensitive to all

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Figure 1 (A) Clinical photograph of the right eye showing corneal scarring, vascularisation and conjunctivalisation on nasal side (1999). (B) Right eye 1 year after deep anterior lamellar keratoplasty showing opacification of the graft in superotemporal quadrant (2006). (C) Same eye showing graft infiltrates (arrow) and opacification of graft all around in the periphery (D). Left eye clinical photograph revealing leucomatous corneal scar with vascularisation and conjunctivalisation (2001). (E) Left eye corneal graft failed due to allograft rejection after two penetrating keratoplasties (2006). (F) Left eye type I Boston keratoprosthesis 1 year postoperatively.

antibiotics except cefuroxime. She was treated with topical antibiotics based on sensitivity and infiltrates resolved over a period of 3 weeks leaving behind a scar. At the last follow-up visit, she had a VA of 20/100 in the right eye and 20/40 in the left eye with corneal scar in the right eye, and in left eye keratoprosthesis was in place with posterior capsule opacification (figure 1F).

Case 2

A 24-year-old man presented with gradual decrease in vision both eyes associated with photophobia for the past 1 year. He was diagnosed to have XP in childhood. There was no other significant history of systemic illness, previous ocular surgery or family history of similar illness. On examination, his VA was FC at 1 metre in the right eye and FC at 1 metre in the left eye. Ocular examination of the right eye revealed corneal stromal scarring, diffuse oedema and conjunctivalisation (figure 2A). The left eye showed diffuse corneal oedema, stromal scarring, vascularisation and conjunctivalisation of the peripheral cornea (figure 2B). A diagnosis of vascularised corneal scar with corneal decompensation and LSCD in both eyes was made. Ultrasound B scans of both eyes were normal. In view of LSCD and corneal vascularisation, he was deemed to be at high risk for graft failure. Based on our experience in a first case, an option of primary implantation of keratoprosthesis (Auro K Pro) was considered. The patient underwent an uneventful extracapsular cataract extraction with posterior chamber intraocular lens implantation and keratoprosthesis implantation in the left eye.

At 1 week follow-up, the unaided VA was 20/25 which is maintained until the last follow-up at 1 year (figure 2C).

DISCUSSION

Patients with XP are poor candidates for PK due to associated lid, tear film abnormalities and corneal vascularisation. Jalali *et al*⁴ has reported high incidence of graft rejection in these patients after PK. Endothelial cells are also found to be dysfunctional in patients with XP⁵ In vivo and histopathological studies have shown decreased cell density and increased coefficient of variation of endothelial cell area in patients with XP as compared with normal individuals. This limits successful surgical outcome of anterior lamellar keratoplasty also.⁵ Keratoprosthesis is a viable option for patients with corneal blindness who are poor candidates for keratoplasty.³ In the first case, two times graft failure occurred following multiple episodes of rejection. Boston type I keratoprosthesis was performed and VA improved to 20/40. Based on our previous experience, we performed primary type I keratoprosthesis in a patient with XP with corneal involvement in the form of vascularisation and conjunctivalisation. VA improved to 20/25. Both cases continue to maintain a good VA without any complications till last follow-up visit. To the best of our knowledge, this is the first report of the use of type I keratoprosthesis in patients with XP and corneal involvement. The type I keratoprosthesis is a viable option for patients with XP with corneal involvement and severe ocular surface disease. It can be considered as a primary procedure of choice for the visual rehabilitation of these eyes.

Figure 2 (A) Clinical photograph of the right eye showing stromal scarring with mild corneal stromal oedema. (B) Left eye revealed corneal scarring, diffuse oedema and conjunctivalisation inferiorly. (C) Left eye 1 year after keratoprosthesis implantation.

Learning points

- ▶ Patients with xeroderma pigmentosum can have severe ocular surface disease including limbal stem cell deficiency, corneal scarring and vascularisation.
- ▶ There is high risk of graft rejection in these patients after penetrating keratoplasty.
- ▶ Type I keratoprosthesis can be considered as a good option for visual rehabilitation in xeroderma pigmentosum patients with ocular surface disease.

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Competing interests None.

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