Heterogeneity of xeroderma pigmentosa: A case series

Prabhjot Kaur Multani[1,*], Dipankar Das[2], Jnanankar Medhi[3], Damaris Magdalene[4], Harsha Bhattacharjee[5]

[1]Resident, [2,3,4]Senior Consultant, [5]Medical Director & Senior Consultant, [1,2]Dept. of Ocular Oncology & Pathology, [3]Dept. of Cornea, [4]Dept. of Pediatric Ophthalmology, [5]Dept. of Cataract & Vitreoretina Services, Sri Sankaradeva Nethralaya, Guwahati, Assam

***Corresponding Author:**
Email: pmultani2008@gmail.com

Abstract

Xeroderma Pigmentosa (XP) results in hypersensitivity to ultraviolet radiation. We intend to report different clinical presentations of XP presenting to a tertiary eye care centre. 3 cases of different age groups i.e. 3, 16 and 25 years old presented with mild dry eye associated with ocular melanosis, ocular surface squamous neoplasia masquerading as conjunctival mass and idiopathic keratitis leading to corneal scarring and spontaneous perforation, respectively. Their clinical presentations, course of disease, response to treatment and outcomes will be discussed in detail. Because of limited literature on ocular presentations of XP, it is extremely vital to have an insight on this subject.

Keywords: Conjunctival melanosis, Keratitis, OSSN, Penetrating keratoplasty, Xeroderma Pigmentosa.

Introduction

Xeroderma Pigmentosa (XP), characterised by defect in DNA repair mechanism, leads to hypersensitivity to ultraviolet radiation. Hebra & Kaposi[(1)] described photosensitivity, photophobia, pigementary changes and early predisposition to malignancy in sun exposed areas of the body. Extreme photosensitivity is the first sign in about 60 percent cases, whereas the rest 40 percent show an increased freckle like pigmentation.([2)] Ocular abnormalities are frequently limited to the ocular surface and eyelids. Chronic sunlight exposure can cause ocular surface neoplasms and keratitis leading to corneal opacification and neovascularization.[(3,4)]

As there are very few reports regarding ocular involvement in XP, this article intends to report three special cases diagnosed with XP and their presentation.

Materials

This is a 3 case series of XP with ocular involvement who presented to our out-patient department at Sri Sankaradeva Nethralaya, a tertiary eye care institute in North east India. Details of each case including its history, clinical presentation, management and their outcomes have been discussed.

Results

Case 1:

A 3-year male child presented with pigmentation in both eyes since 6 months of age with normal birth history and timely achieved milestones. His visual acuity (VA) was normal and anterior segment examination revealed conjunctival melanosis with mild dry eye in both the eyes (Fig. 1). Rest ophthalmic examination was normal. Patient was diagnosed with XP based on typical skin lesions in the sunlight exposed areas (Fig. 2).

He was managed with UV protective glasses, lubricants and regular follow up. His last visit at 6 years of age showed a stable clinical picture.

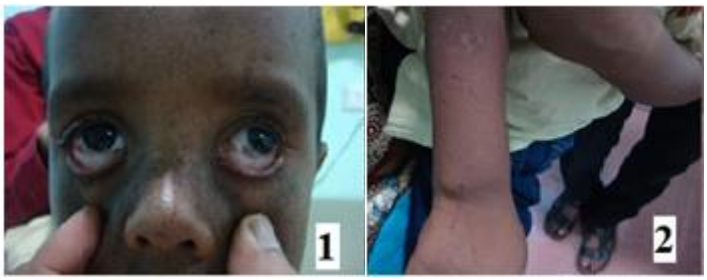

Fig. 1, 2: Conjunctival melanosis in a 6years old child (on last follow up) and dark pigmentation with dry skin of hands and face

Case 2:

A 16-year female presented with mass in right eye since 2 months, gradually increasing in size with history of skin disease since childhood. On examination, conjunctival mass (OD) and conjunctival melanosis (OU) were noted with typical skin lesions suggestive of XP (Fig. 3). UBM showed nasally thickened mass with scleral extension without uveal tract invasion (OD). Thus, a diagnosis of XP with ocular surface mass (OD) was made. She was planned for mass excision with cryotherapy and histopathological examination. Postoperatively, she was started on topical and oral steroids and topical mitomycin C. Histopathological report suggested focal grade III dysplasia with breach of basement membrane and congested dilated vascular channels giving an impression of squamous cell carcinoma (SCC) with microinvasion (Fig. 4). Well along, there was no recurrence. 16 months later, there was similar lesion in the left eye which was timely excised. Presently, she is under observation for 4 years and is maintaining good vision.

Fig. 3 Ocular surface mass (OD) in 16 years old female with typical skin lesions suggestive of Xeroderma pigmentosa

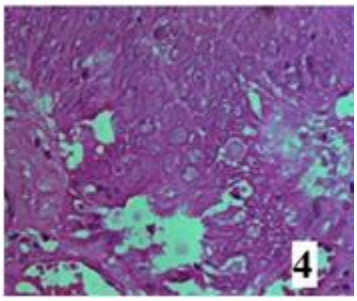

Fig. 4 Histopathology of excised mass showing focal grade III dysplasia with breach of basement membrane and congested dilated vascular channels giving an impression of squamous cell carcinoma (SCC)

Case 3:

A 25-year male presented with painless, progressive diminution of vision in both eyes without history of trauma or contact with vegetative matter. His VA was 20/40 (OD) and hand movement (HM) close to face (OS). There was corneal haze without any inflammation (OD) and corneal ulcer with hypopyon (OS). Patient was started on topical antibiotics, mydriatics and antiglaucoma medication (AGM). Patient was started on topical fluoroquinolones eye drops, mydriatics and antiglaucoma medication timolol maleate 0.5%. Patient reported improvement with medications and was under regular follow up. After 3 months from the initial presentation, there was a recurrent bout of foreign body sensation, pain and redness in both the eyes. Further evaluation revealed ocular surface congestion with corneal haze noted in both the eyes. Patient was then started on lubricants and topical steroid eye drops. After 3 months of topical steroid therapy, eye became quiet. However, the visual acuity in the right eye was 20/60 in the right eye and count finger at 1 meter. Patient was then kept on topical lubricants.

7 months later, patient then presented with loss of vision in the right eye since last 18 days. On examination, sealed central perforation noted in the right eye and total corneal opacity in the left eye (Fig. 5). Systemic evaluation revealed typical skin lesion suggested a diagnosis of Xeroderma pigmentosa (Fig. 6). He underwent therapeutic penetrating keratoplasty in the right eye as it was the only Seeing Eye. However, patient developed graft infection in the post-operative period because of non-compliance to medications. Subsequently there was graft failure with corneal neovascularization in the right eye. He was then planned for and underwent penetrating keratoplasty with extracapsular cataract extraction with K-Pro implantation in the right eye after 1 year. Following surgery patient was comfortable with improvement in vision and could do daily activities. However, vision deteriorated after 1 month follow up and exudates present behind the BCL. On corneal scrapping, infection

with Enterobacter species was present. Patient developed secondary glaucoma and was started on anti-glaucoma medications (AGM). Vitreous cavity was clear with minimal choroidal effusion on USG B scan. On his last visit, 3 years from the initial presentation, ocular condition was stable, intraocular pressure was normal and the vision was hand movement (OD) and counting finger at 1 metre (OS). He was prescribed lubricants and was advised to stop antiglaucoma medications and continue regular follow ups.

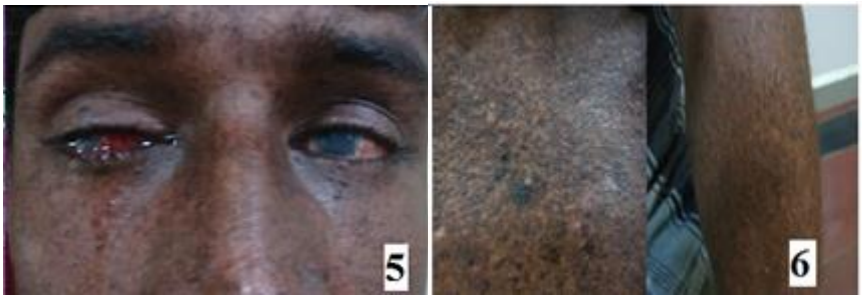

Fig. 5,6: Clinical picture of 25 years old male with corneal perforation (OD) and total corneal opacity (OS) with skin lesions suggestive of XP

Discussion

Ocular involvement in XP is not very well reported in literature. In our case series, ocular involvement in XP ranged from simple conjunctival melanosis not requiring any active treatment to severe recurrent keratitis and corneal ulceration leading to blindness.

Kaur A et al[5] reported a case of XP with eyelid SCC which was a therapeutic challenge because of the non-availability of normal skin required for large defect reconstruction. They used a full thickness trans-midline lateral forehead flap based on superficial temporal artery which had favourable outcomes with satisfactory graft uptake.

Another case reported by Schulze Schwering M et al[6] of a 3 years old child with XP diagnosed with SCC of right ocular surface and left lower eyelid. The child was treated with 3 cycles of intravenous 5-flourouracil (1000 mg/m) for 5 days and cisplatin (50 mg/m) for 2 days every 3 weeks. Patient was completely cured of malignancy following the treatment. The malignancy case reported in our case series was bilateral ocular surface squamous neoplasia (OSSN) in a young female which was completely excised followed by cryotherapy with topical steroids and mitomycin C treatment in the postoperative period. There was complete remission without recurrence thereafter.

Gupta N et al[7] reviewed 14 eyes of 7 cases of OSSN with XP. They found associated limbal stem cell deficiency in 64.3%, dry eye in 100%, conjunctival melanosis in 50% eyes, pseudopterygium in 14.3%, anterior symblepharon in 21.4% and conjunctival inflammatory granuloma in one eye (7.1%). They had emphasized on regular long term follow up due to evidence of late recurrences in XP cases.

Freedman J[8] reported 3 XP cases who underwent corneal transplantation. One case in our series also developed corneal haze in one eye and keratitis with ulceration in the other eye. Therapeutic penetrating keratoplasty, ECCE and Kpro were done which failed subsequently and final VA of hand movement was achieved in the operated eye. Thus, recurrent keratitis without any history of trauma or other systemic illness in a patient with XP can have devastating complications leading to blindness.

Conclusion

Thus, XP has been known to have myriad ocular presentations ranging from dry eye or conjunctival melanosis requiring lubrication and observation to more severe forms such as squamous neoplasia of lids and ocular surface or recurrent keratitis leading to blindness. This article helps to add on to the very limited literature available on the ocular manifestations of this disease. Thus, early identification of XP, regular follow ups, timely diagnosis and management are mandatory for favourable outcomes and to prevent blindness and in these cases.

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