CASE REPORT

Xeroderma pigmentosum with bilateral ocular surface squamous neoplasia and review of the literature

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SUMMARY

Xeroderma pigmentosum is a rare genetic disorder associated with various ocular malignancies. Here we report a single paediatric case of xeroderma pigmentosum with bilateral ocular surface squamous neoplasia (OSSN) presenting with diffuse lesion in one eye and a large mass in the other eye. Diffuse OSSN in one eye was treated with topical chemotherapy using mitomycin-C (0.04%) and the large OSSN in the other eye was treated with a combination of surgery and topical chemotherapy. Long-term follow-up and a multimodality treatment approach are necessary to identify and manage recurrences of OSSN in XP.

BACKGROUND

Xeroderma pigmentosum (XP) is a rare genetic disorder associated with defective DNA repair. It is associated with various cutaneous and ocular surface malignancies such as squamous cell carcinoma, basal cell carcinoma and melanomas. These

malignancies can occur at multiple locations throughout the body on sun exposed areas. Patients of XP need to be evaluated regularly to screen and manage these wide range of malignancies. Ocular surface squamous neoplasia (OSSN) associated with XP has a high chance of recurrence or persistence and requires a multimodality treatment approach.

CASE PRESENTATION

A 12-year-old boy presented with photophobia and slowly growing mass lesions in both eyes (figure 1A) best corrected visual acuity (BCVA) was 3/60, <N36 in the right eye (OD) and no perception of light (NPL) in the left eye (OS). Examination revealed a diffuse mass lesion involving the conjunctiva and 360° of cornea in OD. There was diffuse corneal involvement with xerosis (figure 1B). OS had a large mass due to which the globe was not visible (figure 1C). The largest dimension of mass was 25 mm. Prior informed consent was taken from the attending parent

Figure 1 (A) A young patient with xeroderma pigmentosum showing dry skin and hypopigmented and hyperpigmented patches over the body. (B) Diffuse ocular surface squamous neoplasia (OSSN) in OD. (C) Large OSSN in OS.

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Figure 2 (A) Histopathological examination of the debulked and incision biopsy specimen showing squamous cell carcinoma. (B) Pretreatment lesions in both eyes (OU). Note black arrows showing small melanomas. (C) Fourth month follow-up: post-treatment persistence of tumour in OU (black circle inset). Note black arrows showing enlarged melanomas at post-treatment fourth month follow-up.

(father) to carry out photographic documentation and treatment, since the patient was a minor. Imaging studies did not show orbital infiltration.

Diagnosis of a diffuse OSSN in OD and large OSSN in OS was made.

The patient had numerous hypopigmented macular skin lesions over his body. Dermatological consultation confirmed the diagnosis of XP.

TREATMENT

The diffuse OSSN in the right eye was treated by monotherapy with topical mitomycin-C (MMC) 0.04% four times per day. Four cycles of MMC were given, each consisting of 1 week on and 1 week off treatment. After the four cycles of MMC, there was significant reduction in tumour size and corneal involvement. A further course of two cycles was given. The ocular surface persistently, even after six cycles of MMC, demonstrated residual tumour. The large OSSN mass in the left eye was managed by surgical debulking. On careful excision of the mass, we were able to partially expose the cornea. Complete removal of the tumour was not possible due to its diffuse involvement with the entire ocular surface. Histopathology of the specimen confirmed diagnosis of squamous cell carcinoma (figure 2A). Postoperatively, the patient was started on topical MMC cycles similar to those performed for the right eye. After four cycles of MMC, debulking of the residual tumour mass was carried out and two more cycles of MMC were given postoperatively.

OUTCOME AND FOLLOW-UP

The neoplasia persisted despite six cycles of MMC and debulking surgeries (figure 2B, C and circled area). The patient had two melanomas inferior to the right lower eyelid at presentation, which were found to have grown when he came for his last check up before being lost to follow-up, 4 months after initiation of treatment (figure 2B, C arrows).

DISCUSSION

Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disease with a worldwide prevalence of 1:250 000 and is characterised by defective DNA repair.¹ Exposure to UV radiation leads to varied oculocutaneous manifestations. It starts in early childhood and carries a high risk of various cutaneous and

ocular malignancies such as squamous cell carcinoma, basal cell carcinoma, melanomas and fibrosarcomas.²⁻⁴ Other cutaneous changes include increased photosensitivity, pigmentation and atrophic skin lesions.⁴ Our patient had all the above cutaneous signs though obvious cutaneous malignancies were not present.

Ocular manifestations predominantly consist of conjunctival and corneal involvement, which includes conjunctival xerosis, chronic conjunctival congestion and conjunctival pigmentation. Cornea demonstrates xerosis, limbal stem cell deficiency, band-like nodular keratopathy, scarring and ulcerations.^{3 4} One of the most common ocular surface malignancy in these patients is squamous cell carcinoma.

OSSN is an umbrella term used for all premalignant and malignant epithelial lesions of the conjunctiva and cornea. OSSN is most commonly found in the interpalpebral area at or near the limbus.

Localised well-defined tumours are treated by surgical excision with a no touch technique using lamellar keratectomy with partial sclerectomy (if sclera is involved) with cryotherapy on the conjunctival side.⁵ Topical chemotherapy using MMC 0.04% is used as monotherapy for treatment of diffuse lesions or as an adjuvant after surgical excision. MMC has been shown to decrease the rate of recurrences when used for two to four cycles postoperatively.⁶ Diffuse lesions are difficult to treat and have higher chances of recurrence.^{7 8}

In our case, the diffuse OSSN was treated with topical chemotherapy and large OSSN was treated with a combination of

Learning points

- ▶ Patients of xeroderma pigmentosum (XP) should be screened regularly for early detection and management of a wide range of oculocutaneous malignancies.
- ▶ Ocular surface squamous neoplasia (OSSN) in patients with XP has a high chance of recurrence or persistence and requires rigorous follow-up and a multi-modality treatment approach.
- ▶ Long-term follow-up and a multimodality treatment approach are required to identify and manage recurrences of XP-associated OSSN.

surgery and chemotherapy. Similar to our case, persistence and recurrence of OSSN in patients with XP has been reported in the literature.²

Contributors CK had the idea for this case and report and was involved in the conceptualisation, drafting, analysis, literature review. NR was involved in the drafting, analysis and literature review. AM was involved in the drafting, analysis, literature search and editing. SR was involved in the drafting, literature review and editing.

Competing interests None declared.

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