Content available at: https://www.ipinnovative.com/open-access-journals

IP International Journal of Ocular Oncology and Oculoplasty

Journal homepage: https://ijooo.org/

Case Report

Bilateral ocular surface squamous neoplasia in xeroderma pigmentosum patient - A case report

Sivaraman G[1,*], **Modini Pandharpurar**[1]

[1] *Dept. of Ophthalmology, Sarojini Devi Eye Hospital and regional Institute, Osmania Medical College, Hyderabad, Telangana, India*

ARTICLE INFO

Article history:
Received 31-05-2022
Accepted 25-07-2022
Available online 08-11-2022

Keywords:
Ossn
Ocular surface squamous neoplasia
Nodular ossn
MitomycinC
Cryotherapy

ABSTRACT

A rare case of a 13yr old male patient presented with photophobia in both eyes and gradually progressive painless mass with watering, itching, and redness in the right eye for 2 months. Best-corrected visual acuity in Right eye 20/200 and Left eye 20/120. On examination, Right eye has a 6 x 6 mm fleshy pink oval pedunculated mass lesion arising from the temporal limbus and a 2mm hyperpigmented lesion in the nasal limbus. Left eye has a gelatinous white flat diffuse lesion with the pigmentation of 3mm both in the nasal and temporal limbus. Both eyes' Anterior segment, ocular motility, and fundus are normal. The patient had multiple hyperpigmented spots (freckles) and scaly skin on the face and trunk suggestive of Xeroderma pigmentosum for which he was on irregular treatment. Right eye tumor mass was excised with intraoperative control of surgical margins with adjunctive cryotherapy and sent for histopathology examination, which was confirmed as squamous neoplasia. Postoperatively, Both eyes started on Topical Mitomycin-c 0.02% 4 cycles, and the patient was carefully followed for any recurrence.

1. Introduction

Xeroderma pigmentosum is an autosomal recessive genetic disease that occurs due to defective DNA repair. It is associated with malignancies in the skin and eye such as squamous and basal cell carcinoma. It occurs in multiple areas throughout the body in sun-exposed areas. Ocular surface squamous neoplasia in XP requires multimodality treatment such as topical chemotherapy and surgery. There is a high recurrence rate when both XP and OSSN coexist even with combination therapy with topical drugs and surgery. These patients require long-term follow-up for the management of recurrences.

2. Case Report

A 13-year-old boy patient presented with chief complaints of photophobia in both eyes and gradually progressive painless mass with watering, itching, and redness in the right eye to oculoplastic OPD in Tertiary eye care centre, Hyderabad, Telangana in South India. The patient was referred with ocular complaints from the dermatology department with the diagnosis of Xeroderma pigmentosum. He was diagnosed to have XP 3 years back for which he was on treatment for 2 years and stopped 1 year back. There was no history of any ocular injury or surgery to either eye. Prior informed consent was taken from his parents for documentation and treatment. A complete ocular examination which includes visual acuity, refraction, anterior segment examination by slit lamp, fundus examination, and systemic investigations to rule out HIV are done.

* *Corresponding author*.
E-mail address: sivagsr94@gmail.com (Sivaraman G).

https://doi.org/10.18231/j.ijooo.2022.049

On examination, his best-corrected visual acuity in the right eye was 20/200 and in the left eye was 20/120. Anterior segment examination under a slit lamp revelated a 6x6mm fleshy pink oval pedunculated mass lesion in the temporal limbus and a 2mm hyperpigmented lesion over the nasal limbus in the right eye (Figure 1) and a gelatinous white flat diffuse lesion with pigmentation 3mm over nasal and temporal limbus in the Left eye (Figure 2). Both eyes' cornea, iris, pupil, lens, fundus, and ocular motility are normal. The patient had multiple hyperpigmented spots (freckles) and scaly skin involving the face, eyelid margin, and trunk suggestive of xeroderma pigmentosum.(Figure 3)

Diagnosis of nodular and minimally diffuse OSSN in the right eye and Diffuse OSSN in the left eye with xeroderma pigmentosum was made.

The patient was advised to undergo excision of nodular OSSN in the right eye. After a complete preoperative workup and fitness from an anesthetist. The right eye tumor mass was excised with intraoperative control of surgical margins with adjunctive cryotherapy (Figure 4A-C) and sent for histopathology examination which was confirmed as squamous neoplasia. Postoperatively right eye started on topical mitomycin-c 0.02 % 4 cycles. The diffuse OSSN in the left eye was treated by monotherapy with Topical Mitomycin-c 0.02 % 4 cycles. The patient was carefully followed for any recurrences in both eyes

The patient was followed after 1 month. The right eye shows congestion on the nasal side and no residual tumor growth on the temporal side (Figure 5). The patient was followed after 6 months. The right eye was quiet and had no recurrence and the left eye showed persisting diffuse OSSN despite 4 cycles of chemotherapy.

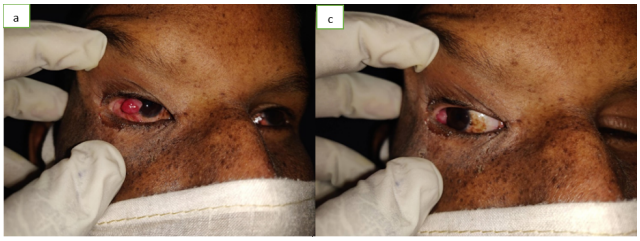

Fig. 1: Right eye, **a:** nodular ossn over temporal limbus; **b:** Diffuse ossn over nasal limbus

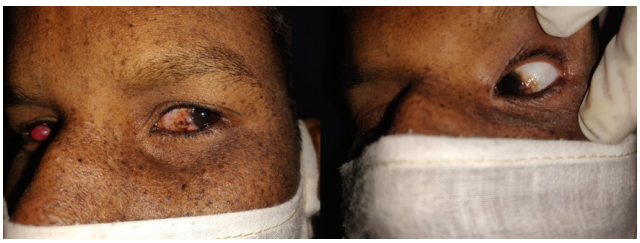

Fig. 2: Left eye diffuse ossn; **a:** nasal and **b:** temporal limbus

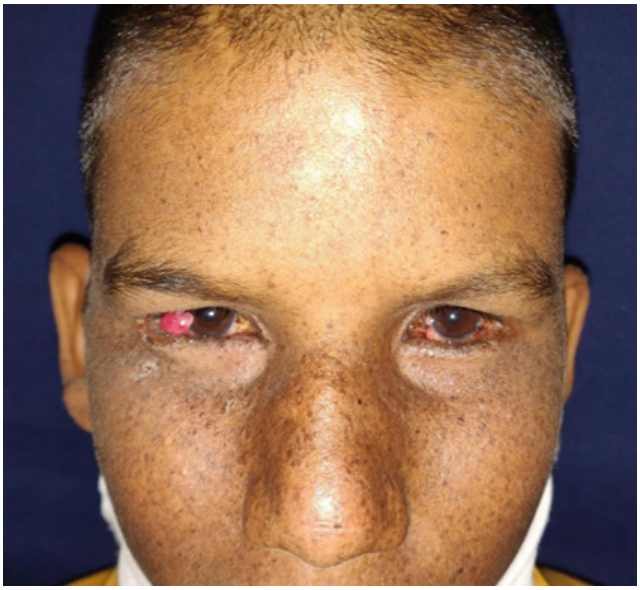

Fig. 3: Oculocutanous lesion in xp and ossn patient.

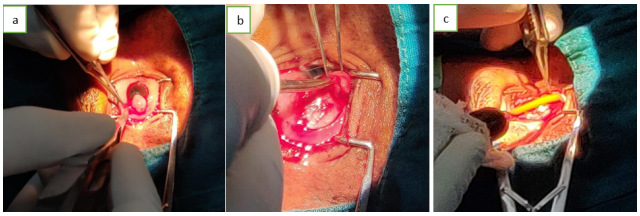

Fig. 4: Intraoperative; **a**: Surgical excision; **b**: Margin excision **c**: Cryotherapy.

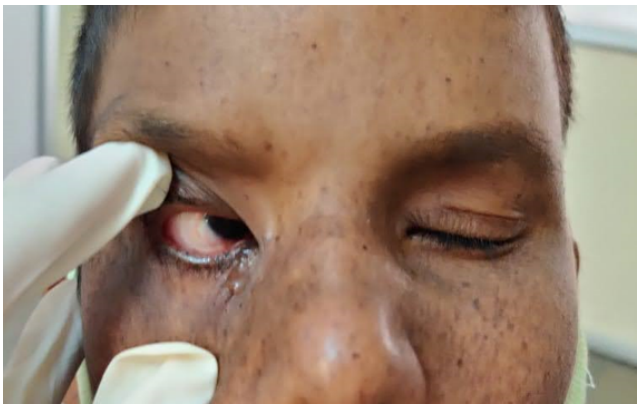

Fig. 5: Postoperative re 1 month

3. Discussion

Xeroderma pigmentosum is associated with oculocutanous malignancies such as basal cell carcinoma, squamous cell carcinoma, and rarely melanoma which are triggered by UV radiation.[1] Localized well-defined tumors are treated by surgical excision and adjunctive treatment. Diffuse lesions are difficult to treat and have a high chance of recurrence. Malignancy associated with XP has a high chance of metastasis. OSSN's usual age onset is between 55 to 65yrs, but patients with XP have a very early onset within 10yrs (range 1-30yrs).[2]

XP is an autosomal recessive genetic disease and is characterized by the inability to repair UV-induced DNA damage leading to malignancy.[3]

Ocular manifestations in XP include xerosis, congestion, pigmentation, keratopathy, ulcerations, scarring, and malignancy such as OSSN, rarely melanoma.[4] Cutaneous changes in these patients include photosensitivity and multiple hyperpigmented spots such as freckles.[5,6]

OSSN with localized nodular variety is treated by surgical excision with a no-touch technique and cryotherapy at the margin, if corneal or sclera are involved lamellar keratectomy and partial sclerectomy are done respectively.[7] Multimodality approaches like combined surgery and topical chemotherapy are usually preferred for nodular or large lesions.[8] Topical chemotherapy post-operative prevents recurrence. Topical chemotherapy is used as mono therapy in a diffuse[9] lesion or as an adjuvant after surgical excision Most commonly used chemotherapy is Mitomycin 0.04% 4 to 8 cycles, other topical drugs like interferon-alpha 2b 1MIU/ml are also used. Histopathological examination is a must for all surgically excised lesions to confirm the malignancy and margin free from tumor.[10]

Patients with XP should be screened regularly for early detection of oculocutaneous malignancy. Management of this patient should be focused on early detection, multi modality treatment, and frequent follow-up. Preventive measures such as sun glass, and covering the sun-exposed area with cloths to minimize the UV radiation-induced DNA damage.[11]

4. Conflict of Interest

The authors declare that there are no conflicts of interest in this paper.

5. Source of Funding

None.

.

References

1. Arya D, Das S, Gandhi A. Ipsilateral presentation of ocular surface squamous neoplasia and conjunctival melanoma in xeroderma pigmentosum: A rare occurrence. *Indian J Ophthalmol*. 2019;67(12):2068–71. doi:10.4103/ijo.IJO_1449_19.
2. Schubert S, Lehmann J, Kalfon L, Slor H, Falik-Zaccai TC, Emmert S, et al. Clinical utility gene card for: Xeroderma pigmentosum. *Eur J Hum Genet*. 2014;22(7). doi:10.1038/ejhg.2013.233.
3. Van Steeg H, Kraemer KH. Xeroderma pigmentosum and the role of UV-induced DNA damage in skin cancer. *Mol Med Today*. 1999;5(2):86–94. doi:10.1016/s1357-4310(98)01394-x.
4. Takebe H, Nishigori C, Tatsumi K. Melanoma and other skin cancers in xeroderma pigmentosum patients and mutation in their cells. *J Invest Dermatol*. 1989;92(5):236–8.
5. Kalamkar C, Radke N, Mukherjee A, Radke S. Xeroderma pigmentosum with bilateral ocular surface squamous neoplasia and review of the literature. *BMJ Case Rep*. 2016;p. bcr2016215364. doi:10.1136/bcr-2016-215364.
6. Goyal JL, Rao VA, Srinivasan R, Agrawal K. Oculocutaneous manifestations in xeroderma pigmentosa. *Br J Ophthalmol*. 1994;78(4):295–7.
7. Sarici AM, Arvas S, Pazarli H. Combined excision, cryotherapy, and intraoperative mitomycin C (EXCRIM) for localized intraepithelial and squamous cell carcinoma of the conjunctiva. *Graefes Arch Clin Exp Ophthalmol*. 2013;251(9):2201–4.
8. Besley J, Pappalardo J, Lee GA, Hirst LW, Vincent SJ. Risk factors for ocular surface squamous neoplasia recurrence after treatment with topical mitomycin C and interferon alpha-2b. *Am J Ophthalmol*. 2014;157(2):287–93.
9. Singh V, Maurya RP, Pal K, S S, Singh V, Anand K, et al. Ocular Surface squamous neoplasia: An overview. *IP Int J Ocul Oncol Oculoplasty*. 2022;8(1):1–11.
10. Gupta N, Sachdev R, Tandon R. Ocular surface squamous neoplasia in xeroderma pigmentosum: clinical spectrum and outcome. *Graefes Arch Clin Exp Ophthalmol*. 2011;249(8):1217–21.
11. Lehmann AR, Mcgibbon D, Stefanini M. Xeroderma pigmentosum. *Orphanet J Rare Dis*. 2011;6:70. doi:10.1186/1750-1172-6-70.

Author biography

Sivaraman G, Post Graduate https://orcid.org/0000-0002-9777-5042

Modini Pandharpurar, Professor

Cite this article: Sivaraman G, Pandharpurar M. Bilateral ocular surface squamous neoplasia in xeroderma pigmentosum patient - A case report. *IP Int J Ocul Oncol Oculoplasty* 2022;8(3):223-225.