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CASE REPORT

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Insights into Ocular Surface Squamous Neoplasia – A Case Study in Diagnosis and Management

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Abstract

We report a case of Ocular surface squamous neoplasia (OSSN) in a thirty-year-old female with Xeroderma pigmentosa who presented with complaints of pain, irritation, photophobia, foreign body sensation and a small mass, gradually increasing in size in the left eye. Examination revealed multiple diffuse mottled erythematous pigmentation over face suggestive of Xeroderma pigmentosa. On Slit lamp examination right eye was unremarkable with 6/6 vision whereas left eye with 6/12 vision showed a round, 1x1 cm lobulated, non-mobile, hard lesion with abnormal vasculature on nasal bulbar conjunctiva in interpalpebral area. CT orbit revealed a lobulated lesion involving infero-medial aspect of anterior surface of sclera showing contrast enhancement with no obvious deeper extensions and negative HIV test. She underwent excisional biopsy, followed by application of Mitomycin C and cryotherapy and later with an amniotic membrane graft on her left eye. Histopathological analysis revealed moderately differentiated invasive squamous cell carcinoma that extended into all horizontal and deep surgical margins. As a result, adjuvant treatment with topical interferon alpha 2B was initiated. At the 10-month postoperative visit, her visual acuity was 6/9, improving to 6/6 with a pinhole test, and no clinical evidence of recurrent squamous cell carcinoma was observed.

Keywords: Conjunctiva; Ocular surface; Neoplasia; Mitomycin C; Xeroderma pigmentosa

1 Introduction

Ocular surface squamous neoplasia encompasses a variety of precancerous and cancerous growths that affect the epithelial tissues of the conjunctiva and cornea. This condition includes lesions ranging from mild dysplasia, Carcinoma in situ (CIN) to invasive squamous cell carcinoma, all originating from the surface cells of the eye.[1–3] Previously used terms for this condition include

intraepithelial epithelioma, Bowens disease, and Bowenoid epithelioma.[4] Ocular surface squamous neoplasia (OSSN) typically presents unilaterally and is most commonly observed in middle-aged and older adults. In rare instances, it can occur bilaterally, especially in individuals with compromised immune systems. Symptoms generally include redness and irritation of the eye, though vision remains unaffected unless the lesion extends into the central cornea. Key risk factors for OSSN involve prolonged ultraviolet light exposure, infection with HPV type 16, and immune system deficiencies. Research has identified connections between AIDS, UV damage, and HPV infection in the development of OSSN. Additionally, systemic conditions such as Xeroderma pigmentosum (XP) and Papillon–Lefèvre syndrome are associated with an increased risk of OSSN.

2 Case Details

A 30-year-old healthy female presented with a mass in her left eye that had been developing over the past 4 months. She initially noticed a small mass, which has since grown accompanied by pain, irritation, light sensitivity and foreign body sensation. The patient reported a history of itchy facial lesions since childhood and had undergone pterygium excision in the same eye 4 years earlier.

Examination revealed multiple diffuse mottled pigmentation with erythema over the malar area of face. Freckles were seen on the anterior aspect of neck and chest suggestive of Xeroderma pigmentosa. Similar pigmentation was noted in younger sibling. No neurological

abnormalities detected. Her uncorrected visual acuity was 6/6 in OD and 6/9 in OS that improved to 6/6 with -0.50 DC at 90^0. On Slit lamp examination her right eye was unremarkable whereas her left eye showed a round, 1x1 cm lobulated, non-mobile lesion on nasal bulbar conjunctiva in the interpalpebral area, that was hard in consistency with abnormal vasculature. Indirect ophthalmoscopy and B-scan showed no intraocular extension.

3 Clinical Course

The patient's clinical presentation strongly suggested squamous cell carcinoma. HIV testing returned negative, and a CT scan of the orbit revealed lobulated lesion involving the infero-medial aspect of anterior surface of sclera showing contrast enhancement with no obvious deeper extensions. One week after the initial presentation, the patient underwent an excisional biopsy following the 'no – touch' technique, followed by cryotherapy performed in a double freeze slow thaw method on the conjunctival margins, intraoperative application of 0.04% mitomycin C, and the placement of an amniotic membrane graft. Histopathological analysis confirmed the presence of invasive, moderately differentiated squamous cell carcinoma, with tumor extension into all horizontal and deep surgical margins.

Adjuvant therapy was initiated with topical interferon alpha-2b (1 million units/mL) applied four times daily, and this regimen was planned to continue for at least 12 months due to the patient's history of recurrence. Additionally, 10 million units of interferon alpha-2b was injected subconjunctivally every month for 6 months. During the 4-month follow up appointment, corrected visual acuity in the left eye was 6/12 with correction with no signs of recurrent squamous cell carcinoma. By the 10-month postoperative visit, corrected visual acuity had further improved to 6/9.

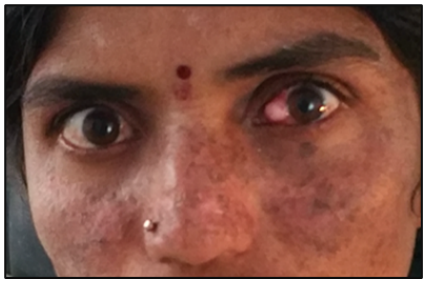

Fig 1. Multiple diffuse mottled pigmentation with erythema over the malar area of face

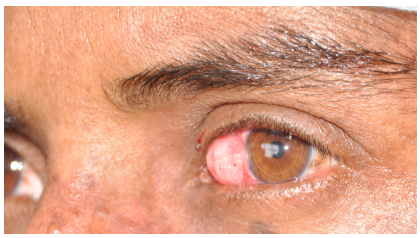

Fig 2. Slit lamp photographs of the left eye. Note the elevated, gelatinous appearance and abnormal vascularity of the lesion

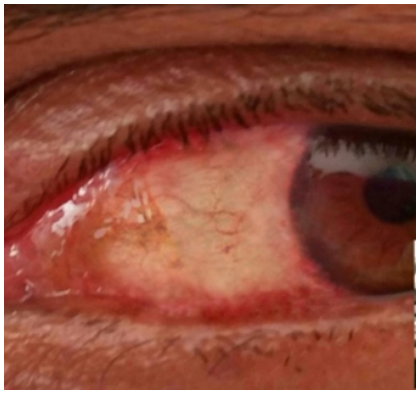

Fig 3. First week post op

4 Discussion

Xeroderma pigmentosum is a rare hereditary disorder caused by autosomal recessive genes. It is marked by extreme sensitivity to ultraviolet (UV) radiation, leading to severe damage to the skin and eyes, which can result in cancer. In some individuals, the condition also involves progressive neurological decline.

Treating xeroderma pigmentosum (XP) is difficult because it affects multiple organs and systems and often involves extensive tissue damage by the time it is diagnosed.[5] Malignant tumors can develop by the third or fourth year of life.[6] Timely diagnosis and prompt application of strict sun-protection measures can significantly increase the lifespan of individuals with XP.[5,7] Approximately two-thirds of individuals with xeroderma pigmentosum who do not receive adequate management do not survive past age 20. In areas with high levels of sunlight and limited access to modern medical care, children with XP who do not comply with sun-protection measures may have a life expectancy of around 10 years.

Individuals with XP must stay away from all sources of UV light, including sunlight, fluorescent, halogen, and mercury-vapor lights. They should always use high SPF sunscreens, UV-absorbing eyewear and protective clothing. Topical therapies such as 5-fluorouracil or imiquimod are suitable for premalignant skin lesions, whereas surgical excision is the recommended course of action for malignant neoplasms affecting the skin, tongue, eyelids, conjunctiva, and cornea. Using a bland ointment at night and eyedrops comprising methyl cellulose or quinodine are important aspects of eyecare.

4.1 Take Home Points

- HIV testing is mandatory in young patients with ocular surface squamous neoplasia.
- It is important to note that individuals with XP, who are adequately protected from sunlight exposure, are at increased risk of developing vitamin D deficiency. Therefore, routine supplementation with vitamin D is recommended.

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