



Case Report

Synchronous malignancy in xeroderma pigmentosum and issues in management

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Xeroderma pigmentosum (XP) is a genetic disorder of autosomal recessive transmission, characterized by photosensitivity, pigmentary changes, premature skin aging and malignant tumour development due to cellular hypersensitivity to ultraviolet radiation. This hypersensitivity results from a defect in DNA repair. The XP is known to have multiple cutaneous malignancies in sun exposed areas, which are usually treated by surgery. The surgical excision of the tumor should be complete and oncologically safe, as the use of postoperative radiotherapy is controversial. This case report describes synchronous basal cell carcinoma and squamous cell carcinoma occurring at the head and neck region of a young individual with xeroderma pigmentosum. Here, attempts have been made to highlight the difficulties encountered in such individuals while managing lesions located in close proximity to each other.

Key words: Xeroderma pigmentosum, synchronous malignancies, squamous cell carcinoma, basal cell carcinoma, hypersensitivity

INTRODUCTION

Xeroderma pigmentosum (XP) is an autosomal recessive genetic disorder of DNA repair in which the ability to repair damage caused by ultraviolet (UV) light is deficient, and this deficiency is due to mutated and non functioning nucleotide excision repair (NER) enzymes. Those affected are hypersensitive to UV light produced by the sun and even with a short exposure, they exhibit various skin lesions ranging from dry, flaking skin and pigmented spots to variety of skin cancers. These skin cancers are usually metachronous but they can be rarely synchronous (Vandana, 2011; Grampurohit, 2011). Individuals with XP are about 1000 times more likely to develop skin cancer than individuals without the disorder (Mohanty, 2001; Patil, 2007). This case report describes synchronous basal cell carcinoma and squamous cell carcinoma occurring at the head and neck region of a young individual with xeroderma pigmentosum. Here, attempts have been made to highlight the difficulties encountered in such individuals while managing lesions located in close proximity to each other.

CASE DETAILS

A 15-year-old male patient who was diagnosed with xeroderma pigmentosum ten years back presented with ulcer over lower lip that endured for the last three months. The patient also complained of multiple blackish spots over that persisted on his face for many years. However, one of the spots on right cheek had recently increased in size and started to ulcerate (Figure 1). Family history was not contributory. The general physical examination showed no neurological deficit. There was no history of neck swelling or any other complaints related to the oral cavity or throat.

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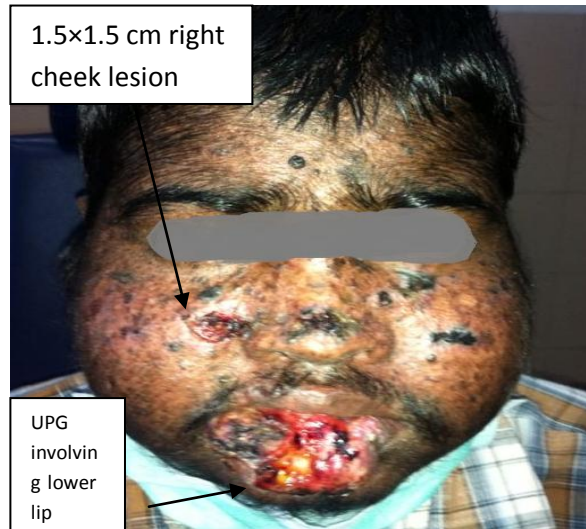


Figure 1. Preoperative photograph showed both lesion in close proximity



Figure 2. Post excision (Black arrow showed advancement direction)



Figure 3. Primary closure by advancement

Examination of eyes showed visual acuity 6/18 both side with bilateral macular corneal opacity and blepharitis. There was no history of neck swelling or any other complaints related to oral cavity or throat.

Biopsy from lip ulcer was reported as moderately differentiated squamous cell carcinoma and that from cheek as basal cell carcinoma. Detailed clinical workup for possible metastasis revealed nothing particular.



Figure 4. Wound dehiscence on 7th post operative day (black arrow)

Afterwards wide local excision of both carcinomatous lesions and reconstruction by local rotation flaps were performed. It was made sure that surgical margins were adequate all around the lesions. The excision of lip lesion eventually resulted in loss of nearly 80% of lower lip (Figure 2). Since the tumour did not involve the adjacent gingival or alveoli, these structures were preserved along with overlying periosteum. After adequate mobilisation of the surrounding cheek and remaining lip, tensionless primary closure was done (Figure 3). Although the patient had good recovery in immediate postoperative period, the wound gaped to certain extent after suture removal on postoperative seventh day. Given the quantity and quality of surrounding skin, no further reconstruction was planned and the wound was left for secondary healing, which subsequently resulted in reasonable appearance (Figure 4). Histopathological examination of the surgical specimens showed moderately differentiated squamous cell carcinoma in lip specimen and no residual primary tumor in cheek specimen. Two cycles of chemotherapy in form of 5 fluorouracil was given.

DISCUSSION

The XP was first described in 1874 and Kaposi coined the term in 1882 because of its characteristic dry and pigmented skin. The basic defect in xeroderma pigmentosum is in nucleotide excision repair, leading to deficient repair of DNA which has been damaged by UV radiation. The use of sunscreens with other sun-avoidance methods can minimize UV-induced damage in patients with xeroderma pigmentosum. The XP patient is known to develop multiple cutaneous malignancies in sun exposed areas, which are usually treated by various surgical methods such as electro-desiccation and curettage, or surgical excision. The

selection of approach for surgical excision will depend on extends of tumor as we did in our case. There are certain peculiar issues related to surgery in this patient. The surgical excision of the tumor should be complete and oncologically safe, as the use of postoperative radiotherapy is controversial (Ben, 2011). For premalignant lesion and in post-operative cases chemotherapy is indicated (Rohan 2013) but few case report mentioned clearance of malignant lesion by chemotherapy only (Jordi, 2009) In literature, palliative radiotherapy advised for anticipated prolong life expectancy in view of non availability literature showed long term side effect in XP. Shetty et al highlight the importance of histopathological sampling of each lesion separately prior to surgery in patients who have multiple lesions due to XP. This report considers the difference in behaviour of various types of skin malignancies and criticizes the surgeons inclination towards conservative approach. The reason is that similar pathology exists in lesions of close proximity patients with XP having multiple lesions (Shetty, 1999;, Rahul, 2013).

Surgical resection should aim to achieve negative surgical margins and, thus, to excise the surrounding regions which are likely to have the neoplastic molecular alterations already during the histopathological examination and subsequent immunohistochemical assessment.

The issue of reconstruction of such a radically resected lip, almost involving the whole of the lower lip is also complicated by the poor quality of the sun exposed skin in such patients (Terziqi, 2009). As for particular patient concurrent resection of the cheek lesion means not only the difficulty in the reconstruction of cheek area and also results in the narrowing of the spectrum of local rotation flaps which could have been otherwise used for lower lip reconstruction.

The patient had a relatively small lesion in cheek which was distinctly located from the lip lesion so that excision could be done separately. Since the surgical resection involved nasolabial folds, both the surgical defects could be closed separately. There was no apparent tension on margins as local flaps were mobilized and rotated as based on facial-angular vascular bundle. This case report indicates that suture removal could be delayed until up to 10 days so that adequate time may be allowed for skin healing, fibrosis and maturation.

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