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Case Report

Xeroderma Pigmentosum with synchronous Squamous cell carcinoma and Basal cell carcinoma

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Abstract:

Xeroderma pigmentosum (XP) is a rare autosomal recessive genodermatosis due mutation in excision repair genes. It is characterized by marked photosensitivity, skin pigmentation, predisposition for development of malignant tumors and sometimes progressive neurological degeneration. Cutaneous tumors develop due to exposure to UV light. Here we report a case of a 23-year young female of known case of Xeroderma Pigmentosum presented with multiple hyperpigmented lesions on face, back and both forearm. On excisional and histopathological examination, tissue from back revealed Basal cell carcinoma (BCC) while tissue from left forearm revealed squamous cell carcinoma (SCC). XP is a rare genodermatosis with accumulation of unrepaired DNA damage due to defect in mismatch repair system. There are eight variants XP A-G and XP, with varying mutation in nucleotide excision repair. Exposed skin is particularly affected due to UV-radiation exposure. SCC, BCC and melanoma are common cutaneous tumors but simultaneous occurrence is rare. There are few case reports describing synchronous development of cutaneous and ocular SCC, cutaneous SCC, BCC and melanoma. Patient require frequent follow-up, self-examination and biopsy of any suspected lesion for early identification of malignancies.

Keywords: Autosomal recessive, Basal cell carcinoma, genodermatosis, squamous cell carcinoma.

Introduction

Xeroderma Pigmentosum (XP) first described by Hebra and Kaposi in 1874. Xeroderma pigmentosum (XP) is a rare autosomal recessive genodermatosis due to mutation in excision repair genes.[1] It is characterized by marked photosensitivity, skin pigmentation, predisposition for development of malignant tumors and sometimes progressive neurological degeneration.[2] Cutaneous tumors develop due to exposure to UV light. Patients with xeroderma pigmentosum (XP) near the age of 20 have more than a 1000-fold increased risk of developing skin cancer.[3] These malignancies are aggressive, grow rapidly, metastasize early and can be fatal. Early detection and treatment of cutaneous cancers are crucial to reduce morbidity and mortality. However, the most effective preventative measure for xeroderma

pigmentosum is genetic counseling, which plays a key role in reducing the risk of the condition being passed on.[4] In this case of 23 year young female who developed synchronous cutaneous Squamous cell carcinoma (SCC) and Basal cell carcinoma (BCC).

Case report:

A 23 year young female of known case of Xeroderma Pigmentosum presented with multiple hyperpigmented lesions on face, back and both forearm [Figure 1].

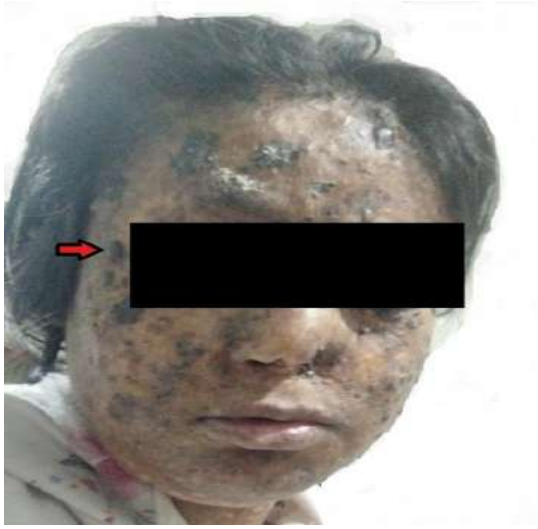

Figure [1] - Patient with multiple pigmented lesions on the face

Largest lesion was present on left forearm measured 8 x 6 cm. There were no neurological symptoms. Patient sister also had similar lesions on face and arms.
Excision of multiple lesions over left forearm and back with split thickness skin graft was performed. We received skin covered tissue in multiple fragments with surface nodular and ulcero-proliferative lesions.

On microscopy tissue from face revealed a tumor comprising of nests of atypical basaloid cells with peripheral palisading and cleft artefacts. Mitosis were frequent with atypical forms. Melanin pigment deposition was also there. Diagnosis of Nodular BCC was given [Figure 2A and 2B].

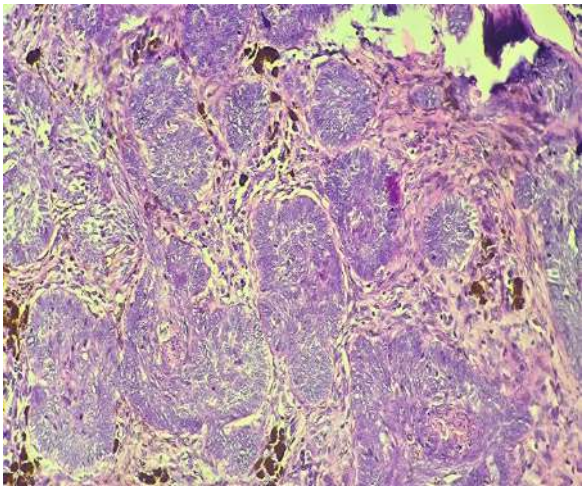

Figure.[2A]- Face nodule (Hematoxylin and eosin, 200x).:- Basal cell carcinoma with nests of atypical basaloid cells.

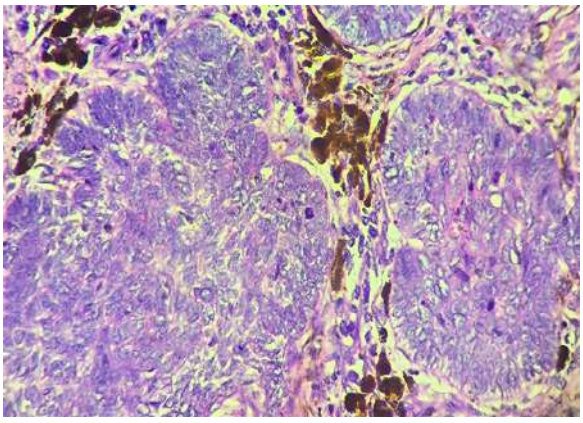

Figure.[2B]- Face nodule (Hematoxylin and eosin, 400x).:- Basal cell carcinoma -Atypical basaloid cells with mitosis and melanin pigment deposition in stroma.

Tissue from left forearm revealed infiltrating tumor comprising of irregular nests of dysplastic squamous cells. Diagnosis of moderately differentiated SCC was given [Figure 3].

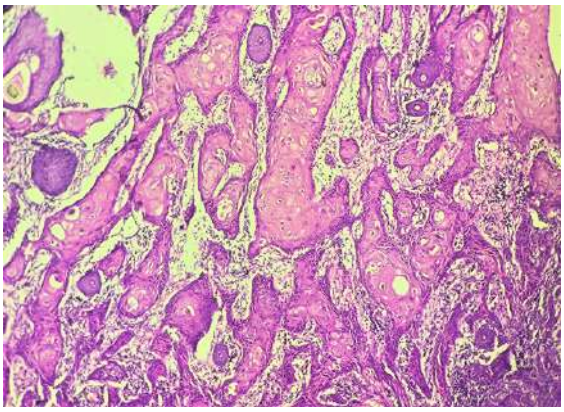

Figure [3]- left forearm swelling- (Hematoxylin and eosin, 100x).

Discussion:

Xeroderma pigmentosum (XP) is a rare genodermatosis with accumulation of unrepaired DNA damage due to defect in mismatch repair system.[5] There are eight variants XP A-G and XP, with varying mutation in nucleotide excision repair . Exposed skin is particularly affected due to UV-radiation exposure.[6] SCC, BCC and melanoma are common cutaneous tumors but simultaneous occurrence is rare. There are few case reports describing synchronous development of cutaneous and ocular SCC, cutaneous SCC BCC and melanoma. This patient had synchronous SCC and BCC. The differential diagnosis of xeroderma pigmentosum (XP) should include two other conditions caused by mutations in excision repair genes: Cockayne's syndrome (CS) and Trichothiodystrophy (TTD). However, skin cancer linked to high sensitivity to UV radiation is commonly seen in XP, but not in CS or TTD. Xeroderma pigmentosum (XP) is diagnosed based on history and clinical examination.[7] Identification of specific mutation require gene sequencing. Patient require frequent follow-up, self-examination and biopsy of any suspected lesion for early identification of malignancies.

Conclusion:

A uncommon, hereditary condition called Xeroderma Pigmentosum (XP) is characterized by an increased sensitivity to ultraviolet (UV) radiation. This condition can cause significant skin damage and raise the chance of developing skin malignancies, such as basal cell carcinoma (BCC) and squamous cell carcinoma (SCC). This case report illustrates the substantial risk of multiple skin cancers in people with XP by highlighting the incidence of both SCC and BCC in a patient with the illness. Preventing life-threatening consequences requires vigorous care of skin lesions, frequent dermatological surveillance, and early detection. People with XP should receive thorough care, including regular skin cancer screenings and teaching on UV protection, due to the genetic origin of the illness. The occurrence of simultaneous SCC and BCC in this instance emphasizes the necessity of prompt action to control and track these tumors, as well as for investigation into better treatments and preventative measures for skin cancers linked to XP.

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