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Malignant melanoma in a case of xeroderma pigmentosum

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Abstract

Xeroderma pigmentosum is a rare autosomal recessive disorder characterized by photosensitivity, pigmentary changes, premature skin aging and malignant tumour development due to cellular hypersensitivity to ultraviolet radiation resulting from a defect in DNA repair. A 47 years old female presented with an increase in size and ulceration of the pre-existing pigmented macules over the face. The patient had these hyperpigmented macules since early childhood, throughout the body, more on sun exposed areas. Histological examination of ulcero-proliferative lesions showed features of malignant melanoma while the surrounding hyperpigmented macules revealed changes of xeroderma pigmentosum. This case highlights a rare clinical entity presenting with a complication which is even rarer.

Introduction

Xeroderma pigmentosum is a rare autosomal recessive disorder. [1] It is seen in all races worldwide and has equal sex incidence. It occurs with an estimated frequency of 1: 250,000 in United states and is somehow more common in Japan, but its incidence is not significant in India. [2,3,4] It is characterized by photosensitivity, pigmentary changes, premature skin aging and malignant tumour development which commonly include squamous cell carcinoma, basal cell carcinoma and rarely fibrosarcoma. Malignant melanoma arises in only about 3% of patients with xeroderma pigmentosum. The basic defect underlying the clinical manifestation is a nucleotide excision repair (NER) defect leading to defective repair of DNA damaged by UV radiation.[3]

Here is a case report of malignant melanoma arising in a patient of xeroderma pigmentosum.

Case Report

A 47 years old female reported with chief complaints of freckles and hyperpigmented macules all over the body as well as photo-sensitivity and increased watering from eyes since early

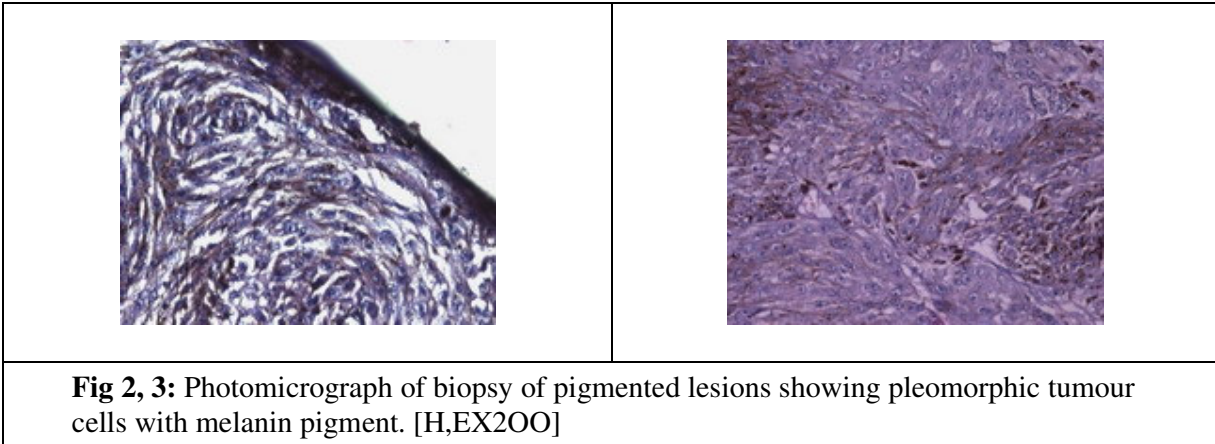
childhood. These skin lesions initially appeared over the face and gradually involved the entire body surface. The skin pigmentation was progressive and more so after exposure to sunlight. She presented with increase in size and ulceration of two lesions over the face since one month. There was no history of consanguinity and the parents and the other siblings were normal.



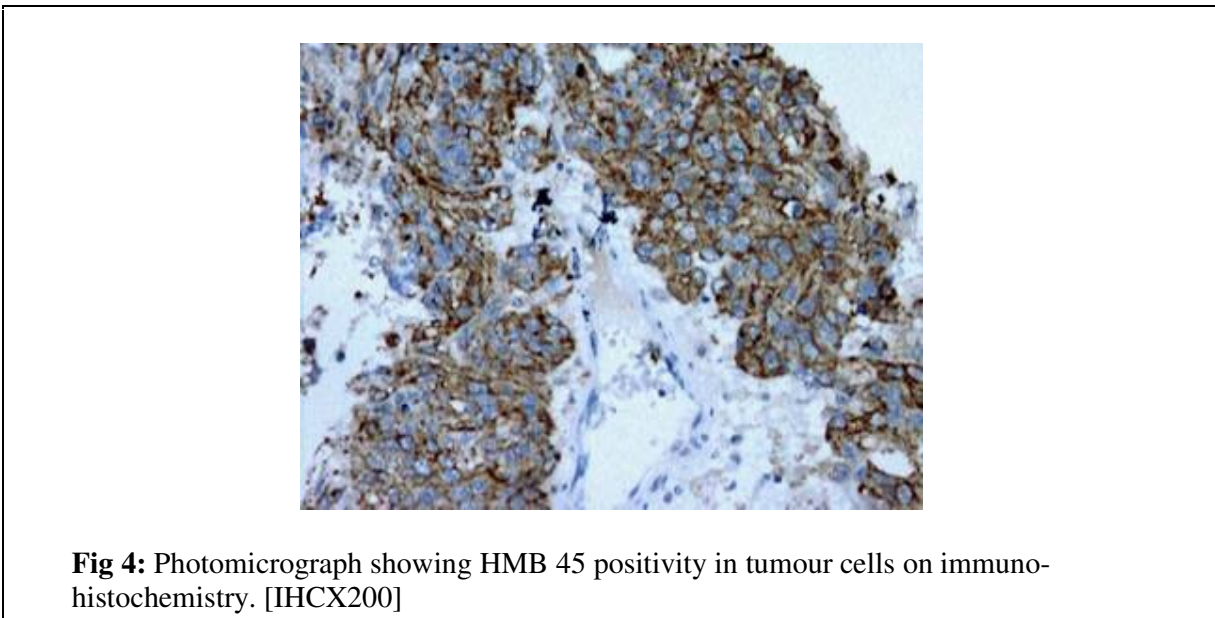
Fig 1: Segmental vitiligo lesions: lesions affect different body sites

Fig.1: Multiple pigmented ulcero-proliferative lesions on the face.

Local examination revealed pigmented ulcero-proliferative lesions over the face varying in size from 1 to 1.5cm with crusting and hemorrhage. Multiple pigmented macules and freckles were noticed over the rest of the body. Lesions were more over the sun exposed areas. There was no significant cervical lymphadenopathy. Systemic examination was essentially normal. Complete haemogram and serum biochemistry was within normal limits. Chest radiography and ultrasonography of the abdomen were normal.



Wedge biopsy of ulcerated skin lesion over the face revealed tumour cells containing melanin pigment. High power view revealed pleomorphic cells with moderate amount of cytoplasm, vesicular nuclei, prominent nucleoli and melanin pigment.



On immuno-histochemistry, the tumour cells were positive for HMB-45 and S-100. The histological features were consistent with those of malignant melanoma.

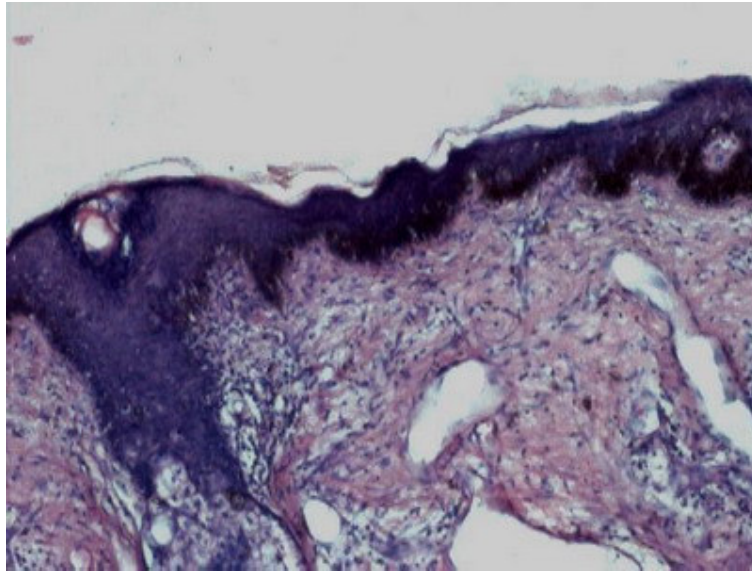


Fig 5: Photomicrograph of biopsy from macules showing features of Xeroderma Pigmentosum. [H,EX200]

Biopsy from hyperpigmented macular lesions over the body showed features consistent with Xeroderma pigmentosum.

The patient was offered conservative treatment with antibiotics and wound care and was referred to plastic surgery where she underwent excision of both lesions.

Discussion

Xeroderma pigmentosum is a rare autosomal recessive disorder. It was first described in 1874 by Hebra and Kaposi. In 1882, Kaposi coined the term Xeroderma pigmentosum. [1,2] It has been reported worldwide in all races with an estimated frequency of 1: 250,000 in US and somehow is more common in Japan.[2,3,4] In these patients excessive solar damage to the skin develops at an early age. The lesions occur chiefly in areas of skin that habitually are exposed to sunlight. Three stages are recognized; the first stage usually starts when the child is 1-2 years old, there occurs slight diffuse erythema which is associated with scaling and small areas of hyper-pigmentation resembling freckles. In the second stage, atrophy of the skin, mottled pigmentation and telangiectasias develops which give the skin an appearance similar to that of a chronic radio-dermatitis. Third stage usually starts in adolescence; various types of malignant tumors of skin appear, often causing death. Malignancies include squamous cell carcinoma, basal cell carcinoma and rarely fibrosarcoma. Only about 3% of patients develop malignant melanoma. The eyes are commonly affected showing conjunctivitis and often keratitis with corneal opacities. [1] Apart from oculo-cutaneous malignancies very rarely it may be associated with neoplasm of other organs like brain, bone marrow, stomach, testis, lungs, pancreas etc.[6]

The basic defect in xeroderma pigmentosum is in the nucleotide excision repair (NER) leading to deficient repair of DNA damaged by UV radiation. There are ten genetic complementation

groups, while one group exhibits defective, post replication repair (XP variant), nine are deficient in excision repair (XP group A-I). Owing to impaired ability to repair, defective or damaged DNA leads to heritable chromosomal mutation and cell death, which possibly cause neoplastic and atrophic clinical abnormalities.[7] Seven XP repair genes, XPA through XPG have been identified. In addition to the defects in repair genes, UVB radiation also has immuno-suppressive effects that may be involved in the pathogenesis of xeroderma pigmentosum.[1,3] The prognosis of this disease is poor with fewer than 40% of patients surviving beyond the age of 20. Individuals with milder forms of disease may however survive beyond middle age. [3]

Although Xeroderma pigmentosum is ultimately fatal, life can be prolonged by paying strict attention to simple preventive measures to minimize sun exposure. The patient should undergo regular checkups for early detection and treatment of any malignancies that may occur to reduce the mortality [3]

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