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XERODERMA PIGMENTOSUM WITH MULTIPLE CUTANEOUS MALIGNANCIES: A CASE REPORT.

Dermatology

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ABSTRACT

Xeroderma pigmentosum is a rare disorder of DNA repair. It is characterized by pigmentary changes, photosensitivity, mucocutaneous malignancies and neurodegeneration. Here we report a case of xeroderma pigmentosum with multiple cutaneous malignancies at the time of presentation.

KEYWORDS

Xeroderma pigmentosum, Basal cell carcinoma, Malignant melanoma

INTRODUCTION:

Xeroderma pigmentosum is a rare disorder of nucleotide excision repair with autosomal recessive inheritance. There is a higher incidence of ultraviolet radiation induced cutaneous and mucosal cancers on sun exposed areas. Most common malignancy associated is basal cell carcinoma followed by squamous cell carcinoma and melanoma[1]. Other important features include severe photosensitivity and neuronal degeneration. Ocular manifestations can be in the form of dryness or development of pterygia. The disease is having poor prognosis with median age of death less than 40 in most cases. We report a case of xeroderma pigmentosum in a young female with extensive malignancies, which were confirmed as basal cell carcinomas and malignant melanoma on histopathology.

Case Report:

A 21 year old female patient born out of non-consanguineous marriage presented with multiple tumours over the face and pigmentary changes all over the body since childhood. Similar history of pigmentary changes was present to her elder sister. On examination multiple lentigines, freckles and hypopigmented macules were present all over the body, along with multiple cauliflower like sessile and pedunculated tumors over the face with pus discharge (Figure 1, 2 & 3). Multiple hyperpigmened macules , papules and plaques were present over the neck and extremities. A clinical diagnosis of xeroderma pigmentosum was made. Dermoscopic examination of one of the hyperpigmented plaques showed asymmetry, blue- gray veil and color variety which were suggestive of malignant melanoma. (Figure 4&5). Dermoscopy of tumours over the face showed large clods and arborizing vessels pointing towards the diagnosis of basal cell carcinoma.(Figure 6). On investigating, she was found to have severe anemia and blood transfusions were given following which she was subjected to excision biopsy and skin grafting. (Figure 7) Histopathological examination was carried out in the biopsy specimen which showed basaloid cells with peripheral palisading consistent with basal cell carcinoma. (Figure 8).

Figure 1- Multiple lentigines, freckles, hypopigmented macules, hyperpigmented papules and plaques over the anterior aspect of trunk and extremities.

Figure 2- posterior aspect of trunk showing multiple hypopigmented and hyperpigmented macules, papules and plaques.

Figure 3: Multiple cauliflower-like sessile and pedunculated tumours. Few lesions are secondarily infected and showing pus discharge.

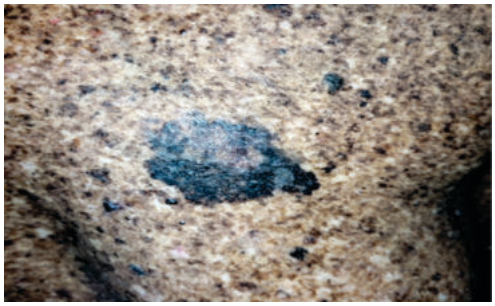

Figure 4: An irregular hyperpigmented plaque present over the anterior aspect of left side of trunk

Figure 5: Dermoscopy showing asymmetry, blue grey veil & color variety- Suggestive of melanoma

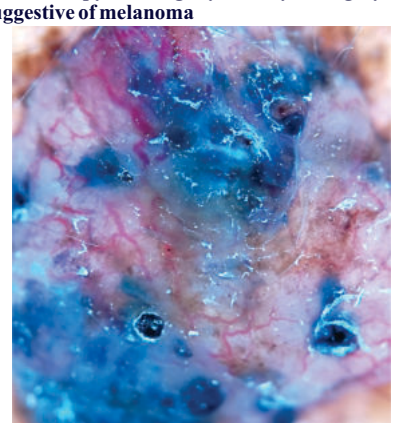

Figure 6: Dermoscopy showing large clods and arborizing vessels- suggestive of basal cell carcinoma

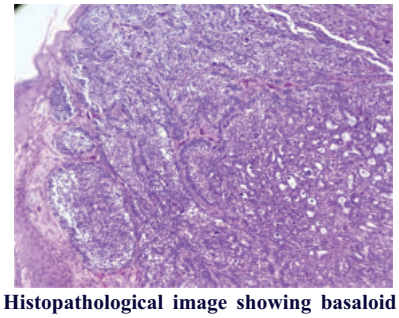

Figure 7: Histopathological image showing basaloid cells with peripheral palisading- Basal cell carcinoma.

Figure 8: Post operative clinical image after excision of tumours.

DISCUSSION:

Most of the patients with Xeroderma pigmentosum are usually normal at birth. Because of extreme photosensitivity, there will be exaggerated sunburning even with minimal exposure. Cutaneous manifestations include abnormal pigmentation, lentigines, freckles, premature aging, atrophic changes, poikiloderma or presence of actinic keratoses[2].

The disease is categorized into eight complementation groups XP-A to XP-G and XP-V. Of these, patients of XP-A to XP-G disease develop cutaneous malignancies before 20 years whereas in XP-V variant, malignancies develop in second or third decade. Incidence of other malignancies such as medulloblastoma, glioblastoma, astrocytoma and schwannoma is also increased in patients of xeroderma pigmentosum[3]. Our patient had multiple basal cell tumours over the face and malignant melanoma over the anterior aspect of trunk and all of them were excised.

Progressive neurodegeneration presenting with cerebellar signs, ataxia and areflexia along with increased incidence of psychological morbidity are also noted in this patients. Our patient did not have any neurological or psychological manifestations. Patients without any neurological abnormalities and those who are being protected from UV radiation are having comparatively better prognosis than the patients with neurological disease. Our patient was treated with excision biopsy and skin grafting. She was advised to have adequate sun protection and is under regular follow up.

CONCLUSION

There is no absolute cure for xeroderma pigmentosusm and the life expectancy is very low. We report this case because of its presentation with extensive cutaneous malignancies. The tumors were excised following which skin grafting and flap reconstruction was done giving good functional and aesthetic results.

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