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XERODERMA PIGMENTOSUM WITH SQUAMOUS CELL CARCINOMA: A CASE REPORT

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ABSTRACT
Xeroderma pigmentosum (XP) with squamous cell carcinoma of skin has been infrequently reported. XP is a rare autosomal recessive disease characterized by a spectrum of symptom complex due to defective DNA repair. A 12-year-old boy having xeroderma pigmentosum presented with extensive ulceration in the face. On histopathological examination, the ulceration was found to be squamous cell carcinoma. The details of this case are presented.

INTRODUCTION

Xeroderma pigmentosum (XP) is heterogeneous group of genetic disease, result from faulty DNA repair. It is a rare autosomal recessive disease characterized by photosensitivity, pigmentary changes, premature skin aging, neoplasia and abnormal DNA repair mechanism [1]. Basal cell carcinoma is found to be associated with xeroderma pigmentosum in majority of the reported cases in Indian literature [2, 3].

We report a case of squamous cell carcinoma of the face in a young boy co-existing with xeroderma pigmentosum.

CASE REPORT

A 12 year old boy presented with history of pigmentation of skin over face,neck and trunk. Freckles present all over the body and photophobia since 6 months of age. Multiple ulceration over the left and right cheeks was present for 9 months. The pigmentation was progressive and more so after exposure to sunlight and over exposed areas. There was no history of difficulty in hearing or seizures. He had a good appetite with bowel movements normal. There was no family history of consanguinity.No history of similar complaints in the family.And he was immunized with no significant perinatal history. Development of milestones was normal.He was also suffering from redness of both eyes and photophobia.
Local examination revealed extensive ulceroproliferative lesion which is 5×6 cm involving left side of the nose, extending to the left cheek with crusting and haemorrhage and the ulcer present over the right is 2×2 cm in diameter [Fig 1].

There was no significant cervical lymphadenopathy. Systemic examination including neurological functions was normal. Haemogram and serum biochemistry was within normal limits. Chest radiography and ultrasonography of abdomen were normal.Biopsy of the skin lesion revealed characteristic features of squamous cell carcinoma such as pseudo epitheliomatous hyperplasia, atypical squamous cells, with keratin pearls and inflammatory infiltrate [Fig 2,3&4].

Figure 1. Clinical photograph showing multiple ulceroproliferative lesions along with numerous freckles and perforation of the right ala nasi.

Figure 2. Histopathology showing pseudo epitheliomatous hyperplasia and atypical squamous cells in epidermis which is extending into the dermis.

Figure 3. Arrow mark showing individual cell keratinization in the epidermis

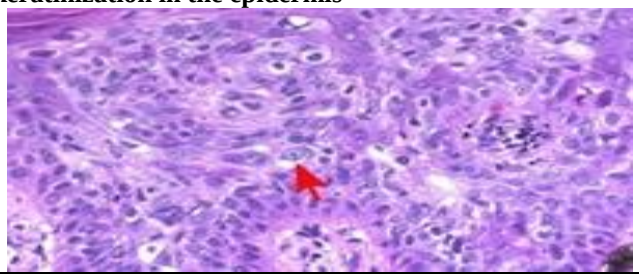
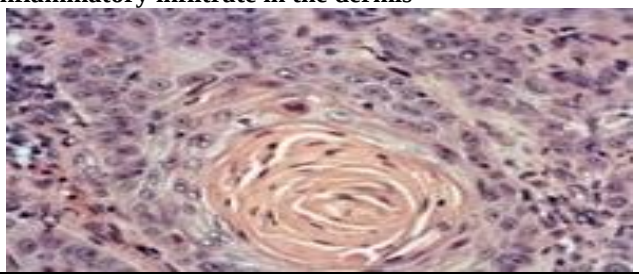


Figure 4. Microscopic view showing horn cysts and inflammatory infiltrate in the dermis



DISCUSSION AND CONCLUSION

Photosensitivity, pigmentary changes, premature skin aging, and malignant tumor development characterize XP. The manifestations of XP mainly due to cellular hypersensitivity to ultraviolet (UV) radiation resulting from a defect in DNA repair. The basic defect in XP is in nucleotide excision repair (NER), leading to deficient repair of DNA damaged by UV radiation [4]. Two types of NER exist: global genome (GG-NER) and transcription coupled (TC-NER). Seven complementation groups of XPA-XPG correspond to defects in the corresponding gene products of *XPA-XPG* genes and they differ with respect to disease severity (e.g., XPG is severe, whereas XPF is mild) and clinical features. Cockayne syndrome can rarely occur with XPB, XPD, and XPG.

Clinically, XP passes through 3 stages. The skin is healthy at birth. Typically, the first stage makes its appearance after the age of 6 months. Diffuse erythema, scaling, and freckle like areas of increased pigmentation characterize this stage. The second stage is characterized by poikiloderma, giving rise to an appearance similar to that of chronic radiodermatitis. The third stage is heralded by the appearance of numerous malignancies, including basal cell carcinoma, squamous cell carcinomas, fibrosarcoma and malignant melanoma. De Sanctis-Cacchione syndrome refers to the combination of XP and neurologic abnormalities (including cerebellar ataxia and mental retardation), dwarfism and hypogonadism [4, 5].

Children with this disease develop multiple cutaneous neoplasms at a young age. Two important causes of mortality are metastatic malignant melanoma and squamous cell carcinoma. Patients younger than 20 years have a 1000 - fold increase in the incidence of nonmelanoma skin cancer and melanoma [5]. The mean patient age of skin cancer is 8 years in patients with XP compared to 60 years in the healthy population. Actinic damage occurs between 1 and 2 years of age. An equal incidence has been reported in males and females. The disease is usually detected at age 1 or 2 years [5].

Histopathologically, hyperkeratosis and increased melanin pigment (corresponds to the clinical freckling) in the basal cell layer. Some rete ridges may be elongated, whereas other rete ridges may be atrophic. These findings may be accompanied by a chronic inflammatory infiltrate in the upper dermis. In the second stage, atrophy, hyperkeratosis, hyperpigmentation and telangiectasia may be prominent. In addition, the epidermis may exhibit architectural disorder and atypia, and the dermis may be elastotic. The histologic appearances of the various tumors that complicate XP are seen in the third stage of XP [6]. Differential diagnosis should include Bloom syndrome, ephelides (freckles), Hartnup disease, hydroa vacciniforme, Leopard syndrome, Progeria (Werner syndrome), Rothmund Thomson syndrome, Cockayne Syndrome, basal cell nevus syndrome and Porphyria [6].

The diagnosis of XP can be established with studies performed in specialized laboratories. These studies include cellular hypersensitivity to UV radiation and chromosomal breakage studies, complementation studies, and gene sequencing to identify the specific gene complementation group [6].

The goal of treatment is to protect the child from sunlight. The use of sunscreens in conjunction with other sun avoidance methods (e.g., protective clothing, hats, and eyewear) can minimize UV-induced damage in patients

with XP. Oral retinoids have been shown to decrease the incidence of skin cancer in patients with XP. A new approach to photo protection is to repair DNA damage after UV exposure by delivery of a DNA repair enzyme into the skin by means of specially engineered liposomes. Complete excision of the malignancies associated with XP should be performed. An ophthalmologic consultation is recommended because of the ocular problems associated with XP. A neurologic consultation is recommended as well because neurologic abnormalities are seen in 20% of patients with XP [6].

As multiple cutaneous neoplasm develop in persons with XP at a younger age, early diagnosis and management would be life saving. Genetic counseling should be offered for families. Follow up care should be geared to educate the patient and the parents about effective sun protection and early recognition of skin cancer.

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