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Case Report

Xeroderma Pigmentosum with multiple squamous cell carcinomas: A case report with literature review

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Abstract

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease, in which the ability of the DNA repair is impaired, symptom presentation is majorly observed over the skin exposed to sunlight and can range from minor hyperpigmentation to ulcerative nodules. Frequently this can be associated with cutaneous malignancies Early detection of these malignancies is necessary because they are fast growing, metastasize early and leads to death. This paper describes a case of xeroderma pigmentosum with multiple cutaneous squamous cell carcinomas in a 12-year old boy.

Key words: Xeroderma pigmentosum, squamous cell carcinoma

Introduction

Xeroderma Pigmentosum (XP) first described by Hebra and Kaposi in 1874. It is a rare autosomal recessive genetic disorder characterized by defective DNA repair which leads to clinical and cellular hypersensitivity to ultraviolet radiation and other carcinogenic agents.[1,2] In 1882, Kaposi coined the term xeroderma pigmentosum for the condition, referring to the characteristic dry, pigmented skin seen in these patients.[3] XP is a genodermatosis characterized by photosensitivity, cutaneous pigmentary changes, premature skin aging and the development of cutaneous and internal malignancies at an early age. These patients exhibit enhanced sensitivity to ionizing radiation.[4] Synchronous occurrence of multiple cutaneous malignancies in a patient of XP is extremely rare.

Case report

A 12-year-old boy presented with multiple ulceroproliferative nodules in the scalp. History reveals the patient was apparently well before the age of 3 years, when his mother started noticing mild hyperpigmentation over his face and neck. He was taken for regular check-up and advised to limit sun exposure and use sun protective measures. Over the next years, he was exposed to sunlight and protective measures were not taken. The condition became aggressive and the macules spread aggressively over the face, scalp and throughout the body particularly in the sun exposed areas.

There was no history of other family members having XP. On examination, he had severely damaged facial skin (Figs 1), 'salt and pepper' pigmentory changes all over his body. The scalp nodules were approximately 8×4 cm and 4x3 cm sized and were ulceroprolifeartive. The crusted lesions were more over the sun-exposed areas. The findings were consistent with classic XP.

The patient underwent punch biopsy followed by histopathological examination. Cut surface of the mass is gray white with areas of hemorrhage and necrosis. Microscopic examination shows sheets and cords of squamous epithelial cells having intercellular bridges (Figs: 2). Keratin pearls are present as well. Histopatholoigically it is diagnosed as Invasive squamous cell carcinoma, well differentiated. The patient was reffered to National Institute of Burn and Plastic surgery for wide excision of the scalp lesions followed by skin grafting.

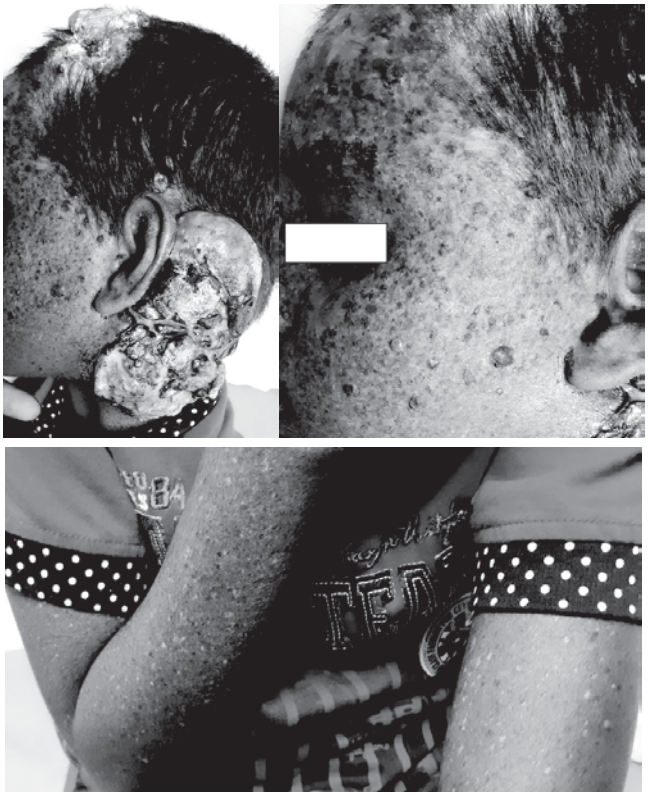

Fig 1: Ulceroproliferative lesion overs scalp and pigmentory changes all over his body which is more marked in face.

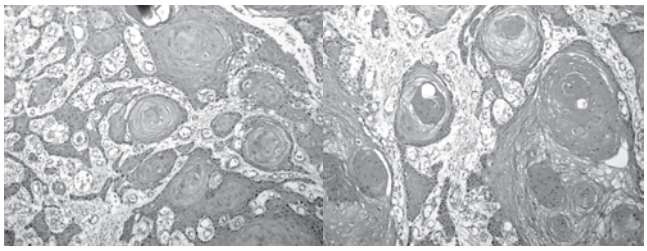

Fig 2: Histopathology shows a well differentiated squamous cell carcinoma with keratin pearls

Discusssion

XP affects males and females equally and is frequently symptomatic since early childhood.[5] The incidence of XP in the United States, Western Europe, and Japan is 1 in 250 000, 1 in 500 000 and 1 in 22,000, respectively,[6] with higher incidence in communities where consanguinity is common.[7] The incidence in the Indian population is insignificant.[8]

The basic defect in XP is in nucleotide excision repair (NER), leading to deficient repair of DNA damaged by UV radiation. NER involves removal and the replacement of damaged DNA with new DNA. Two types of NER exist: global genome NER (GG-NER) and transcription coupled NER (TC-NER). In addition to defects in the NER genes the immunosuppressive effects of UV-B radiation may also be involved in the pathogenesis of XP. XP patients below 20 years of age have a >1000-fold increased risk of developing skin cancer.[8]

The two most common types of cancer found in XP patients are Basal cell carcinoma and Squamous cell carcinoma, mainly occurring on the face, head, and neck. Melanomas occur in one-fourth of cases, and one-third of these occur in the head and neck.[9] Eyelid and conjunctival cancers have been reported, [10] conjunctival involvements includes squamous cell carcinoma developing from the interpalpebral zone. Limbal tumor includes pterygia, squamous cell carcinomas, and malignant melanoma. The iris can rarely be affected by melanoma. Orbital tumors include basal cell carcinomas, squamous cell carcinomas, and melanomas.[11] The oral manifestations are mainly related to the occurrence of malignant tumors in the lips, tongue and buccal mucosa. [12]

Xeroderma pigmentosum (XP) is characterized by an extreme sensitivity to ultraviolet (UV) rays from sunlight. Infants and children are reported to be most commonly affected by XP as they have immature immune system. The sunburn commonly causes redness and blistering but some children also present with normal tanning of exposed area. Usually by age 2, almost all children with xeroderma pigmentosum develop freckling of the skin in sun-exposed areas (such as the face, arms, and lips. In affected individuals, exposure to sunlight often causes dry skin (xeroderma) and changes in skin coloring (pigmentation). This combination of features gives the condition its name, Xeroderma Pigmentosum.[13]

The recommendation for preventing these neoplasms in patient with XP is the avoidance of UVR exposure from sunlight through the application of high-factor sunscreen, UVR-protective clothing, hats, gloves and sunglasses, and UVR-blocking window films at home and in cars. In Bangladesh many of our patients have little access to adequate information about the disease

and importance of UV protection, on top of late diagnosis and difficulty to find medical assistance. High cost of UV protective sunscreen and barriers like UV protective hats and clothes also present a challenge.[14]

Conclusion

XP patients need periodic skin examination by dermatologist and ocular exam by an ophthalmologist. Besides dermatological, ophthalmological management, XP patients require constant follow-up in order to control the occurrence of new lesions. Although, early detection and treatment of cutaneous malignancies will reduce morbidity and mortality, genetic counseling remains the most important measure for preventing XP. There are very few cases of xeroderma pigmentosum reported in literature from Bangladesh. In Bangladesh there is no data regarding XP incidence and prevalence. Reporting every case might help us to know the incidence and prevalence of XP in Bangladesh which is yet unknown.

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