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Xeroderma Pigmentosum with Squamous Cell Carcinoma of Eye

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Abstract: *XerodermaPigmentosum(XP) is a rare autosomal recessive disorder, characterized by photosensitivity, pigmentary changes, premature skin ageing and marked increase in risk of developing malignant neoplasms of the skin and eyes. Here we present case of a sibling, 8 years old boy and 6 year old girl (Figure. 1). Girl had developed squamous cell carcinoma of left eye.*
Keywords*: XERODERMA PIGMENTOSUM, PHOTOSENSTIVITY, SQUAMOUS CELL CARCINOMA*

I. Introduction

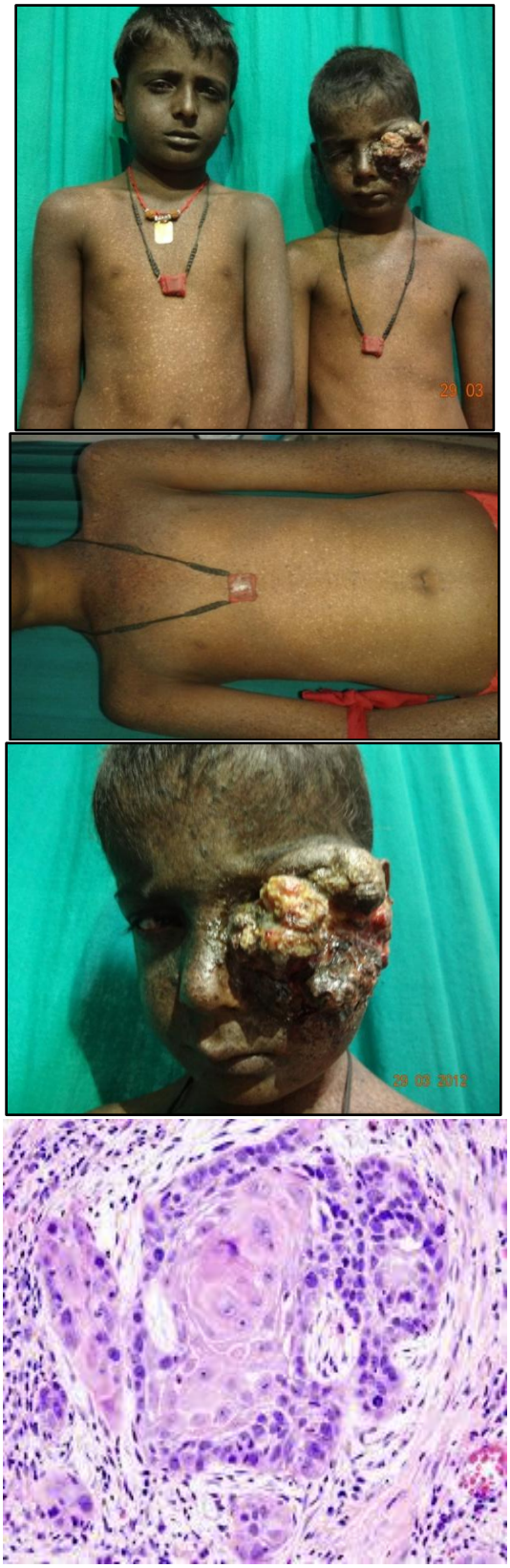
XP is a hereditary autosomal recessive disorder, characterized by mucocutaneous and ocular hypersensitivity to ultraviolet radiation. Herbaand Kaposi first described XP in 1974 [1,2]. Kramer et al found an equal sex predilection and significant parental consanguinity confirming an autosomal recessive inheritance patner [3]. The incidence of XP in United states and Europe are 1:250000 and in Japan and other countries at a higher frequency 1:40000. Its incidence is not that significant in context to other part of the world [4]. The basic defect underlying the clinical manifestation is a nucleotide excision repair (NER) defect leading to a defective repair of DNA damaged by ultra violet (UV) radiation [3,5]. XP is an autosomal recessive disorder with 100% penetrance. In general the signs and symptoms of Xeroderma Pigmentosum starts from the age of 1-2 years. The disease begins with photosensitivity and burning sensation after normal sun exposure in 60% cases. Cutaneous manifestation include dryness of skin, pigmentation, freckling and telangiectasis. Occular abnormalities include photophobia, ectropion, conjunctival infection, keratitis with incidence of tumors like SCC, melanoma and epithelioma. There is 1000 fold increased risk of skin malignancy on sun exposed sites. A onefifth of patients (20-30%) have associated abnormalities such as gait disturbance, aflexia, difficult swallowing, deafness, growth delay, and low intelligence[4]. Historically, the disease was classified as classical XP with only skin abnormalities and the De-Sanctis- Cacchione syndrome with skin abnormalities and extreme neurological degeneration wasevident[1]. XerodermaPigmentosum is variably also known as Kaposi Disease, Xeroderma Pigmentosum Variant type, XP-V and XP.

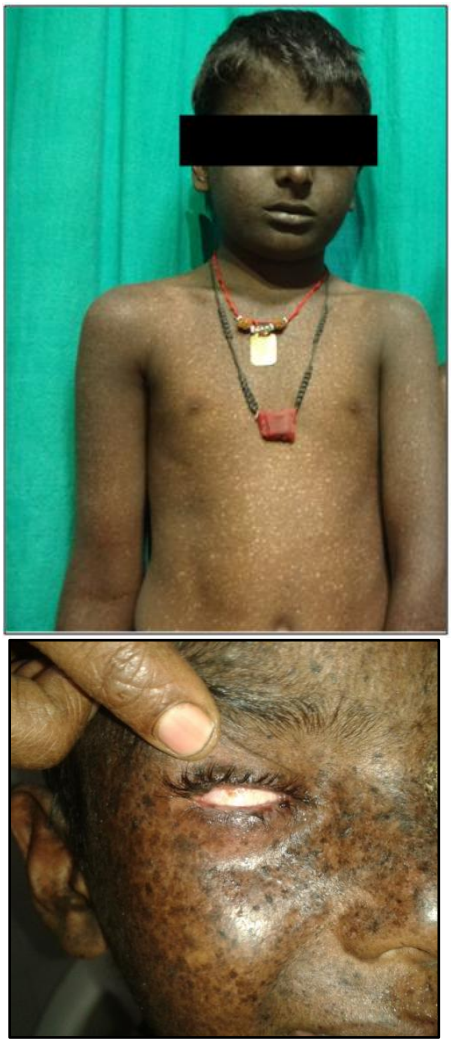
II. Case Report

Case 1: We present the case of an 6 year-old girl who presented to the out-patient department of UPRIMS & Research Saifai, Etawah (U.P), after her mother concern regarding progressive ocular lesions. At the age of 8 months she presented with numerous brownish pigmentation of the face, which was initially less in number, although were confined to sun-exposed areas. Over time these lesions had enlarged and become progressively more numerous and raised and spread to unexposed areas like abdomen, groin and thighs (Figure.2). Xerosis of face and hands were observed. Scarring at multiple places. The patients's mother gave the history that girl have severe burning sensation on exposure to sunlight. In the last year her mother had noted gradually enlarging corneal lesions on left side of face. The growth was approximate 8x8 cm in size covered entire left eye, round to oval in shape, indurated and was bleeding at the time of examination (Figure.3). In the last year her mother had noted gradually enlarging corneal scarring in right eye also. On asking relevant history to her mother it was found that following normal vaginal delivery at home with no pre-natal care, the patient had low IQ and delayed milestones. The mother have history of consanguinity in the patient's recent lineage. Incisional biopsy was taken from the face for the histopathological diagnosis. Histopathological diagnosis was moderately differentiated squamous cell carcinoma (Figure.4). Immunohistochemistry staining for p53 was also carried out and was found to be positive. The patient was advised for the surgical resection of the tumour. The patient was also advised to apply sunscreen on exposed skin and to avoid sun exposure.

Case 2: Her 8 year old brother was also diagnosed with xerodermapigmentosum on clinical examination. He presented with brownish-black pigmentation on sun exposed parts of the body like face, neck, hands and legs at the age of 2 years (Figure.5). Xerosis of skin is also evident. He also complained of burning sensation on exposure to sunlight. Corneal scarring of right eye also present (Figure.6). On asking relevant history to his

mother, he had no developmental delay and his mental IQ was average. The patient was advised to apply sunscreen and avoid sun exposure. Patient was recalled for follow up every month. Unfortunately, both the patient didn't turn up to our institution. Thus, we don't have follow up record for the patient.





III. Discussion

Xerodermapigmentosum is autosomal recessive genetic disease caused by defects in the normal repair of DNA of various cutaneous and ocular cell types damaged by exposure to sunlight [6,7]. From an early age patients are sensitive to even minimal sun exposure developing erythema, vesicles and oedema. By the age of two years solar lentigos, xerosis and pigmentation occur. Later in childhood dysplastic and neoplastic lesions occur with the development of actinic keratosis, keratocanthoma, basal cell carcinoma, squamous cell carcinoma and malignant melanoma[8]. In one study the median age for development of malignant melanoma was 8 years of age [9]. Ocular complications are nearly as common as skin lesions with keratitis progressing to corneal opacification, loss of eyelashes, ectropion, entropion and benign and malignant lesions of the cornea and eyelids. Neurological complications occur in approximately 30% of cases and can be severe [8,10]. XP is more commonly seen in populations where marriage of close blood relatives is common [11]. XerodermaPigmentosum has been reported worldwide in all races with an overall prevalence of 1–4% per million [12]. Treatment of the disorder includes avoidance of ultra violet radiation, topical application of 5-fluorouracil to treat actinic keratosis and experimental treatments with topical DNA repair enzymes and oral retinoids are showing promise for the future [13,14] and regular evaluation by an ophthalmologist, dermatologist, and neurologist. Genetic counseling is an important aspect as an increased incidence of consanguineous marriages has been reported with this disorder [11]. A case of xeroderma pigmentosum should be give utmost importance by the panel of doctors, to improve the life expectancy of the affected individual. There are very few cases of xerodermapigmentosum reported in literature from India. Reporting every case might help us to know the incidence and prevalence of XP in India which is yet unknown.

Abbreviations :
DNA: Deoxyribonucleic Acid; UV: ultra-violet; XP: XerodermaPigmentosum.

Figures legends:

Figure.1 Image of sibling.
Figure. 2Lesions spread to photoprotected sites in girl.
Figure. 3Indurated mass over left side of face.
Figure. 4Well differentiated squamous cell carcinoma.
Figure. 5Lesions spread to photoprotected sites in boy.
Figure. 5 Right corneal scarring in boy.

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