XERODERMA PIGMENTOSUM: A CASE REPORT OF TWO INDIAN SIBLINGS

INTRODUCTION:
Xeroderma pigmentosum is a rare autosomal recessive disorder of DNA repair characterized by clinical and cellular hypersensitivity to ultraviolet radiation and carcinogenic agents which leads to progressive pigmentary abnormalities and increased incidence of UVR-induced skin and mucous membrane cancers at sun-exposed sites. Ocular involvement has been reported in 40%-80% of cases, 10 % of them present with ocular malignancies.

CASE DETAILS:
Two male siblings of 7 and 6 years of age presented with hyperpigmented macules and papules and ocular manifestations. They were born to non-consanguineous marriage. Elder child was absolutely normal till 1 years of age, thereafter, he developed hyperpigmented spots and bilaterally distributed freckles, first over face and neck then over chest and both extremities. History of photophobia and watering of eyes was present on exposure to sunlight. At 1 and half years of age, history of ocular malignancy is there for which evisceration was done which leads to loss of vision in left eye.

DISCUSSION:
Xeroderma pigmentosum was first described by Hebra and Moriz Kaposi in 1874. The incidence of XP is 1:250,000 births in the USA, in Japan 1:20,000 and other countries at a higher frequency 1:40000. The term xeroderma pigmentosum was introduced in 1882. The prevalence of Xeroderma pigmentosum is higher in North Africa and the Middle East, more in communities in which consanguinity is common. It affects males and females equally. XP is an autosomal recessive disorder and results from mutations in any one of eight genes (XP-A to XP-G complement groups) and post replication repair defect (XP-Variant). We present a case of 2 Indian male siblings of 7 and 6 years of age with Xeroderma pigmentosum.

KEYWORDS
Xeroderma pigmentosum, photosensitivity, DNA-repair defect disorders.
approach which should include regular skin, eye and dental reviews, early management of any malignancy. Patients with XP must avoid exposure to sunlight and other sources of UV light and must wear protective clothing, hats, gloves, UV-absorbing sun glasses. Use of high-factor sun screen, topical application of 5-fluorouracil or imiquimod for premalignant lesions and surgical excision for malignant neoplasm of the skin, tongue, eyelids, conjunctiva, and cornea. Eye care include use of sunglasses, methyl cellulose or quinodine containing eye drops and bland ointment at night. Vitamin D deficiency is common and supplements should be prescribed. Smoking is prohibited. Retinoids may have a role in the prevention of skin cancer.

Genetic counselling is also an important component in management of patients with XP, especially in a family that has an affected child and is considering having more children.

Psychosocial issues need to be addressed which include social isolation from peers at school and at home, limited career prospects etc. In vitro and ex vivo experiments have established that correction of the underlying genetic defect in different forms of XP is possible. Animal studies using viral vectors have also established that gene therapy approaches for patients with this disease may become possible.

Xeroderma Pigmentosum has been reported worldwide in all races with an overall prevalence of 1–4% per million. In India, there are very few cases of xeroderma pigmentosum has been reported in literature till date. Reporting every case might help us to know the exact incidence and prevalence of XP in India which is yet unknown.

CONCLUSION:
Patients with XP are more prone to the development of malignancies at an early age which can progress rapidly with drastic consequences thus emphasizing the early diagnosis and treatment. Thus, a case of xeroderma pigmentosum should be give utmost importance by the panel of doctors, to improve the life expectancy of the affected individual.

REFERENCES