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XERODERMA PIGMENTOSUM: A CASE REPORT OF TWO INDIAN SIBLINGS

Dermatology

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ABSTRACT

Xeroderma pigmentosum is a rare type of genodermatosis, inherited as an autosomal recessive disorder, characterized by photosensitivity, freckles, premature skin ageing, telangiectasis, pigmentary changes and skin cancers along with ocular, oral and neurological complications. These manifestations are due to mutation in seven nucleotide excision repair gene (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.

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 mucus membrane cancers at sun-exposed sites. Ocular involvement has been reported in 40%-80% of cases, 10 % of them present with ocular malignancies.[1]

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. Right eye examination shows congested conjunctiva and hazy cornea (Fig. 1). His younger sibling also developed brownish-black pigmentation and freckles over sun-exposed sites at 2 year of age. There is history of watering, redness and purulent discharge in both eyes after sun exposure (Fig. 2). Systemic examination of central nervous system, respiratory system and cardiovascular system was normal in both siblings. They were advised photo protection in the form of sun screens and appropriate clothing and referred to ophthalmologist for ocular complications.

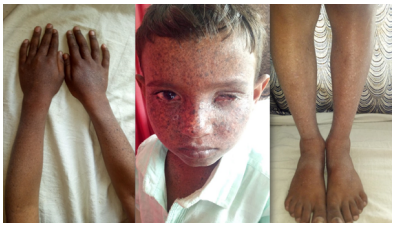

Figure 1: Sibling 1 with Xeroderma pigmentosum with pigmentary changes and loss of left eye due to ocular malignancy.

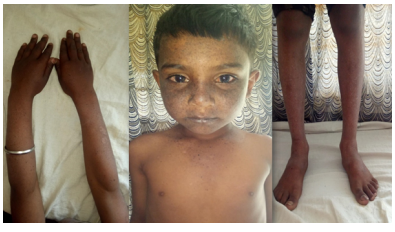

Figure 2: Sibling 2 with Xeroderma pigmentosum with pigmentary changes and ocular manifestations.

DISCUSSION:

Xeroderma pigmentosum was first described by Hebra and Moriz Kaposi in 1874[2]. 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.[3] The term xeroderma pigmentosum was introduced in 1882.[3] 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 (*XPA-XPG*), encoding for cyclobutane pyrimidine dimers (CPDs) and pyrimidine (6.4) pyrimidine photoproducts (6.4PPs) involved in the recognition and repair of UVR.induced photoproducts by the process of nucleotide excision repair (NER).[4]

XP is characterised by dermatological manifestations like exaggerated sunburn, persistent erythema, marked freckle-like pigmentation of the face before 2 years of age, dry pigmented skin, atrophy, keratosis, telangiectasis, and neoplasm. Patients with multiple primary lesions, with XP can develop skin cancer by 8 years of age.[5] These children may present with ocular changes before skin lesions which include photophobia, keratitis, atrophy of the skin of the lids, cataract, and eye tumors. 25-30% of XP individuals develop neurological manifestations in infancy or second decade. The neurological abnormalities include microcephaly, progressive intellectual impairment diminished deep tendon reflexes, sensory neural hearing loss, spasticity, ataxia, seizures, pyramidal syndrome, and peripheral neuropathy. Neurological abnormalities are due to loss of neurons, particularly in the cerebrum and cerebellum, primary axonal degeneration in peripheral nerves, and secondary demyelization. Commonest causes of death include skin cancer (BCC, SCC etc.) neurological degeneration, or internal cancer, median age of death is reported as 32 years.[3]

Ocular malignancies can occur in early age which needs early intervention to decrease disease related mortality. Lower eyelid neoplasm occurs more common than upper eyelid neoplasms. Either ectropion or entropion following surgical treatment of the eyelid neoplasms can cause exposure keratitis, pterygium, corneal opacities which can ultimately lead to loss of vision.

Clinical variants include Xeroderma pigmentosum variant (XP.V-20%), Xeroderma pigmentosum/Cockayne syndrome complex (XP/CS), Xeroderma pigmentosum/trichothiodystrophy (XP/TTD) syndrome. It has to be differentially diagnosed from trichothi odystrophy, Cockayne syndrome, cerebro, oculo, facio, skeletal syndrome, UV.sensitive syndrome, erythropoietic protoporphyria and Rothmund–Thomson syndrome.

Definitive treatment of XP is not possible, since DNA injury is cumulative and irreparable. Management requires a multidisciplinary

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 sunscreen, 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.[5] Vitamin D deficiency is common and supplements should be prescribed. Smoking is prohibited. Retinoids may have a role in the prevention of skin cancer.[6,7]

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.[8,9]

Xeroderma Pigmentosum has been reported worldwide in all races with an overall prevalence of 1–4% per million.[10] 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.

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