Xeroderma Pigmentosa; Review and Case Report

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ABSTRACT

Xeroderma Pigmentosa is caused by an autosomal recessive allele. It is characterized by dry, pigmented skin, spidery blood vessels in the skin, skin cancers, and sometimes other abnormalities of both the eyes and brain. A harsh reaction to sunlight, such as severe sunburn and blistering at only a slight exposure, is a notable symptom and should be distinguished within the first year or two of life. The majority of the people who are born with these disorders die by early adulthood due to malignant cancers. This article reports a case of 6-year-old child suffering from XP with dental implications.

Key words: Recessive, Basal cell carcinoma, Skin Cancer, Spidery

Xeroderma Pigmentosa is caused by an autosomal recessive allele. It is characterized by dry, pigmented skin, spidery blood vessels in the skin, and sometimes other abnormalities of both the eyes and brain.

Clinical Features

Characteristics of Xeroderma Pigmentosa are skin atrophy (the thinning of skin), telangiectasia (spidery blood vessels in the skin), and skin cancers. Basal cell carcinoma, squamous cell carcinoma, and malignant melanoma are examples of the skin cancers. While basal cell cancers do not spread easily and are typically easy to treat, malignant melanomas spread quickly to other organs, and squamous cell cancers are much more difficult to treat.(1-2)

Diagnostic Methods

Xeroderma Pigmentosa can typically be detected in the first year of the patient’s life. Most diagnoses are made visually. Molecular genetic testing exists only on a research basis. A family history of XP may be relevant, but since the parents are heterozygous and show no sign of the disorder, the history would be very hard to detect.(1-2)

Etiology

Cleaver suggested that early increase in sunlight-induced cancers was a direct consequence of an increase in mutated cells in the skin of XPs. He showed that patients with xeroderma pigmentosum (XP), who have a propensity for developing light-induced cancers early in life, possessed cells that were defective in the excision repair of UV-induced pyrimidine dimers from their DNA. This defect was correlated with hyper-mutability, when XP cells were exposed to ultra violet radiations (UV).

An alternative hypothesis was proposed which argued that the crucial effect of sunlight which led to the early appearance of skin cancers was not the excessive induction of mutations but the exacerbation by UV of a defect in immune surveillance which resulted in existing transformed cells being able to grow and express their malignant pheno-type.(3-5)

Case Report

A 6-year-old male child reported in the Department of Pediatric Dentistry, SDM college of Dental sciences, Dharwad. He reported with the chief complain of decayed teeth since 4 months. The patient was a known case of Xeroderma Pigmentosa since 4 years. The personal history suggested that this child had been adopted from an orphanage when he was 6 month old. Therefore the family history suggested that this child was extremely sensitive to sun and distinct freckling was seen since the child was.
two years old (Fig. 1). Skin atrophy, telangiectasia, and ocular and neurological problems had developed when the child was five years old.

The Intraoral examination revealed deep occlusal caries with 64,65, caries with 63,83,84,85, pit caries with 55, deep proximal caries with74, chronic irreversible pulpitis with 75 and dark pigmentation with respect to the attached gingiva (Fig. 2, Fig. 3).

The clinical findings suggested early childhood caries. The treatment planning was considered to be oral hygiene instructions with diet counselling and oral prophylaxis followed by topical fluoride application. Restorations with 55,63, 83,84,85. The pulp therapy was done with 64,65,74,75 followed by stainless steel crowns (Fig. 4, Fig. 5).

Removal partial denture (functional space maintainer) was given in the maxillary arch (Fig. 6) and the occlusion was reestablished (Fig. 7).

The Patient was asked to maintain regular recalls and checkup. The appointments were kept short and early in the morning for the convenience of the patient.

**Discussion**

Xeroderma Pigmentosa is characterized by dry, pigmented skin, spidery blood vessels in the skin, skin cancers. Approximately 80% of XP patients have ocular complications. These complications may include severe keratitis, which could be followed by corneal opacification and vascularization. XP patients may lose their eyelashes, and in severe cases, they may also lose their entire eyelid. Neurological symptoms such as microcephaly, progressive sensorineural hearing loss, and cognitive impairment can be found in around 30% of XP patients. Berkel and Kiran divided a group XP patients according to the extent of their cutaneous disease there appeared to be an inverse relation between disease severity and the development of contact allergy. Thus light-exposed XP patients are susceptible to higher risk of skin cancer because their defect in DNA repair results in an increased frequency of initiated (mutated) skin cells which are able to grow into tumorous colonies early in life, probably because of failure of the immune system to restrict their growth. This failure may be two-fold: a constitutive defect (probably in NK cell
function) exacerbated by a UV-dependent impairment (probably of cell-mediated immunity and possibly also of residual natural killer cell function. With the possibility of ophthalmic malignancies, the need to avoid acute exposures of UV radiation to the eyes by use of protective eye gear, UV block glasses or goggles early in the course of disease can not be neglected and should form part of the plan in management of the disease.(9-11)

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