CASE REPORT

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.

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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.

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 could not be retrieved. The child was extremely sensitive to sun and distinct freckling was seen since the child was

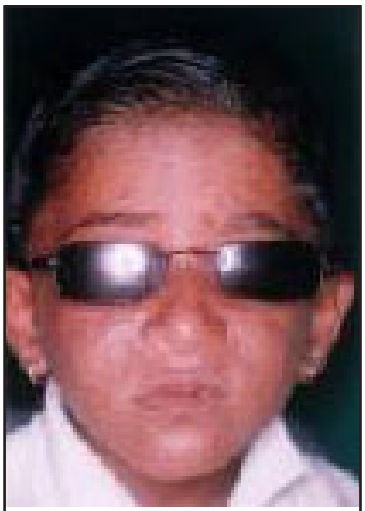

Fig. 1: Photograph of face showing frecking of skin

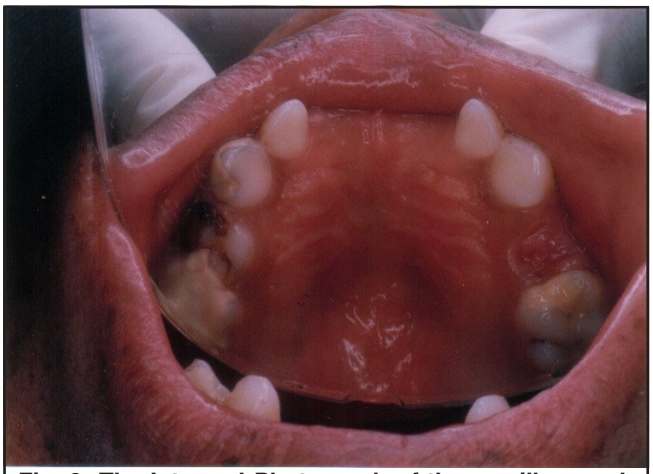

Fig. 2: The Intraoral Photograph of the maxillary arch showing the carious teeth (55, 63, 64 65). (Mirror View)

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).

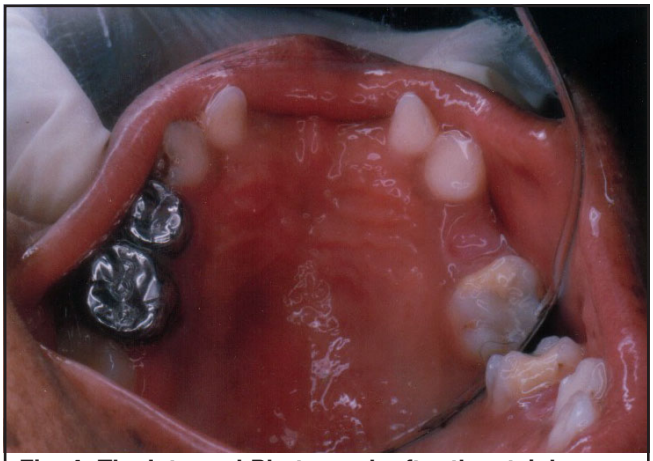

Fig. 4: The Intraoral Photograph after the stainless steel crown placement with 64,65 and restoration with 55&63. (Mirror View)

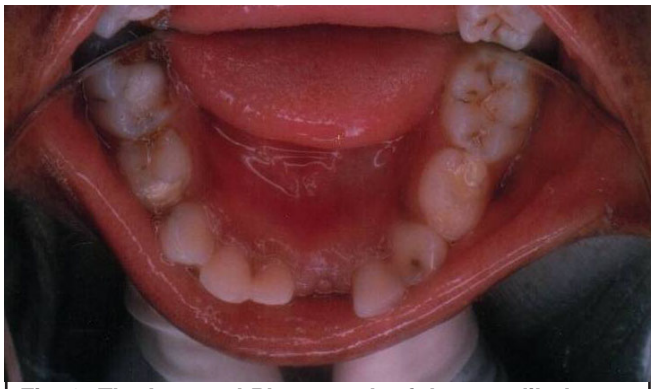

Fig. 3: The Intraoral Photograph of the mandibular arch with carious teeth 74,75,83, 84, 85. (Mirror View)

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.(1-2) 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.(6-8) This failure may be two-fold: a constitutive defect (probably in NK cell

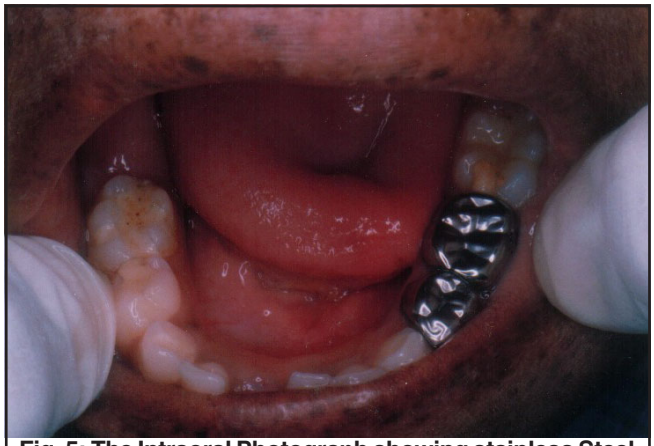

Fig. 5: The Intraoral Photograph showing stainless Steel crowns with 74,75 and Restorations with 83,84,85.

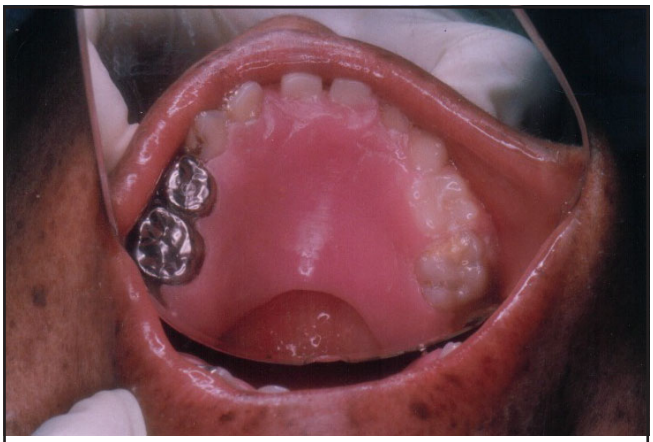

Fig. 6: The Intraoral Photograph of the maxillary arch showing the Functional Removable Space maintainer. (Mirror View)

function) exacerbated by a UV-dependent impair-ment (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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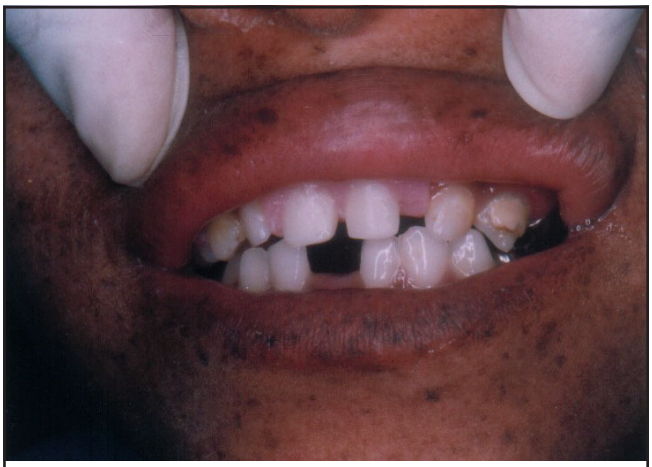

Fig. 7: The frontal view depicting the occlusion

References

1. Rook A, Wilkinson, DS, Ebling FJ. Textbook of Dermatology 1968: I: Oxford, Blackwell.
2. Gartler SM. Inborn errors of metabolism at the cell culture level. 2nd International Conference on Congenital Malformations 1963 Edited by M Fishbein. New York, International Medical Congress, Ltd, p 94.
3. Cleaver JE. Defective repair replication of DNA in xeroderma pigmentosum. Nature (Lond) 1968;218:652-656.
4. Cleaver JE. DNA damage and repair in light-sensitive human skin disease. Journal of Invest Dermatology 1970;54:181.
5. Bridges BA. How important are somatic mutations and immune control in skin cancer? Reflections on xeroderma pigmentosum. Carcinogenesis 1981;2:471-472.
6. Berkel AI, Kiran O. Immunological studies in children with xeroderma pigmentosum. Turkish Journal of Pediatrics 1974;16: 43-52.
7. Wysenbeek AJ, Weiss H, Duczyminer-Kahana M, Grunwald MH, Pick AI. Immuno-logic alterations in xeroderma pigmentosum patients. Cancer 1986;58:219-221.
8. Morison WL, Bucana C, Hashem N, Kripke ML, Cleaver JE, German JL. Impaired immune function in patients with xeroderma pigmentosum. Cancer Research 1985;45:3929-3931.
9. Bridges B. Sunlight DNA damage and skin cancer: A new perspective. Jpn Journal of Cancer Research1990;81:105-107.
10. Dilek FH, Akpolat N, Metin A. Ugras: Atypical fibroxanthoma of the skin and the lower lip in xeroderma pigmentosum. British Journal of Dermatology 2000;143(3):618-20.
11. HoferA, Kaddu S, Seidl H, *et al.* Collision of squamous-cell carcinoma with melanoma in situ in a child with xeroderma pigmentosum. Dermatology 2001;203(1):66-69.