

## Case series

**Xeroderma pigmentosum with aggressive periodontitis - A rare case series**Shanmugapriya SundarRaj<sup>1</sup>, SuganthaPriya SaiRam<sup>1</sup>, SaravanaPriya Mohan Kumar<sup>2</sup><sup>1</sup>Department of Dental Surgery, Thanjavur Medical College, Thanjavur, 600 004, Tamil Nadu, India<sup>2</sup>Department of Dental Surgery, Government Sivagangai Medical College, Rajaduraiasingam, Sivagangai, 630 561, Tamil Nadu, India

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Corresponding author: **Shanmugapriya SundarRaj**. Email: brightsmilesever@mail.com**ABSTRACT**

Xeroderma pigmentosum (XP) is a rare hereditary disease which is characterized by photosensitivity, abnormal pigmentations with freckling, actinic hyperkeratosis, scarring and prematurity of skin along with malignancies of ectodermal origin. The defect is in the normal DNA repair gene which causes the typical clinical manifestations depending on the degree of ultraviolet light exposure, especially in the sun-exposed areas of the skin and mouth. The disorder is often found among populations where marriage of close blood relatives is common. Oral manifestations usually present as actinic cheilitis, and squamous cell carcinoma of lip and tongue. Here we report two cases of xeroderma pigmentosum with oral findings as generalized aggressive periodontitis which is a rare manifestation.

**Keywords:** Xeroderma pigmentosum; hyperpigmentation; skin malignancies; aggressive periodontitis.

**INTRODUCTION**

**X**eroderma pigmentosum (XP) is an autosomal recessive inherited rare geno-dermatoses, characterized by photosensitivity, pigmentations, premature aging and malignancies of skin at an early age, caused by mutations of DNA repair gene (1). In particular, affected individuals have a 1,000-fold increase in the incidence of sunlight-induced skin cancers (2). Here we report two cases, who are cousins and presented with oral manifestations.

**Case report 1**

An 18-year-old girl reported a complaint of a painful tooth on the left lower jaw lasting two days. She presented with numerous brownish-black pigmentation of the face and hands. History revealed that she was one of the two children born to a consanguineously married parents and her younger brother reported not to have such features. The pigmentation which was initially less in number had progressed with age. She also revealed that she has a burning sensation of skin when exposed to sunlight.

On examination, generalized hyper-pigmented skin spots were present on the hands, neck, trunk, groin, thighs, and legs. Extra oral examination revealed that the skin of the face, limbs were dry and had few gray hairs on the scalp (Fig.1.A). Eyelids presented with sparse eyelashes with mild depigmentation on the upper and lower marginal periocular skin (Fig.1.B). There were few pigmented spots on the lips also.



**Fig. 1(A): Case 1:** Photograph of face showing numerous hyperpigmentation of face and lips



**Fig. 1(B):** Photograph of the left eye showing hypopigmented periocular skin (black arrow) with scarce eyelashes

Intra-orally, generalized mild deposition of calculus with severe periodontitis. Grade 3 mobility of 16, 11, 21, 26, 36, 31, 41, and 46 was present (Fig.1.C). OPG revealed generalized severe interdental bone loss suggestive of aggressive periodontitis (Fig.1.D).



**Fig.1(C):** Intra-oral photograph showing aggressive periodontitis.



**Fig.1 (D):** OPG revealing, generalized severe interdental bone loss.

## Case report 2

The second case is the cousin of the first girl who was a 21 years old girl, who also reported to the department to replace her loose upper and lower partial dentures. She also had generalized hyper-pigmented spots (Fig.2.A). Intraorally, multiple missing teeth with mobility of the remaining teeth were present (Fig.2.B).



**Fig. 2(A): Case 2:** Photograph of the face with hyperpigmentation



**Fig. 2(B):** Intra-oral photograph showing presence of few teeth with periodontitis

Denture stomatitis was evident because of the poor oral hygiene. OPG revealed extensive alveolar bone loss in both the arches with almost all the teeth floating (Fig.2.C).



**Fig. 2(C):** OPG revealing, multiple missing teeth and extensive interdental bone loss in the upper and lower arches

## DISCUSSION

Xeroderma pigmentosum (XP) is portrayed as a skin disorder with hypersensitivity to sunlight, subsequent pigmentation and greater chances of skin malignancy. In late 1800, Moritz Kaposi, a Wiener dermatologist initially put forth the term “xeroderma” to particularize the dry, dyspigmented alterations of skin in these patients (3). Herba and Kaposi first described Xeroderma Pigmentosum in 1974 (4). In 1968, James Cleaver (1) conducted a study which demonstrated that fibroblasts from patients with XP were unable to perform nucleotide excision repair (NER) of DNA damaged by UV radiation. XP arises due to inefficient Nucleotide Excision Repair (NER) pathways that are necessary in recognizing UV induced cyclo-pyrimidine dimers (CPDs), 6-4 photoproducts(6-4PPs) and clearing UV radiation induced photolesions. Mutations in any of the seven genes that encode proteins for the NER pathway end up in XP (5).

Hence XP has seven genetic subgroups from XP-A to XP-G. XP-V is a variant with normal NER pathway but an inadequate DNA polymerase that are integral for replicating damaged DNAs (6).

Xeroderma pigmentosum has worldwide prevalence in all races with an overall prevalence of 1–4% per million (7) and with equal gender predilection. The frequency of incidence is higher in countries where higher levels of consanguinity exist, like in India, incidence is quoted at 1:10,000-30,000 (8). Both familial and sporadic forms are reported.

The patients are often offspring of consanguineous marriages. The parents, the presumed heterozygotes, are clinically normal. The present cases were born to consanguineously married parents.. Several agents that cause DNA-damage includes UV radiation, Psoralens plus-long wavelength-UV radiation (PUVA), chlorpromazine, nitrofurantoin, mitomycin C, anthramycin, cisplatin, carmustine, aflatoxin, benzopyrenes, nitroquinoline oxide derivatives,

phenanthrene derivatives are some of the drugs found to be causative agents in the etiopathogenesis.

Clinical manifestations depend on the degree of UV light exposure at the sun exposed sites. Dermatological changes typically, in the early stage, present as disseminated erythema with small pale brown spots over light-exposed unprotected areas on the face, neck, trunk and limbs. Thinning and mottling of skin, telangiectasias, are seen in advanced cases.

XP patients are in a perilous state of developing melanoma and basal cell carcinoma during early decades of life (9). Ocular findings like photophobia, periocular skin changes, conjunctival and corneal damages are seen which on protracted period leads to corneal scarring and visual impairment.

Mucocutaneous malignancies are common in tip of the tongue and lips, being reported as squamous cell carcinomas, basal cell carcinomas, melanomas and sarcoma in descending frequency (10). Leukoplakia, erythroplakia and actinic cheilitis, desquamative gingivitis, restricted mouth opening, fissured tongue, atrophy of tongue papillae are also associated. The key points in clinical differential diagnosis of carcinomas of oral cavity between non-XP and XP populations, is that, the previous group are elderly, Sought to have tobacco consumption, site predilection to base, posterior-lateral borders and floor of the mouth. Whereas, the later individuals are in early decades of life, sites affected are the tip of the tongue and lips with slow progression (11).

In our report, both case 1 and case 2 presented with generalized aggressive periodontitis with extensive interdental bone loss which is a finding rarely reported. Aggressive periodontitis may be attributed by several immunological deficiencies in XP patients such as reduced T4 positive lymphocyte subset (12). Neurological problems and intellectual deficiency also have been reported in 20% to 30% patients. The period of onset varies from 2 to 30 years of age (13).

No cure is available for XP. Affected individuals have to abstain themselves from exposure to all sources of harmful radiations and to strictly adhere to all protective measures. Chemotherapy with topical 5-fluorouracil is useful for actinic keratosis. Complete excision of associated malignancies is the surgical care. Avoidance of smoking and environmental carcinogenic agents is also recommended.

Prognosis is good for the patients without neurological symptoms, precancerous and cancerous lesions. However, they should follow the strict protective protocols. Besides, neurological, dermatological and ophthalmic care, dental and oral care should also be strictly adhered to with constant follow-ups. In the reported two cases, extraction of

all hopeless teeth and rehabilitation was recommended.

## CONCLUSION

As XP patients are more prone to dermal and oral malignancies, it is mandatory that regular surveillance, early diagnosis and treatment of all malignancies are essential. Keen photo-protection measures must be ensured and a multidisciplinary team approach is critical in securing a better lifestyle in the affected population. Genetic counseling can be emphasized to discuss the effects of consanguineous marriages in communities where it is common.

## CONFLICT OF INTEREST

The authors declare no conflicts of interest.

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