Xeroderma pigmentosum with significant periodontal findings: a rare clinical report

Introduction

Xeroderma pigmentosum (XP) is a very rare skin disorder where a person is highly sensitive to sunlight (photosensitive), has premature skin aging and is prone to developing skin cancers. It is an autosomally recessive inherited disease, therefore, one recessive xeroderma pigmentosum gene is inherited from each parent. XP is characterised by dry, pigmented skin, spidery blood vessels in the skin and skin cancers. Approximately 80% of XP patients have ocular complications. These complications may include severe keratitis, which could be followed by corneal opacification and vascularisation. XP patients may lose their eyelashes, and in severe cases, 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 patients1,2.

Light-exposed XP patients are susceptible to a higher risk of skin cancer. This is because of a defect in DNA repair that results in an increased frequency of initiated (mutated) skin cells that are able to grow into tumorous colonies early in life, probably because of failure of the immune system to restrict their growth3-5.

Berkel and Kiran5 divided XP patients according to the extent of their cutaneous disease and there appeared to be an inverse relationship between disease severity and the development of contact allergy. XP is classified into eight complementation groups (or genetic subtypes), based upon different defects in the body’s ability to repair DNA. Only Type B (XP, Cockayne syndrome) is associated with dental findings, which are namely dental caries, delayed eruption of teeth and malocclusion. To date, no case of XP with periodontal findings has been reported. Here we present a rare case of XP with oral and periodontal findings. The authors further hypothesised that periodontal destruction in XP patients is due to immune defects present in such patients.

KEY WORDS  bone loss, immunologic abnormalities, periodontitis, xeroderma pigmentosum
findings, and these include: dental caries, delayed eruption of teeth and malocclusion. To the authors' knowledge, this is the first report of xeroderma pigmentosa associated with significant oral and periodontal findings.

Case report

A 14-year-old female patient with xeroderma pigmentosum (XP), the elder of a two-child family, was referred to the Department of Periodontics, Government Dental College and Hospital, Bangalore for oral and periodontal evaluation. The medical history revealed that the patient had been diagnosed with XP at the age of 2. The patient was apparently normal in the first year of her life, after which she developed freckling, which began on the face and spread to the arms, neck, trunk, and legs. There was also a history of photophobia and burning sensation of the skin on sun exposure. Family history revealed consanguineous marriage of her mother, and the patient's cousin also suffered from XP.

The following clinical extra-oral findings were observed: physical examination showed that the patient had numerous localised small (1 to 4 mm) hyperpigmented macules on the face, as well as on all sun-exposed areas (Figs 1 and 2). Additionally, there was skin dryness and marked skin changes on areas that were exposed to sunlight. Ophthalmic examination revealed a heavily pigmented iris (Fig 3) and freckles on the eyelids. Intra-oral examination revealed severe gingival inflammation, and periodontal probing resulted in gingival bleeding (Figs 4 and 5). Gingival bleeding had been noticed by the patient's mother a few months prior to the examination. Probing pocket depths ranged from 4 to 6 mm and attachment loss ranged from 6 to 7 mm (Fig 6). Mobility was assessed through Miller's mobility index and it was found that grade II mobility was present at teeth 31, 32, 41 and 42. Radiographs revealed periodontal breakdown around the molars and incisors. The maxillary central incisors had lost more than 70% of their supporting alveolar bone (Fig 7). In addition, furcation involvement was found in all of the molars. The amount of local factors were not commensurate with the amount of alveolar bone destruction. Delayed eruption of teeth was observed, with unerupted teeth showing little or no bone covering the occlusal surface of the teeth. Microbiological testing revealed mainly Gram-negative strict anaerobes.

Routine blood investigations revealed a normal blood picture, and a skin biopsy was performed from the ventral surface of the arm. Histopathological investigations revealed mild hyperkeratosis, partial atrophy of epidermis, irregular accumulation of melanin in the basal layer and focal chronic inflammatory infiltration seen in the upper dermis, and these features are suggestive of XP (Fig 8).

Periodontal treatment, which included professional prophylaxis, was performed after informed consent had been received. In addition, detailed oral hygiene instructions were given to the patient's mother, who assured routine care for the patient.
would be carried out. Periodontal treatment consisted of thorough scaling and root planing for the whole dentition. Revaluation was performed 1 month after completion of the treatment. Plaque index (PI)\textsuperscript{8} was measured and at the beginning of the study, the mean PI score was 2.0. This value was reduced to 0.7 after the treatment. The percentage reduction in bleeding on probing was 64% at 1 month post-therapy.

The pocket depths were reduced, and recession of the gingival margin could be noted in several regions of the dentition. However, the inflammatory component was present at the end of the three sessions of scaling and root planing.

\section*{Discussion}

XP is a very rare skin disorder where a person is highly sensitive to sunlight (photosensitive), has premature skin aging and is prone to developing skin cancers. At present there is no cure for XP. The main goal of the treatment is to protect the patient from UV radiation exposure by application of sunscreens and, thus, prevent the damaging effects it can have on the skin.

The case reported here presented signs similar to those of severe periodontal disease, namely gingival bleeding, gingival recession, pocket formation, tooth mobility and alveolar bone loss, both in the maxilla and in the mandible. In this case, the occurrence of

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\includegraphics[width=\textwidth]{severe_gingival_inflammation.png}
\caption{Severe gingival inflammation.}
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\includegraphics[width=\textwidth]{deep_periodontal_pockets.png}
\caption{Deep periodontal pockets.}
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\caption{Orthopantomograph.}
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\caption{Histopathological picture.}
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periodontal disease associated with the systemic disease was suggested. It is well known that if the patient presents with systemic involvement, the periodontal tissue may respond in an altered manner when encountering an offending agent, which leads to the classification of periodontal disease associated with systemic diseases. The result is often the occurrence of rapidly progressive periodontitis.

Further, presence of advanced periodontal destruction can be explained in many possible ways. Patients with XP have immunological abnormalities, which are characterised by defects in cellular immunity, impaired natural killer cell activity and reduced interferon production. Altered cytokine induction may also contribute to the immune defect in XP patients. In addition, XP patients present with a low number of circulating T cells. All these factors may affect the regulation of immune responses against intracellular and extracellular antigens and, hence, can be further hypothesised to have caused periodontal destruction in the present case.

Further, XP patients show premature aging of the skin, eyes and mouth. This is a specific feature of XPB. The periodontist plays an important role in the surveillance of oral and perioral structures, in addition to reinforcement of preventive oral care in XP patients. The clinician is advised against the use of UV light-curing units in these patients because UV-induced epithelial damage may cause dysplasia, as DNA repair mechanisms are dysfunctional in such patients.

This case report shows XP with very significant periodontal findings, which have not been previously reported.

References