

# Xeroderma Pigmentosum: Case Report

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## ABSTRACT

**Aim** Xeroderma pigmentosum, which is commonly known as XP, is an inherited condition characterized by an extreme sensitivity to ultraviolet (UV) rays from sunlight. This condition mostly affects the eyes and areas of skin exposed to the sun. Some affected individuals also have problems involving the nervous system. In affected individuals, exposure to sunlight often causes dry skin (xeroderma) and changes in skin coloring (pigmentation). The aim of this report is to present a case of xeroderma pigmentosum in a female patient.

**Case Report** A 33-year-old female patient was referred to Marmara University, Faculty of Dentistry Oral Diagnosis Clinic. A 33-year-old female patient was admitted to Marmara University Faculty of Dentistry Oral Diagnosis Clinic with the complaint of pain. All systemic findings of the syndrome were observed. In the intraoral examination, no findings were found in the oral mucosa.

**Discussion** People with xeroderma pigmentosum have a greatly increased risk of developing skin cancer. Without sun protection, about half of children with this condition develop their first skin cancer by age 10. Most people with xeroderma pigmentosum develop multiple skin cancers during their lifetime. These cancers occur most often on the face, lips, and eyelids. Cancer can also develop on the scalp, in the eyes, and on the tip of the tongue.

**Conclusion** Patients should be kept under control because of squamous cell carcinoma that can be observed in the oral mucosa.

**Keywords** Oral cancer, Oral diagnosis, Oral mucosa, Squamous cell carcinoma, Xeroderma pigmentosum

## Introduction

Xeroderma pigmentosum (XP) is an autosomal recessive disease characterized by severe photosensitivity, abnormal pigmentation and a more than 1000-fold increase in the frequency of all types of major skin cancers (basal cell cancers, squamous cell cancers, malignant melanoma) in areas exposed to sunlight compared to normal population (1,2).

The prevalence of XP is relatively high in the Middle East and Japan. In addition, several cases have been reported in Africa. Xeroderma pigmentosum is estimated to affect about 1 in 1 million people in the United States and Europe (3-9).

Researchers have identified at least eight inherited forms of xeroderma pigmentosum: complementation group A (XP-A) through complementation group G (XP-G) plus a variant type (XP-V). The types are distinguished by their genetic cause. All of the types increase skin cancer risk, although some are more likely than others to be associated with neurological abnormalities. This condition mostly affects the eyes and areas of skin exposed to the sun (10).

The signs of xeroderma pigmentosum usually appear in infancy or early childhood. Many affected children develop a severe sunburn after spending just a few minutes in the sun. The sunburn causes redness and blistering that can last for weeks. Other affected children do not get sunburned with minimal sun exposure, but instead tan normally. By age 2, almost all children with xeroderma

pigmentosum develop freckling of the skin in sun-exposed areas (such as the face, arms, and lips); this type of freckling rarely occurs in young children without the disorder. In affected individuals, exposure to sunlight often causes dry skin (xeroderma) and changes in skin coloring (pigmentation). This combination of features gives the condition its name, xeroderma pigmentosum (2,11).

People with xeroderma pigmentosum have a greatly increased risk of developing skin cancer. Without sun protection, about half of children with this condition develop their first skin cancer by age 10. Most people with xeroderma pigmentosum develop multiple skin cancers during their lifetime. These cancers occur most often on the face, lips, and eyelids. Cancer can also develop on the scalp, in the eyes, and on the tip of the tongue. Studies suggest that people with xeroderma pigmentosum may also have an increased risk of other types of cancer, including brain tumors. Additionally, affected individuals who smoke cigarettes have a significantly increased risk of lung cancer (11).

The eyes of people with xeroderma pigmentosum may be painfully sensitive to UV rays from the sun. If the eyes are not protected from the sun, they may become bloodshot and irritated, and the clear front covering of the eyes (the cornea) may become cloudy. In some people, the eyelashes fall out and the eyelids may be thin and turn abnormally inward or outward. In addition to an increased risk of eye cancer, xeroderma pigmentosum is associated with noncancerous growths on the eye. Many of these eye abnormalities can impair vision (12).

The aim of this report is to present a case of xeroderma pigmentosum in a female patient.

## Case Report

A 33-year-old female patient was referred to Marmara University, Faculty of Dentistry Oral Diagnosis Clinic. The patient

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*\*This study was presented as a poster presentation at the 22nd BaSS Congress, Thessaloniki, Greece, 4 - 07 May 2017.*

Received: 16.03.2023 / Accepted: 03.04.2023 / Published: 31.08.2023

came to the faculty to have dental treatment. She has had a xeroderma pigmentosum since 8 years old and she has 40 operations since the age of 10. Because of these surgical procedures, she lost her nose and part of her upper and lower lip. She has been using nose prosthesis since 2002. She went through unilateral exenteratio orbitae due to cancer invading orbital spaces (Figure 1,2).



**Figure 1:** Atrophic, hypo-hyperpigmented skin in a female patient affected by Xeroderma pigmentosum.

Cutaneous examination revealed area of mottled hyper and hypopigmentation involving the face, neck and the hand base of the patient (Figure 1, 2). She had limitation of mouth openness because of xeroderma pigmentosum and in the intraoral examination, no signs of malignancy in the oral mucosa were detected (Figure 3), and the tooth causing pain was extracted, and the dental examination and treatment was completed (Figure 4). The patient was under plastic surgery control.

## Discussion

The first persistent cutaneous change seen in patients with this illness was dry, dyspigmented skin, which was initially described as “xeroderma” by Moritz Kaposi in 1870. In the first dermatological textbook that he co-wrote with Professor Ferdinand Hebra in 1874, Kaposi documented four patients who had xeroderma, also known as “parchment skin”. Dr. Albert Neisser initially recorded an XP case with neurological symptoms, and DeSanctis and Cacchione helped create the name “DeSanctis-Cac-

chione syndrome” in 1932 to refer to XP individuals with severe neurological deficiencies. The condition was previously divided into two categories: classical XP, which only causes skin abnormalities, and DeSanctis-Cacchione syndrome, which causes both skin abnormalities and severe neurological degeneration. There are at least eight hereditary types of xeroderma pigmentosum (11,13,14).



**Figure 2:** Loss of part of the nose, upper and lower lip after surgical procedures in the patient.

The complementation group, the specifics of the mutation, and other factors all affect the clinical characteristics. As a result, there is a large range in clinical characteristics. Bright environments, outdoor activities, fair skin, smoking, a lack of diagnostic resources, a delay in diagnosis, and inadequate sun protection will increase cutaneous abnormalities, leading to a variety of pigmentation alterations, a variety of skin malignancies, and early death (15). While the other 40% of cases do not exhibit any sunburn reaction, about 60% of cases exhibit acute sensitivity to sunlight as the initial symptom, which takes many days or weeks to cure. A common condition is photophobia (16).



**Figure 3:** Decreased mouth opening affecting oral hygiene

Without UV protection, the skin ages and atrophies, becoming dry and harsh. Lentigines proliferate and become warty as well as becoming more numerous, darker, and clinically difficult to identify from the numerous, flat, pigmented seborrheic warts. Tiny, hypopigmented macules are frequently found among the lentigines, giving birth to the distinctive salt-and-pepper pattern of skin's mottled hyperpigmented and hypopigmented look. In these patients, the skin of the nose frequently exhibits an atrophic, hypopigmented region. Telangiectasia may appear later. There may be stucco keratosis, which are easily distinguished from sun keratosis (17).



**Figure 4:** Panoramic radiograph of the patient

Although UV radiation exposure is the cause of all skin changes, the degree of protection the skin has from sunlight, the Fitzpatrick skin type, and the duration of sun exposure all have a direct impact on how severe these changes will be. Individuals experience a wide range of consequences. Due to the photoprotective qualities of melanin, those with darker skin often experience a lower incidence of skin cancer than those with lighter skin. Dark-skinned and light-skinned individuals with XP, however, had comparable rates of skin cancer, highlighting the crucial significance of DNA repair mechanisms even in the presence of melanin protection (18).

Actinic cheilitis and SCC of the lips, as well as leukoplakia, erythroplakia, and SCC of the tongue's tip, are linked to XP [18]. UV radiation is thought to be the cause of the precancerous and cancerous lesions on the tip of the tongue, which are uncommonly affected in the general population. Although it is the only one provided, this explanation is not compelling. The posterolateral and ventral sides of the tongue and the floor of the mouth are most frequently affected by SCC in the general population, and it progresses aggressively among older smokers and drinkers. XP-related SCC, on the other hand, affects those under the age of 20 and manifests as a slowly progressing condition that affects the tip of the tongue. When a patient opens their mouth for breathing, speaking, eating, or doing an oral hygiene routine, a fibrous area that has been the result of multiple labialplasties stretches and causes pain. Because of this, the patient has bad hygiene practices, which contribute to a high prevalence of dental plaque, caries, and periodontal disease. Fissured tongue, keratoacanthoma, and persistent desquamative gingivitis cases have all been documented (19-21).

Although the skin and perioral findings of the case pre-

sented in this study are consistent with the literature findings, no pathology was detected in the intra-oral findings.

## Conclusion

The quality of life and life expectancy of those who have XP can be significantly increased even if there is no known treatment. This is due to increased awareness, crucially early detection, strict sun protection, and attentive patient management.

## Declarations

**Author Contributions:** Conception/Design of Study- F.N.P.; Data Acquisition- F.G.D., A.N.S.; Data Analysis/Interpretation- F.N.P., F.G.D., A.N.S.; Drafting Manuscript- F.N.P., F.G.D., A.N.S.; Critical Revision of Manuscript- F.N.P.; Final Approval and Accountability- F.N.P.; Material and Technical Support- F.N.P., F.G.D., A.N.S.; Supervision- F.N.P.

**Conflict of Interest:** Authors declared no conflict of interest.

**Financial Disclosure:** Authors declared no financial support.

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