Case Report ISSN: 2836-3647

American Journal of Pathology & Research

Xeroderma Pigmentosum Complicated by Squamous Cell Carcinoma of the Base of the Tongue: A Case Report from the University Clinics of Lubumbashi

Pierre Kitha[1,3*], Didier Munganga[1,2], Mika Mulewa[1,5], Florent Zeng[1,6], Didier Mwenze[1,4], Veronique Cathy Kyabu[1,4], Mamy NKumuyaya[1,2], Lydia Mwelwa[1,2], Maryel Ragi Lya Lushule[1,3] and Robert Lukamba[1,2]

[1]*Faculty of Medicine, University of Lubumbashi, Democratic Republic of the Congo.*

[2]*University Clinic, Pediatric Oncology Unit, Democratic Republic of the Congo.*

[3]*University Clinic of Dermatology–Venereology, CUL, Democratic Republic of the Congo.*

[4]*University Clinic of Pathology, CUL, Democratic Republic of the Congo.*

[5]*University Clinic of Medical Imaging, CUL, Democratic Republic of the Congo.*

[6]*University Clinic of Pediatric Surgery, CUL, Democratic Republic of the Congo.*

***Correspondence:**
Pierre Kitha, Assistant, Faculty of Medicine, University of Lubumbashi, Democratic Republic of the Congo, Tel: +243 97 54 75 439.

Received: 06 Dec 2025; **Accepted:** 14 Jan 2026; **Published:** 26 Jan 2026

Citation: Pierre Kitha, Didier Munganga, Mika Mulewa, et al. Xeroderma Pigmentosum Complicated by Squamous Cell Carcinoma of the Base of the Tongue: A Case Report from the University Clinics of Lubumbashi. American J Pathol Res. 2026; 5(1): 1-3.

ABSTRACT

Xeroderma pigmentosum (XP) is a rare disease characterized by a defect in the repair of UV-induced damage. It is marked by photosensitivity with a tendency to develop skin burns after minimal sun exposure, early freckles, and the development of lentiginous pigmentation, as well as other features of poikiloderma and a predisposition to developing skin cancer at an early age. We report the case of a 10-year-old schoolboy, born to non-consanguineous parents, with a history of photophobia beginning at the age of 2. He presented to the pediatric oncology department of the University Clinics of Lubumbashi with an asymptomatic growth on the tongue that had been progressively enlarging for 3 years. The persistent increase in size and difficulty eating prompted the consultation. A diagnosis of xeroderma pigmentosum (XP) complicated by poorly differentiated squamous cell carcinoma of the base of the tongue, without metastasis, was established. The family's socioeconomic situation and the unavailability of radiotherapy in Lubumbashi did not allow for adequate management of the tumor. A palliative excision of the mass was proposed, but the patient left the hospital against medical advice.

Keywords

Xeroderma pigmentosum, Squamous cell carcinoma, Base of the tongue, Lubumbashi.

Introduction

Xeroderma pigmentosum (XP) is a rare disease characterized by a defect in repairing UV-induced damage. It is marked by photosensitivity, with a tendency to develop skin burns after minimal sun exposure, early freckles, and the development of lentiginous pigmentation, as well as other features of poikiloderma and a predisposition to developing skin cancer at an early age [1,2]. Beyond cutaneous complications, patients are susceptible to ocular disorders, neurodegenerative processes, central nervous system tumors, and other neoplasms resulting from exposure to UV radiation and its byproducts. The life expectancy of patients with XP is reduced due to skin cancer and neurodegenerative sequelae.

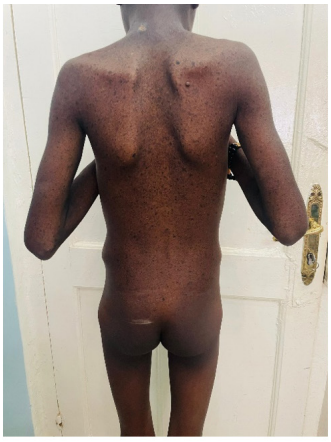
However, aggressive preventive measures aimed at minimizing UV exposure and the resulting damage can improve disease progression and extend survival. There is no causal treatment for this rare autosomal recessive disease, highlighting the importance of early diagnosis and UV protection measures, such as reducing exposure to environmental UV radiation and undergoing regular skin cancer screening [3,4].

We report a case of XP complicated by squamous cell carcinoma of the nasopharynx.

Observation

The patient was a 10-year-old schoolboy born to non-consanguineous parents, with a history of photophobia beginning at the age of 2. He presented to the pediatric oncology department of the University Clinics of Lubumbashi with an asymptomatic growth on the tongue that had been continuously evolving for 3 years, with a progressive increase in size. The persistent enlargement and difficulty eating motivated the consultation.

On examination, a globular tumor mass was observed, measuring approximately 12 × 18 cm in diameter. It was firm in some areas and soft in others, bled on contact, and was tender on deep palpation. The surface was ulcerated with fibrous areas in some regions. In addition, photophobia and diffuse poikiloderma were noted.



Figures 1 and 2: Poikiloderma of XP on the back and face, and photophobia of both eyes.

The biopsy was performed and revealed a cellular proliferation forming clusters and solid sheets that proliferated with marked cytonuclear atypia and fibroconnective infiltration. The thoraco-abdomino-pelvic CT scan did not show any pleural or parenchymal lesions of metastatic nature.

A diagnosis of xeroderma pigmentosum (XP) complicated by poorly differentiated squamous cell carcinoma of the base of the tongue, without metastasis, was established.

The family's socioeconomic situation and the unavailability of radiotherapy in Lubumbashi did not allow for adequate management of the tumor. A palliative excision of the mass was proposed, but the patient left the hospital against medical advice.

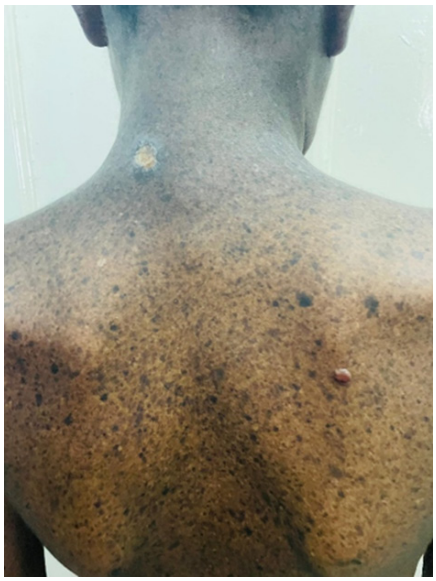


Figure 3: Poikiloderma of the back.

Figure 4: Squamous cell carcinoma of the base of the tongue in a patient with XP.

Argument

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease that disrupts the repair of deoxyribonucleic acid (DNA) following ultraviolet (UV) radiation. XP is characterized by extreme sensitivity to sunlight, photophobia, and skin lesions in the form of hyperpigmented macules resembling freckles, along with skin neoplasms. Malignancy is a common complication observed in UV-exposed areas. Squamous cell carcinoma (SCC) is the most frequent malignant tumor in patients with XP [5,6].

Ocular symptoms are often the initial signs of this disease, alongside the poikilodermic appearance of the entire body. XP is a fertile ground for malignant degeneration. Squamous cell carcinoma is typically the first cancer to appear, followed by basal cell carcinomas and, much later, melanomas. Involvement of the nasopharynx remains very rare in this disease [7,8]. The most commonly reported site of squamous cell carcinoma in patients with XP is the tongue. Nasopharyngeal involvement in our patient is therefore unusual.

Conclusion

XP is a rare genodermatosis that is more commonly observed in contexts where consanguinity is present. It can be complicated by tumors, particularly squamous cell carcinomas. Nasopharyngeal involvement is rare.

References

1. Black JO. Xéroderma Pigmentosum. Head Neck Pathol. 2016; 10: 139-144.
2. Lehmann J, Seebode C, Martens MC, et al. Xeroderma Pigmentosum - Facts and Perspectives. Anticancer Res. 2018; 38: 1159-1164.
3. Cleaver JE. Common pathways for ultraviolet skin carcinogenesis in the repair and replication defective groups of xeroderma pigmentosum. J Dermatol Sci. 2000; 23: 1-11.
4. Boyle J, Ueda T, Imoto K, et al. Persistence of repair proteins at unrepaired DNA damage distinguishes diseases with ERCC2 (XPD) mutations: cancer-prone xeroderma pigmentosum vs. non-cancer-prone trichothiodystrophy. Hum Mutat. 2008; 29: 1194-1208.
5. Effendi RMRA, Fadhlih A, Diana IA, et al. Xeroderma Pigmentosum with Simultaneous Cutaneous and Ocular Squamous Cell Carcinoma. Clin Cosmet Investig Dermatol. 2022; 15: 157-161.
6. Brooks BP, Thompson AH, Bishop RJ, et al. Ocular manifestations of xeroderma pigmentosum: long-term follow-up highlights the role of DNA repair in protection from sun damage. Ophthalmology. 2013; 120: 1324-1336.
7. Lim R, Sethi M, Morley AMS, et al. Ophthalmic Manifestations of Xeroderma Pigmentosum: A Perspective from the United Kingdom. Ophthalmology. 2017; 124: 1652-1661.
8. Leung AK, Barankin B, Lam JM, et al. Xeroderma pigmentosum: an updated review. Drugs Context. 2022; 11: 2022-2-5.