Our Dermatology Online

Brief Report

# Xeroderma pigmentosum: Twelve cases at the National Hospital of Niamey, Niger

**Salissou Laouali[1], Nouhou Diori Adam[2], Hamani Issaka[3], Oueodraogo Mamadou Maïmouna[1], Laouali Idi Mamane Sani[1], Moussa Doulla[1], Nouhou Hassan[4]**

*[1]Department of Dermatology Venereology, National Hospital of Niamey, Niger, [2]Department of Ophthalmology, National Hospital of Amirou Boubacar Diallo of Niamey, Niger, [3]Laboratory of Cytology and Genetic, Faculty of Health Sciences, ABDOU Moumouni University of Niamey, Niger, [4]Laboratories of Pathological Anatomy, Faculty of Health Sciences, ABDOU Moumouni University of Niamey, Niger*

**Corresponding author:** Salissou Laouali, MD, E-mail: danmata@yahoo.com

## ABSTRACT

**Background:** Xeroderma pigmentosum (XP) is a usually autosomal recessive disorder linked to a deficiency of the enzyme systems of DNA repair. The pathological sensitivity to the sun exposes the patient to develop multiple cancers. **Materials and Methods:** We conducted a retrospective study over a period from January 2006 to December 2014. A total of twelve patients were enrolled. **Results:** These were nine male cases and three female, with a sex ratio of 3/1. The average age was 7.9 years, ranging from 1 to 40 years. Consanguinity between the parents was found in ten cases (83.3%). The first non-tumor cutaneous manifestations appeared in eight patients before the age of six months. During follow-up, seven patients, including three (43%) at the age of eight years, died from metastasis. **Conclusion:** XP is complicated in the development of cancers, even in children, and is linked to the intensity of solar radiation in Niger.

**Keywords:** xeroderma pigmentosum; skin cancers; tongue amputation; Niamey, Niger

## INTRODUCTION

Xeroderma pigmentosum (XP) is a usually autosomal recessive disorder, linked to a deficiency of the enzyme systems of DNA repair. It is characterized by a pathological sensitivity to the sun, exposing the patient early to develop cancer. It was first described in 1970 by Hébra and Kaposi. It is a relatively rare disease whose frequency varies from 1/10,000 (Tunisia) to 4/1,000,000 in Europe and the U.S. [1-3]. No currently available treatment seems completely effective; the evolution is constantly toward aggravation and death by cachexia due to the outbreak of multiple cancers, in particular mucocutaneous, ocular, and/or neurological [4-6]. The objective of this study was to describe the epidemiological, clinical, and evolutionary profile of XP in Niger (West Africa), a tropical and especially sunny country.

## MATERIALS AND METHODS

This was a retrospective study conducted over a period of eight years from January 2006 to December 2014 at the National Hospital of Niamey, Niger. The diagnosis of XP was essentially clinical and based on the characteristic skin and/or mucous lesions: photophobia, xeroderma, conjunctivitis hemorrhagic, corneal pillowcases, and ulcerations. The epidemiological data studied were: age, sex, the duration of evolution on the first consultation. For each family of the patient, the following were researched: a notion of consanguinity among the parents, the existence of one or more similar cases in the family, and the economic situation of the parents. The diagnosis of a tumor lesion was confirmed by pathological examination.

**How to cite this article:** Laouali S, Adam ND, Issaka H, Maïmouna OM, Mamane Sani LI, Doulla M, Hassan N. Xeroderma pigmentosum: Twelve cases at the National Hospital of Niamey, Niger. Our Dermatol Online. 2023;14(1):56-59.

**Submission:** 26.05.2022; **Acceptance:** 17.08.2022

**DOI:** 10.7241/ourd.20231.11

## RESULTS

We collected twelve cases of XP, which were the subjects of this study, including nine male cases and three female, giving a sex ratio of 3/1. The average age was 7.9 years, ranging from 1 to 40 years. Consanguinity between the parents was found in ten cases (83.3%) out of the twelve, and the existence of similar conditions in the family was noted in nine cases (75%). All patients came from a poor socioeconomic background. The first non-tumor cutaneous manifestations appeared in eight patients before the age of six months. The average consultation time was 45 months, ranging from 2 to 360 months. Xeroderma was the most frequent reason for consultation on the cutaneous level (six cases) (Fig. 1a) and photophobia on the ophthalmological level (four cases) (Fig. 1b). The first cutaneous tumor appeared between the age of four and six years in seven cases (Table 1). On clinical examination, photophobia and xeroderma were found in all patients, who, moreover, all presented with a malignant tumor—six squamous cell carcinomas (SCC) and six basal cell carcinomas (BCC)—and the SCCs and BCCs were associated in three cases (Table 2). On average, there were three tumors in one patient, ranging from two to six, eight times out of the twelve patients (Figs. 2a and 2b). The locations were: ocular (five cases), labial (one case), nasal (three cases), and the tip of the tongue (four cases, including two amputations of the tongue). (Figs. 3a and 3b). The other mucocutaneous manifestations observed were, among others lentigines, dyschromic macules, corneal sheaths, corneal ulcerations, and hemorrhagic conjunctivitis (Table 3). Classic XP was observed in eleven cases; only one (aged forty) had a variant form (Fig. 4). On the therapeutic level, the patients received advice on photoprotection and the excision of tumor lesions was performed (twelve cases). During follow-up, seven patients, including three (43%) at the age of eight years, died from metastasis (Figs. 5a and 5b B).

## DISCUSSION

XP is a rare autosomal recessive disease whose sexual predominance varies depending on the study. The male predominance observed in our sample was already reported by some authors [2,6-8]. On the other hand, Chidzonga et al. [9] in Zimbabwe found eight females and four males, giving a sex ratio of 2/1. Khatri et al. [10] and Fazaa et al. [11] also found a female predominance, with rates of 23/19 and 7/5, respectively. The annual average concerning our series was 1.5 cases and was more important in consanguineous marriages [2,11,12]. In our study, as in most of the series reported, patients with XP were generally from consanguineous marriages. Gul et al. reported these in 83.3% of cases [8], Boujard et al. in 95% [11], and Dieng et al. in five out of six cases [13]. This could also explain the high frequency of similar cases in the family. Similar cases noted in the families of patients (83.3%) were reported in certain series [8,13-15]. The predominant achievement of the age group in our study was that of one to five years. For Witold [6], the most affected age group was that of ten months and twenty-one years and, for El

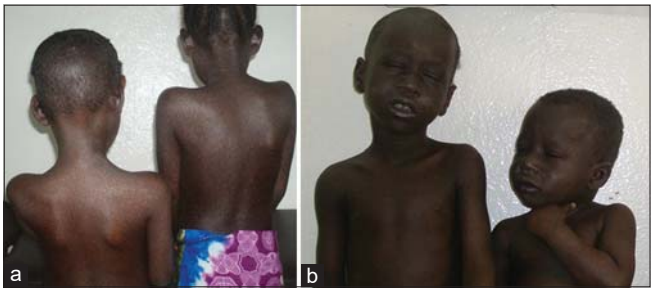



**Figure 1:** (a) Xeroderma, poïkiloderma (brother and sister); (b) photophobia (two brothers).


**Figure 2:** Ulcerative budding tumors of the cheeks in two patients with left corneal pillowcase in one patient: (a) Left cheek injury (SCC), (b) Right cheek injury (BCC).

**Table 1:** Distribution of the patients according to the age at the appearance of the first tumor

| Age of Onset of First Tumor (Year) | Number of Cases | Percentage |
|---|---|---|
| ≤ 3 | 2 | 16.7 |
| 4–6 | 7 | 58.3 |
| > 6 | 3 | 25.0 |
| Total | 12 | 100.0 |

**Table 2:** Types and numbers of carcinomas observed

| Type | Number of Cases | Percentage |
|---|---|---|
| Squamous cell carcinoma (SCC) | 6 | 60.0 |
| Basal cell carcinoma (BCC) | 6 | 60.0 |
| SCC and BCC | 3 | 30.0 |

In ten cases, the types and numbers of cancers were determined, that is, 60% of squamous cell carcinomas (SCCs), 60% of basal cell carcinomas (BCCs), and 30% of the two forms associated.

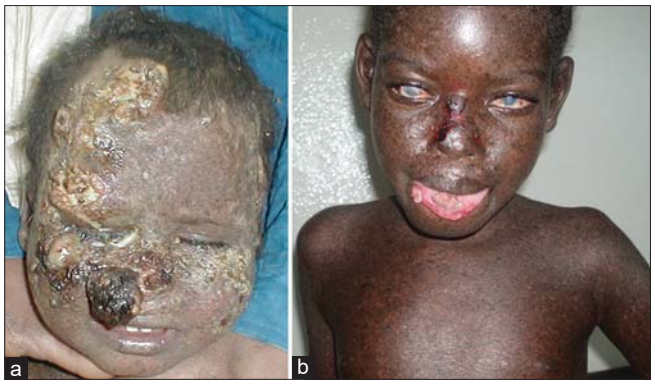
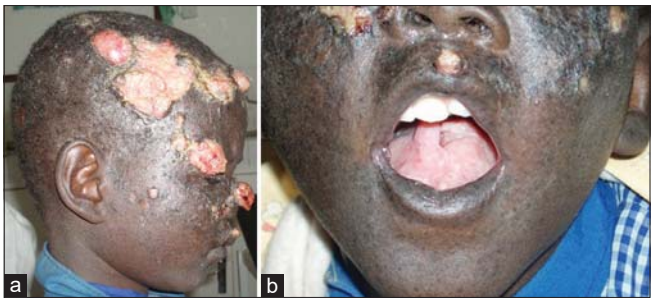


**Figure 3:** (a) Squamous cell carcinoma of the temple, check, nasal mucosa on the right and eyelid; (b) squamous cell carcinoma of the lip, tongue tip, and bilateral corneal pillowcase.

**Figure 4:** XP variant form in the forty-year-old adult (onset of signs at the age of thirty): xeroderma, skin tumors on the face, and a bilateral corneal sheath.



**Figures 5:** Multiple end-stage tumors in the same patient: (a) cutaneous involvement of the right scalp, (b) lingual involvement with its mutilation.

**Table 3:** Characteristics of the mucocutaneous and ophthalmological lesions

| Areas Affected and Type of Lesions | Number of Cases | Percentage |
|---|---|---|
| Damage to photo-exposed parts | 12 | 100.0 |
| Damage to hidden parts | 12 | 100.0 |
| Dyschromic macules | 12 | 100.0 |
| Xeroderma | 12 | 100.0 |
| Lentigine | 12 | 100.0 |
| Ulcerovetting tumors | 9 | 75.0 |
| Kerato-acanthomas | 5 | 41.6 |
| Botriomycomas | 6 | 50.0 |
| Tongue tumor | 4 | 33.3 |
| Tongue amputation | 2 | 16.6 |
| Actinic keratosis | 1 | 8.3 |
| Involvement of the eyelids | 12 | 100.0 |
| Hemorrhagic conjunctivitis | 12 | 100.0 |
| Corneal pillowcase | 12 | 100.0 |
| Photophobia | 12 | 100.0 |
| Corneal ulceration | 10 | 83.3 |
| Eye tumors | 5 | 41.7 |

Fékih et al., that of 17.6 ± 11.04 years [2]. As in the report by Witold [6], our patients also all came from families of low socioeconomic level, which explained the often long average consultation time (45 months). The early appearance of signs of the disease in countries south of the Sahara may be explained by the significant and aggressive sun exposure in this pediatric environment [16,17]. The clinical presentations of XP are in its classic form with seven complementation groups (from XPA to XPG) and in the so-called variant form (XPV) [1]. Without the possibility of conducting biological studies for the distinction of these forms, we resorted to clinical classification [18,19]. Based on the absence of neurological signs [1-2,17] in our patients, we classified eleven cases with the classic form of the XPC group and the twelfth case with the variant form. In classic XP, photophobia and xeroderma, the usual and early signs, were constant in our patients and in those reported by some authors [9,10]; meanwhile, they are late in the variant form [12]. The lingual involvement with amputation that we noted was also reported in some series [8,9]. The ocular and palpebral manifestations, such as hemorrhagic conjunctivitis, ulcerations, and corneal cavities or tumors observed in the classic form of XP as well as in the variant form, were regular in our study and were also reported by some authors [1,8,12]. We noted no cases of leukemia, contrary to the literature [2,10,14]. Basal cell carcinoma and squamous cell carcinoma were observed equally in six cases each. The latter were the most frequently reported [1,6,17] and highly often

as in our series without melanoma [9,10]. Almost all of our patients received photoprotection and an excisional biopsy as treatment because the extent and number of the tumor lesions were contraindications to surgical removal. Chidzonga et al. [9], as well as Bouadjar et al. [12], opted for surgical excision in the event of a tumor in patients seen early. Patients with XP usually die especially young. Seven cases of death before the age of eight were noted following infectious complications, hemorrhage, malnutrition, and the multiplication of carcinomatous tumors. Chidzonga et al., in their study [9], noted that the patient who survived the longest died at the age of eighteen. At the time of this study, we have two patients lost to follow-up and three alive, including the forty-year-old presenting the XP variant. Would photoprotection prescribed early in these patients have had a positive impact on the prevention of skin tumors in a country in which sunshine is present all year round? It is difficult to say, given that patients are received at the stage of tumor lesions. However, in those living at the time of this study, we see the beneficial effect of photoprotection by slowing down the birth of new tumor lesions. Thus, monitoring live cases will allow us to better appreciate the beneficial effect of long-term photoprotection despite its significant cost.

## CONCLUSION

Xeroderma pigmentosum is a rare yet serious genetic disorder in Niger. While photoprotection is currently the mainstay of treatment, genetic therapy is raising hope for affected families. We must concentrate our efforts on genetic counseling and antenatal diagnosis in families at risk and dermatological follow-up of cases, given the early onset of skin cancers in Niger.

### Statement of Human and Animal Rights

All the procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the 2008 revision of the Declaration of Helsinki of 1975.

### Statement of Informed Consent

Informed consent for participation in this study was obtained from all patients.

## REFERENCES


1. Kraemer KH, Lee MM, Scotto J. Xeroderma Pigmentosum: Cutaneous, ocular, and neurologic abnormalities in 830 published cases. Arch Dermatol. 1987;123:241-50.
2. El Fékih N, Fredji M, Aounallah-Skhiri H, Fazaa B, Zghal M, Ridha Kamoun M. Neurological damage in xeroderma pigmentosum. Rev Neurological. 2009;165:967-70.
3. Zghal M, Fazaa B, Abdelhak S, Mokni M. Xeroderma pigmentosum. Ann Dermatol Venereol. 2018;145:706-22.
4. Moussala M, Behar-Cohen F, D'hermies F, Bisseck A-C, Renard G. Xeroderma pigmentosum and its ocular manifestations. J Fr Ophthalmol. 2000;23:369-74.
5. Christen-Zazech S, Imoto K, Khan S G, Tamura D, DiGiovanna J J, Boyle J, et al. Unexpected occurrence of xeroderma pigmentosum in an uncle and nephew. Arch Dermatol. 2009;145:1285-91.
6. Witold K J. Xeroderma Pigmentosum in black South Africans. Intern Jr Dermatol. 1999;38:511-4.
7. Hebert J.C, Lefait vJ-F, Hebert O. Xeroderma Pigmentosum in black-skinned children: Five observations in Mahorais children. Ann Dermatol Venereol. 1994;121:382-6.
8. Gul U, Kihç A, Gonul M, 9akmak SK, Soylu S. Xeroderma pigmentosum: A Turkish case series. Intern J Dermatol. 2007;46:1125-8.
9. Chidzonga MM, Mahomva L, Makunike-Mutasa R, Masanganise R. Xeroderma pigmentosum: A retrospective case series in Zimbabwe. J Oral Maxillo Fac Surg. 2009;67:22-31.
10. Khatri M L, Bemghazil M, Shafi M, Machina A. Xeroderma pigmentosum in Libya. Intern J Dermatol. 1999;38:520-4.
11. Fazaa B, Zghal M, Zeglaoui F, Goucha S, Mokhtar I, Kharfi M, et al. Melanoma and xeroderma pigmentosum: 12 cases. Ann Dermatol. Venereol. 2001;128:503-6
12. Bouadjar B, Belkacem FA, Daya-Grosjean L, Larbaoui LS, Ferhat R, Cherid MC et al. Xeroderma pigmentosum: Study of 40 Algerian patients. Ann Dermatol Venereol. 1996;123:305-6.
13. Dieng MT, Niang SO, Dangou JM, Ndiaye B, Xeroderma pigmentosum: Report of 6cases in Dakar, Senegal. Bull Cancer. 2001;88:199-202.
14. Zghal M, Fazaa B, Zghal A, Mokhtar I, Sarasin A, Kamoun MR, et al. Xeroderma pigmentosum: Clinical and genetic particularities of a family in which all members are affected. Ann Dermatol. Venereol. 2003;130:31-6.
15. Rubaie SMA, El Darouti MA. Clinical and genetic study of xeroderma pigmentosum. Intern J Dermatol. 1990;29:126-8.
16. Salissou L, Moumouni H, Sani R, Nouhou H. Xeroderma pigmentosum: First observation in Niger. Ann Univsers A M Niamey. 2013;XV-A:1-8.
17. Salissou L, Doulla M, Harouna MZ, Salha I, Timmi N, Dan Sono AA, et al. Xeroderma pigmentosum: Squamous cell carcinoma infiltrating and disfiguring facial, in a girl of 3 years and a half. Our Dermatol Online. 2017; 8(Suppl. 1):28-31.
18. Ahmed H, Hassan R Y, Pindiga U. Xeroderma in three consecutive siblings of a Nigerian family: Observations of oculocutaneous manifestations in black African children. Br J Ophthalmo. 2001;85:110-1.
19. Zghal M, Fazaa B, Kamoun MR. Xeroderma pigmentosum. EMC (Elsevier SAS, Paris), Dermatology. 206;98-660-A-10.




**Source of Support:** Nil, **Conflict of Interest:** None declared.