Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Dermatology

Xeroderma Pigmentosum (XP) Type A and its Cutaneous, Ophthalmological, and Neurological Manifestations: Report of Two Cases Diagnosed at the Dermatology Hospital of Bamako, Mali

Dembélé R[1]*, Koné A[2], Dao K[3], Doumbia AA[2], Berthé BB[4], Guindo B[1], Kané M[5], Faye O[1,6]

[1]Dermatology Hospital of Bamako, Mali
[2]Department of Medicine and Endocrinology, Hôpital du Mali, Bamako, Mali
[3]Department of Internal Medicine, Gabriel Touré University Hospital, Bamako, Mali
[4]Bamako Armed Forces Medical and Surgical Center, Mali
[5]Mohamed VI Perinatal Clinic, Bamako, Mali
[6]Faculty of Medicine and Odontostomatology, USTTB, Bamako, Mali

DOI: https://doi.org/10.36347/sjmcr.2026.v14i11.013 | **Received:** 09.09.2026 | **Accepted:** 31.10.2026 | **Published:** 04.11.2026

***Corresponding author:** Dr. Dembélé Ramatou
Dermatology Hospital of Bamako, Mali

Abstract | **Case Report**

Background: Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disorder characterized by extreme sensitivity to ultraviolet (UV) radiation, progressive cutaneous pigmentation abnormalities, and an increased risk of skin malignancies. Ocular and neurological manifestations may also occur, depending on the genetic subtype. XP is particularly associated with consanguinity and remains rarely documented in sub-Saharan Africa. ***Objective:*** To describe the clinical characteristics, cutaneous manifestations, ophthalmological abnormalities, neurological involvement, and management of two familial cases of xeroderma pigmentosum evaluated at the Dermatology Hospital of Bamako, Mali. ***Case Presentations:*** We report two cases involving siblings aged 8 and 18 years, born to consanguineous parents, who presented with characteristic manifestations of XP. The first patient, an 8-year-old girl, exhibited multiple hyperpigmented macules distributed over sun-exposed areas and an ulcerative proliferative lesion involving the nasal ala. Ophthalmological and neurological examinations were normal. Genetic testing confirmed xeroderma pigmentosum type A (XPA). Treatment included 5% urea-containing petroleum jelly, photoprotective cream, and protective clothing recommendations, resulting in marked improvement in cutaneous lesions after several weeks. The second patient, an 18-year-old male sibling, presented with extensive lentigines, hypopigmented macules, poikilodermatous changes, cutaneous atrophy, and supernumerary teeth. Ophthalmological examination revealed corneal dystrophy, while neurological assessment demonstrated an ataxic gait, positive Romberg sign, apraxia, upper-limb hypertonia, absent deep tendon reflexes, impaired proprioception, and growth retardation. A clinical diagnosis of XP was established, and genetic investigations were ongoing. Management included topical emollients, photoprotection, retinoid therapy, and lifestyle modifications. ***Conclusion:*** Xeroderma pigmentosum is a rare hereditary genodermatosis associated with significant dermatological, ophthalmological, and neurological complications. These familial cases highlight the importance of early clinical recognition, genetic testing, rigorous lifelong photoprotection, genetic counseling, and multidisciplinary management. In highly sun-exposed regions such as Mali, preventive measures are particularly important for reducing the risk of cutaneous malignancies and improving patients' quality of life.

Keywords: Xeroderma pigmentosum; Carcinoma; Genetic counseling; Dermatology Hospital of Bamako.

Copyright © 2026 The Author(s): This is an open-access article distributed under the terms of the Creative Commons Attribution **4.0 International License (CC BY-NC 4.0)** which permits unrestricted use, distribution, and reproduction in any medium for non-commercial use provided the original author and source are credited.

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare genetic disorder with autosomal recessive inheritance, clinically characterized by extreme cutaneous photosensitivity, pigmentary abnormalities of sun-exposed skin, an increased risk of developing skin cancers, and ophthalmological and neurological involvement [1].

The condition was first described in 1870 by M. Kaposi [2]. XP is associated with defects in enzymes involved in repairing the oncogenic damage caused by ultraviolet exposure [3–5]. The condition affects all

ethnic and racial groups worldwide. Its prevalence is relatively higher in the Middle East and Japan [6].

In French-speaking sub-Saharan Africa, we found only one case reported in Senegal [7] and three cases reported in northern Mali in 2019 by Conaré I. *et al.,*[8]. The patients described here, one aged 18 years and the other 8 years, had normal skin at birth. At approximately one month of age, both developed typical, highly characteristic skin lesions, allowing the diagnosis to be readily established without additional investigations. The age at onset and severity of symptoms vary according to the gene involved, the XP subtype, and environmental factors.

In this report, we describe two familial cases of XP in Malian patients, incidentally identified during consultation on the basis of their cutaneous manifestations.

Case 1

AS, an 8-year-old girl, was brought by her parents to the Dermatology Department in Bamako because of skin spots.

History taking revealed first-degree consanguinity (the parents are cousins). She was the ninth of 10 children, of whom the sixth was also affected by XP. The pregnancies had been medically monitored, and delivery took place at a health center.

XP had never previously been diagnosed. Examination of the skin revealed numerous small hyperpigmented macules scattered over uncovered areas exposed to ultraviolet radiation, together with a proliferative ulceration of the nasal ala.

The ophthalmological examination was normal. The neurological examination was also normal.



Figure 1: Hyperpigmented lesions of the face and exposed areas, with ulceration of the nasal ala.

Based on these findings, the diagnosis of xeroderma pigmentosum type A was confirmed by genetic testing. Treatment consisted of petroleum jelly containing 5% urea, a photoprotective cream, and advice on protective clothing. After several weeks of treatment, the skin lesions showed marked improvement.

Case 2

MS, an 18-year-old male and the brother of AS, was brought by his parents for dermatological consultation because of skin spots and psychomotor impairment. History revealed that he was the sixth of 10 children and that the parents were first-degree cousins. Cutaneous and mucosal examination revealed numerous dark lentigines and hypopigmented macules on sun-exposed areas (face, neck, and upper limbs), extending to areas covered by clothing and producing a poikilodermatous appearance. These findings were associated with atrophic lesions of the neck and shoulder and supernumerary teeth.



Figure 2: Lentigines associated with atrophic lesions of the neck and shoulders

Ophthalmological examination revealed corneal dystrophy.

Neurological examination revealed an unsteady, ataxic gait, a positive Romberg sign, apraxia, hypertonia of the upper limbs, absent deep tendon reflexes, impaired proprioception, and growth retardation.

On the basis of these findings, we diagnosed XP. Genetic testing had been requested and was ongoing. Treatment included petroleum jelly containing 5% urea, a photoprotective cream, a retinoid, and advice on hygiene and protective clothing.

DISCUSSION

XP is a worldwide disorder described in all populations, with geographic distributions that differ by subtype. Its prevalence is estimated at 1 in 20,000 in Japan, where XPA predominates [4]; 1 in 250,000 in the United States; and 2.3 per million in Europe. It is relatively common in regions with high rates of consanguinity and large families, such as the Middle East and the Maghreb (estimated incidence of 1 in 10,000 in Tunisia). Six cases were described in a South African Bantu royal family. In Cameroon, a single case of XP was identified in a 9-year-old boy attending the Dermatology Department of Yaoundé Central Hospital. Our observations constitute the first descriptions of cutaneous manifestations of XP in Mali. In addition to a suggestive family history, the diagnosis of XP is relatively straightforward. Given the characteristic clinical presentations, we do not consider the absence of genetic testing to be an argument against diagnosing XP in our patients. XP follows an autosomal recessive pattern of inheritance. Consanguinity and parental kinship play an essential role in the transmission of cases reported among Black African populations [9]. Consanguinity is reported in 31% of patients with XP, while 21% have parents who are first cousins. Our patients' parents are first cousins, which explains the occurrence of the disease in these cases. Ultraviolet A radiation (320–400 nm) induces the formation of free radicals that damage DNA bases. These changes lead to DNA strand breaks and generate DNA–protein crosslinks [10]. Pyrimidine dimers and pyrimidine (6–4)

pyrimidone photoproducts are thought to arise from the absorption of UVB radiation by adjacent pyrimidines. This distorts the DNA double helix and interferes with its normal functioning (blocking transcription of active genes and DNA replication, and promoting translesion synthesis leading to mutations or chromosomal rearrangements) [11]. These molecular abnormalities (primarily crosslinks at pyrimidine pairs) are normally recognized and repaired by a multienzyme complex, of which the excision–resynthesis repair system is the most effective and best understood; deficiencies in these repair systems cause XP. According to Stary A, "the presence of unrepaired lesions can, during the replication of damaged DNA, cause the insertion of abnormal bases opposite the lesion and thus lead to errors (mutations) in the genome of the daughter cell" [12]. This results in cell death or the development of somatic mutations that confer oncogenic properties on affected cells through irreversible alteration of the regulation of cell proliferation. Genetically, seven complementation groups are recognized, designated XPA through XPG, together with a more recently identified group known as XPV (variant). The severity of cutaneous involvement and the occurrence of neurological impairment therefore vary among XP groups.

Cutaneous manifestations progress through three stages:

In the first stage, persistent erythema appears on exposed areas following even minimal initial sun exposure; its intensity correlates with disease severity and the duration of exposure. It is accompanied by photophobia and xerosis with cheilitis.

In the second stage, pigmentary disturbances gradually develop, including lenticular pigmented macules, hypopigmented macules, skin atrophy, and telangiectasias.

In the third stage, cutaneous and mucosal tumors appear at an early age, as an apparently inevitable progression. This is responsible for the severity of the disease. Lesions may be benign precancerous tumors (actinic keratoses), but are often malignant tumors, mainly basal cell carcinomas (Case 1) and squamous cell carcinomas, with a risk reported to be 10,000 times greater than that of the general population. XP is relatively easy to diagnose, especially at an advanced stage, primarily on clinical grounds (Case 2).

DNA repair tests following UV exposure, or unscheduled DNA synthesis (UDS) testing, can confirm the diagnosis. These techniques are now being replaced by molecular genetic methods that detect the mutations responsible for the disease.

Treatment is primarily preventive, since there is currently no curative therapy capable of altering the course of XP.

Prevention focuses on photoprotection, which should be introduced as early as possible. It can improve cutaneous and ophthalmological prognosis and delay the onset of malignant tumors, but cannot prevent neurological degeneration. Sun exposure should be avoided between 9:00 a.m. and 6:00 p.m.; especially in summer, a predominantly nocturnal lifestyle may be necessary. Wearing a hat, appropriately filtered glasses, and protective clothing is essential. Regular application of high-SPF sunscreen is indispensable when sun exposure cannot be avoided. Such rigorous photoprotection should be accompanied by a vitamin D-rich diet [15].

Genetic counseling aims to discourage consanguineous marriages and limit the number of births in families at risk.

The use of retinoid compounds prevents tumor progression.

Surgical removal of tumors and autografting of skin from a sun-protected area of the patient are the preferred therapeutic measures for treating established lesions.

CONCLUSION

Xeroderma pigmentosum is a rare genodermatosis. The intensely sunny environment of Mali is an important contributing factor to the development of skin cancers. Photoprotection is the only measure that reduces the occurrence of skin cancers in affected patients. Genetic counseling is necessary, particularly when there is a family history of XP. At present, no curative treatment exists. Symptomatic management requires a multidisciplinary approach.

REFERENCES

1. Stary A, Sarazin A. Xeroderma pigmentosum. Presse Med. 1997;26:1992–1997.
2. Hebra S, Kaposi M. On Diseases of the Skin Including Exanthemata. New Sydenham Soc. 1874;61:252–258.
3. Sarazin A, Renault G, Blanchet-Bardon C, Boue J, Dumez Y. Xeroderma pigmentosum: Clinical and cellular characteristics; development of a prenatal test. Médecine/Sciences. 1988;4:608–617.
4. Hirai Y, Kodama Y, Moriwaki S-I, Noda A, Cullings HM, Macphee DG, *et al.,* Heterozygous individuals bearing a founder mutation in the XPA DNA repair gene comprise nearly. [Incomplete citation in source.]
5. Copeland NE, Hanke CW, Michalac JA. The molecular basis of xeroderma pigmentosum. Dermatol Surg. 1997;23:447–455.
6. Kraemer KH, Lee MM, Scotto J. Xeroderma pigmentosum: Cutaneous, ocular, and neurologic abnormalities in 830 published cases. Arch Dermatol. 1987;123:241–250.

7. N'Diaye B, Ball MD, Strobel M, Niang I. Xeroderma pigmentosum: First Senegalese case report. Dakar Medical. 1983;28:167–172.
8. Conaré I, *et al.,* Xeroderma pigmentosum revealed by its ocular manifestations in northern Mali: Report of three cases. Revue SOAO. 2019;(02):53–55.
9. N'Diaye B, Ball MD, Strobel M, Niang I. Xeroderma pigmentosum: First Senegalese case report. Dakar Médical. 1983;28:167–172.
10. Copeland NE, Hanke CW, Michalak JA. The molecular basis of xeroderma pigmentosum. Dermatol Surg. 1997;23:447–455.
11. Sarasin A. Human DNA repair genes. Médecine/Science. 1994;10:43–54.
12. Stary A, Sarasin A. Xeroderma pigmentosum. Presse Med. 1997;26:1992–1998.