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Original Article

The burden of xeroderma pigmentosum in two families followed at the Department of Dermatology and Venereology of the National Hospital in Niamey, Niger

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ABSTRACT

Background: Xeroderma pigmentosum (XP) is a highly complex autosomal recessive disease linked to an enzymatic DNA repair disorder. Herein, we report the clinical and evolutionary aspects of XP in Niger. **Materials and Methods:** Our study included patients diagnosed with XP from two families. **Results:** We collected eight patients with an average age of 5.5 years, with extremes of two and thirteen years. The sex ratio was 1. Consanguinity was found in both families. The first tumor appeared between three and five years of age in six cases and around eight years of age in two cases. The tumors were cutaneous in seven cases, and extra-cutaneous in five cases. Histology made it possible to identify basal cell and squamous cell carcinomas without any case of melanoma. Five out of eight died between the age of eight and twelve years. **Conclusion:** Prevention through the reduction of new cases by genetic counseling and antenatal diagnosis in families at risk is necessary.

Key words: XP; Basal cell carcinoma; Squamous cell carcinomas; Early death; Niamey, Niger

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare autosomal recessive disease characterized by pathological sensitivity to UV rays [1,2]. The damage to the DNA caused by UV exposure creates distortions in the DNA helix and disrupts transcription mechanisms. Without total and effective protection against the sun, patients experience accelerated skin aging, burns, pigmentation disorders and the inevitable development of cutaneous and extra-cutaneous lesions that may lead to multiple cancers. The clinical presentation of XP is in its classic form with nine complementation groups (from A to I) and in the so-called variant form [1,3,4]. The incidence of the disease is around 1/250,000 births in the U.S. and 1/20,000 in Japan [3,5]. The aim of this study was to determine the sociodemographic, clinical, histopathological, and evolutionary aspects of XP in two different families in Niger.

MATERIALS AND METHODS

This was a cross-sectional, descriptive study conducted from January 1, 2007, to December 31, 2019 (thirteen

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years), at the Department of Dermatology and Venereology of HNN. The affected patients came from two families whose diagnosis of XP was reached on the basis of clinical arguments. We employed a pre-established survey sheet that included the following variables: sociodemographic (age, sex, origin, mode of admission), clinical (history of the disease, functional signs, general signs, physical signs), paraclinical, in particular, histopathology and progression (age of the onset of the first tumors, complications, age of death).

RESULTS

Eight patients were collected, including four per family. The average age of the patients was 5.5 years, with extremes of two and thirteen years. The sex ratio (male-to-female) was 1. The socioeconomic conditions of both families were low level. The tracing of the family tree allowed us to note the consanguinity in the two families (Figs. 1a and 1b).

The onset of classic signs, such as fixed erythema, xeroderma, and photophobia, was reported by the parents in the first twenty-four months in all patients (Fig. 2a, V4). Compared to the non-consanguineous sister whose skin was normal (Fig. 2b, V5), the two patients descended from family II. It should be noted that, in both families, the V10 of family I and the V4 of family II were born after the remarriage of the parents, who were divorced for several years. The age of onset of the tumors was between three and five years in five children, at seven years in two children, and at twelve years in one case. The consultation period was greater than or equal to 48 months in five cases and less than 48 months in three. During the physical examination, we noted, in the eight patients, photophobia, dyschromic macules, xeroderma, freckles, and tumor lesions. These tumors were located particularly on the scalp, face, oral mucosa (Figs. 2c and 2d), and the tongue (Figs. 3a and 3b). Four patients had less than three tumors and three more than three tumors. All eight patients had ophthalmological involvement characterized by an eyelid tumor, hemorrhagic conjunctivitis, and corneal sheath. One patient also presented with corneal ulceration. No patient had neurological impairment, such as hyporeflexia, during our study. Skin biopsy and histological examination were performed in six patients. This histological examination revealed two cases of associated basal cell carcinoma (BCC) (Fig. 4a) + squamous cell carcinoma (SCC) and four cases of SCC (Fig. 4b). None case of melanoma was noted. Most of the treatments performed were symptomatic based on photoprotection. Chemotherapy could not be done for lack of means. None of the patients received

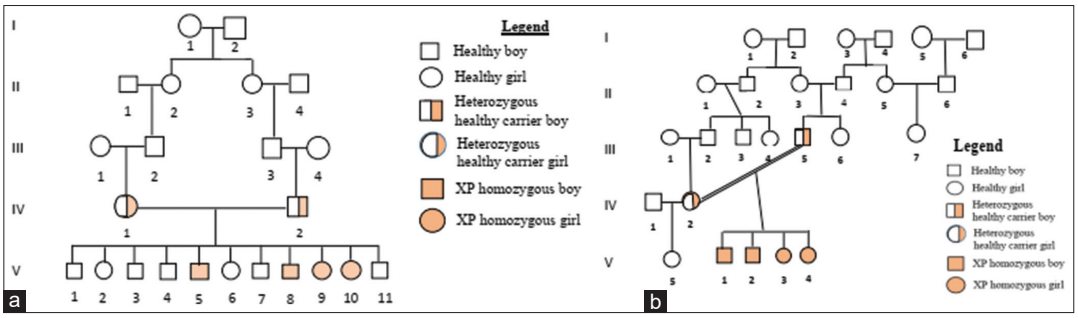

Figure 1: (a) Family tree I. (b) Family tree II.

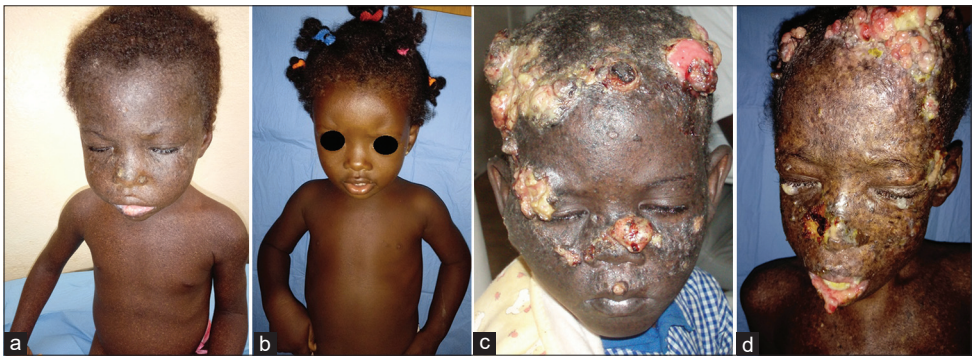

Figure 2: (a) Child V4, poikiloderma and cheilitis (family II). (b) Healthy V5 child from a non-consanguineous marriage (family II). (c) Budding cauliflower tumors of the scalp and face of child V8 (family I). d) Budding ulcerative lesions of the scalp face and lower lip in the third child (family II).

surgical treatment. The evolution was made toward the appearance of mutilating cancerous lesions, which led to the death of the eight patients practically between the ages of eight and thirteen years.

The causes of death were most often infectious complications of tumor lesions in three patients, hemorrhagic complications (Fig. 5a, V8 of family I) in three patients, and the deterioration of the general condition in two (Fig. 5b, V2 of family II) (Table 1).

DISCUSSION

The transmission of XP most often occurring in the autosomal recessive mode explains its relative frequency in countries in which inbreeding is high and families are large [4,6,7]. In our study, consanguinity was found in both families, for which divorce counseling was a failure; this resulted in the birth of the fourth case in family II (fourth of the V4 sibling) and the tenth child (V10) of the sibling in family I. The average age of our patients (5.5 years) was lesser when compared to data in the literature (10 years) [8], and without a female predominance, unlike series from Zimbabwe [9] and Tunisia [10], In other series, the predominance was male [8]. Similarly to another study [11], the socio-economic level of the families of our patients was especially low. The ulcero-budding tumor was the most common reason for consultation, in seven out of the eight cases in our series, whereas it was observed later elsewhere [1]. The first signs observed were between the age of six months and twenty-four months, as in some sources in which the age of onset was between three months and twenty-six months [12,13]. Several studies [2,14] reported the appearance of tumors between the age of two years and eight years, which was proportional in our series, in which the tumors appeared between three and five years of age. The time to consultation was more than forty-eight months in five cases and was earlier than in the literature, in which patients consulted later [1]. In our study, all patients presented, as reported by several authors, with poikilodermic-like [15], lingual [6], and oculo-palpebral [1,6,16,17] aspects. Some authors [8,13,16,18] reported neurological damage, unlike our cases, in which not all patients of the two families presented the damage. Based on the absence of neurological signs [3], all our patients were classified as cases of the classic form of the XPC group. Histopathologically, we noted four patients

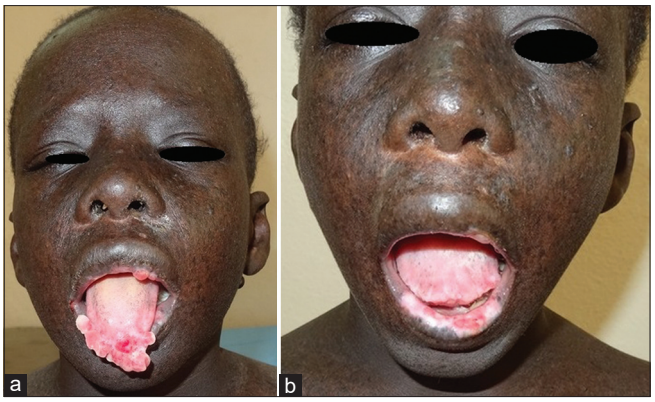

Figure 3: (a-b) Tumor involvement of the tip of the tongue. (a) Spontaneous partial amputation of the tongue, (b) of the first sibling V1 (family II).

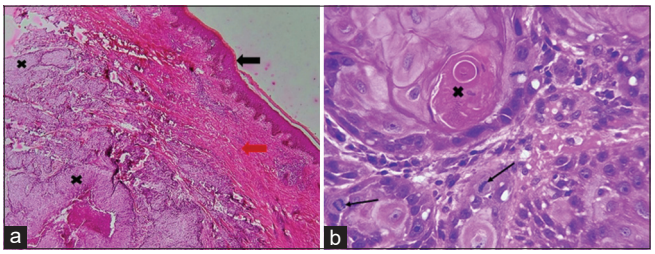

Figure 4: (a) Basal cell carcinoma. Proliferation of lobular architecture with a palisade-like peripheral arrangement and made of basaloid cells (cross) in a fibrous stroma (red arrow) (HE/GX 40). (b) Squamous cell carcinoma. The centered keratin lobules (cross) and the nuclei are highly atypical with numerous mitoses (arrows) (HE/GX 400).

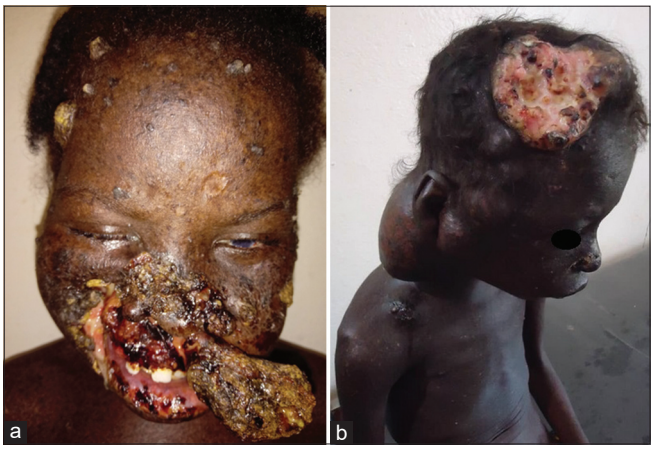

Figure 5: (a) Ulcerative vegetative tumor, hemorrhagic and infected, mutilating the nose and the right cheek, children V10 (family I). (Department of Dermatology and Venereology). (b) Multiple tumors (scalp and neck) in a poor general condition.

with squamous cell carcinoma, two cases of the association of squamous cell carcinoma and basal cell carcinoma, and no cases of melanoma. Some studies [19,20] reported six cases of XP, among which five presented squamous cell carcinoma, two presented the association of squamous cell carcinoma and basal

Table 1: The age of onset of the tumors and the vital prognosis of the patients.

Family	Case	Age at first consultation	Age at onset of tumor lesions	Vital prognostic (yrs.)			
				A5	A8	A12	A13
Family I	1st case V5	5 years	5 years	living	death		
	2nd case V8	5 years	5 years	living	death	death	death
	3rd case V9	3 years	3 years	living	living	living	
	4th case V10	13 years	12 years	living	living		
Family II	1st case V1	5 years	7 years	living	living	death	
	2nd case V2	2 years	7 years	living	living	death	
	3rd case V3	3 years	3 years	living	death		
	4th case V4	2 years	3 years	living	death		

cell carcinoma, and no case presented with melanoma. Squamous cell carcinoma has seemed to be the most encountered [3,19,20]. In our series, six out of the eight patients died of infectious and hemorrhagic complications and two of malnutrition. However, in one study [7], the causes of death were infections in five cases out of twenty-five. Death occurred between 8 and 13 years. Some literature [7] reported cases of death with an average age of fourteen years and extremes of eight and sixteen years, and even a death at the age of eighteen years [5]. This study made it possible to highlight the risks of inbreeding, which is frequent especially in West Africa, the consequences of which are at the origin of serious diseases, in particular, xeroderma pigmentosum. The evolution of the latter has always been precocious and fatal as it constantly leads to death at a young age [3].

CONCLUSION

XP is a complex pathology, yet its management is even more so. The sure way to avoid the disease is to suspend consanguineous marriages through the sensitization and education of the populations. Failing this, one must employ a method of contraception most suitable for couples and adopt children as soon as a case occurs in the offspring and the desire to have children remains (NB families most often poor). Prevention through the reduction of new cases by genetic counseling and antenatal diagnosis in families at risk is necessary.

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.

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