Xeroderma Pigmentosum: A Retrospective Case Series in Zimbabwe

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Purpose: To present our experience with the clinical features and management of black African patients with xeroderma pigmentosum (XP).

Patients and Methods: Twelve patients with XP were seen over a 25-year period, and were retrospectively reviewed for age, gender, clinical features, treatment, and follow-up.

Results: There were 8 females and 4 males with an age range of 3 to 18 years. One patient, the longest survivor, was followed until death at 18 years. Nine patients had the severe form of XP and 3 had the mild form. All patients had early ocular involvement with photophobia and early blindness. Squamous cell carcinoma (SCC) was present on the skin, lip, and tongue in most patients. One patient had ocular surface SCC. There was marked skin photosensitivity. No history of consanguinity was noted in the parents of the patients. Surgery was the treatment modality of choice. Follow-up was poor.

Conclusion: XP is uncommon in our black population, and presents in the severe form with SCC as the malignant skin, lip, and tongue lesion. It is common in early childhood with severe photosensitivity, photophobia, and eventual blindness. Follow-up is difficult in our environment.

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Xeroderma pigmentosum (XP) is a rare autosomal recessively transmitted genetic disease characterized by clinical and cellular sensitivity to ultraviolet (UV) radiation.[1-5] Patients with this disease show hypersensitivity to sunlight and extreme susceptibility to sunlight-induced cutaneous cancers.[1-5] XP also is associated with microcephaly and mental retardation. XP is classified into 8 genetic complementation sub groups, XP-A to XP-G and a variant group XP-V.[3] There are different gene alterations in each of the groups. Seven of these groups (XP-A to XP-G) are involved in deoxyribonucleic acid (DNA) excision repair with the variant XP-V involved in replication of damaged DNA on the leading strand.[3]

The defect in one of the genetically distinct nucleotide excision repair genes (complementation groups A to G) confers increased susceptibility to UV-induced killing and mutation with degeneration and a propensity for the early development of cutaneous malignancies in sun-exposed areas.[3,6] Many enzymatic defects are also observed in these patients.[3] This provides an opportunity for mutant malignant growth resulting in all forms of cutaneous malignancies in the sun exposed areas. These are mainly squamous cell carcinoma (SCC), basal cell carcinoma (BCC), fibrosarcoma (FS), malignant melanoma (MM), keratoacanthomas, angiomas, and sarcomas.[6-8] Death tends to occur in early adulthood due to these cutaneous and ocular malignancies and metastatic disease.[9]

XP is characterized by clinical and cellular hypersensitivity to UV radiation manifesting as intolerance of skin and eyes to light. The skin lesions are comprised of freckles on limbs and face with a dry skin covered with a mixture of mottled, hypopigmented and hyperpigmented, atrophic roundish and oval macules, giving the entire skin a checkered appearance associated with generalized actinic keratoses, manifesting on black skin as palpable, rough, blackish spots covered with adherent scales.[4] These skin le-

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sions cover both sun-exposed and covered areas. The skin later develops cutaneous malignancies. Ocular changes include photophobia, ocular pigmentary changes, conjunctivitis, corneal keratitis, ulcers, blindness, and malignancies.

XP has been described in all races but is exceedingly rare in the Negroid race, although some cases have been reported in both the black American and black African people.[3,4,10] Children are mostly affected in early childhood with a female preponderance although some reports show equal male and female affliction.[3,4,10] It is most common in children of consanguineous parents.[7] Frequency of XP has been reported from a low of 1:250,000 in the United States[11] to a high of 1:400,000 in Japan.[12] High frequencies have also been reported in Morocco, Egypt, and Libya, presumably related to genetic factors and consanguinity in parents of the patients.[10,13,14]

The clinical manifestations occur primarily on the sun-exposed areas of the skin and eyes.

The skin is normal at birth with the first cutaneous symptom being acute photosensitivity in early infancy followed by pigmentation, freckling, atrophy, telangiectasia, and later skin tumors. Clinically it can also be classified into 3 subgroups depending on the clinical presentation of the patients. These include: *1*) mild; those with light brown freckles on the face alone; *2*) moderate; dark brown freckles with burning on the face, neck, ears, chest, hands, and photophobia but without other associated cutaneous and ocular changes; and *3*) severe, extensive, dark brown freckles all over the body with cutaneous changes such as ulcers and skin malignancy.[3]

Treatment modalities for XP include isotretinoin prophylaxis, avoidance of light exposure, surgical excision of premalignant and malignant tumors, resurfacing with skin grafts, dermabrasion, radiation therapy, and Mohs micrographic surgery.[10,15]

This retrospective case series studies 12 patients with XP seen in Zimbabwe over a 25-year period. It presents their clinical features, surgical management, and follow-up. These features are discussed and compared with those of XP patients in other reported case series.

Report of Cases

CASE 1

Patient 1, a 3-year-old girl, presented in February 1982 with severe XP, severe skin photosensitivity, and severe photophobia. She had multiple facial tumors with an average diameter of 1.5 cm. Excision biopsies confirmed the tumors to be SCC. She was lost to follow-up after 6 months without any recurrence or new tumors developing.

CASE 2

Patient 2, a 9-year-old boy, presented in May 1983 with severe XP, severe skin photosensitivity, and severe photophobia. He presented with a left cheek tumor 4 cm in diameter. Excision biopsy confirmed the tumor to be SCC. He was lost to follow-up after 1 year without recurrence or new tumors developing.

CASE 3

Patient 3, a 9-year-old girl, presented in September 1984 with severe XP, severe skin photosensitivity, and blindness (Fig 1). She had the following tumors: *1*) tip of tongue (2 cm in diameter); *2*) left upper lip (93 cm in diameter); and *3*) left cheek (4 cm in diameter). Excision biopsy confirmed all the tumors to be SCC. She was lost to follow-up after 18 months without recurrence or new tumor developing.

In December 1985, now 10 years old, she reappeared with a 3.5 cm in diameter tumor of 6 months duration attached to the right alae nasi. Excision biopsy confirmed SCC. She was again lost to follow-up after 6 months without recurrence or new tumor developing.

CASE 4

In June 1987, patient 4, a 3-year-old girl, presented with severe XP and an exophytic 2 cm in diameter fleshy tumor of 6 months duration on the mid-dorsum of the tongue. She had mild photophobia and severe skin photosensitivity. Her parents recalled that some pigmentation was present at birth but was less intense and sparsely distributed. However, over the years the pigmentation had been increasing in intensity and generalized spread. Her 2 female siblings were not affected. She had normal vision and mild photophobia. Her medical examination was otherwise noncontributory. A wide excision biopsy confirmed the tumor as SCC.

In December 1987, she presented with a tumor 1 cm in diameter over the tip of the tongue. Wide excision biopsy confirmed the tumor as SCC. In both excision biopsies the specimen margins were reported to be clear of tumor.

In October 1989, now 5 years old, she presented with a right cheek tumor 1.5 cm in diameter along with a right upper lip tumor 0.5 cm in diameter. Excision biopsy confirmed both tumors as SCC.

In September 2000, now 16 years old (Fig 2), she presented with a left cheek tumor 1.5 cm in diameter and a nodular forehead tumor 3 cm in diameter, both of 8 months duration. Excision biopsies confirmed both tumors as SCC. She presented 3 months later, December 2000, with palpable right submandibular lymph nodes. Right submandibular dissection was done and confirmed metastatic SCC in the lymph nodes.

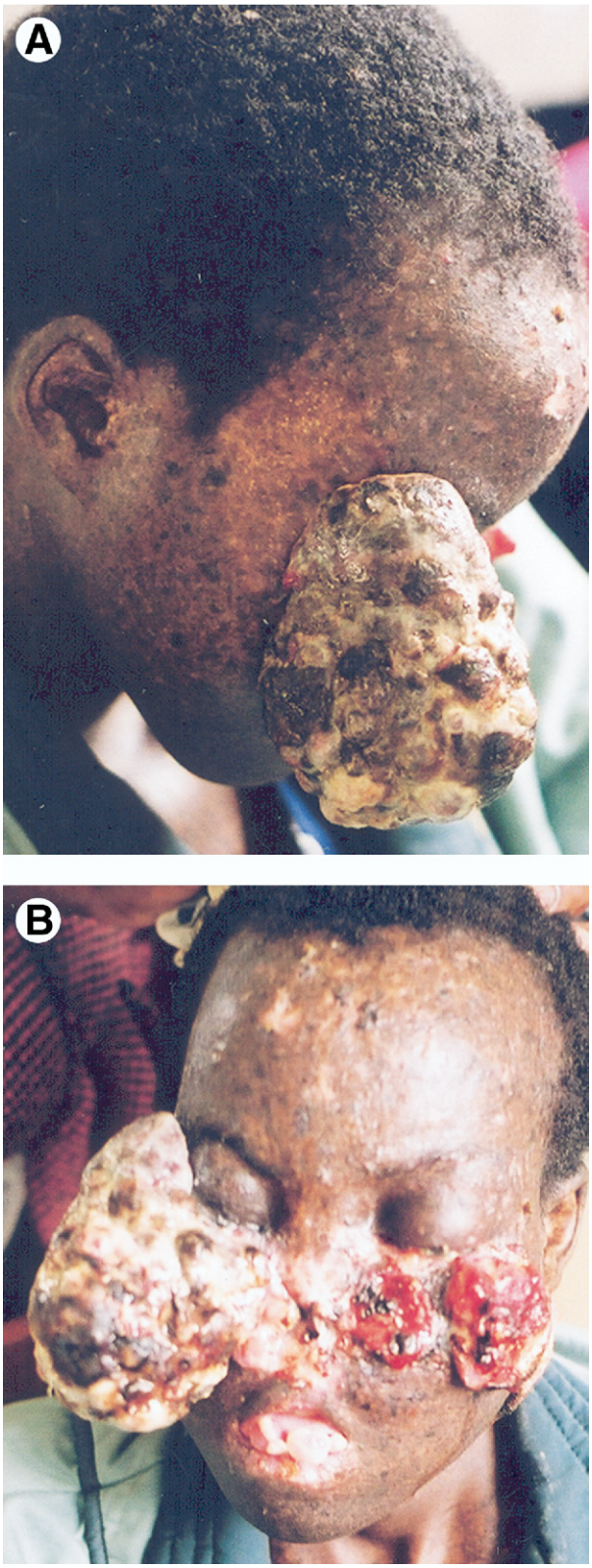

FIGURE 1. Case 3, patient 3. A 9-year-old girl. Florid cutaneous SCC. Presented 4 years previously with tongue and lip SCC.

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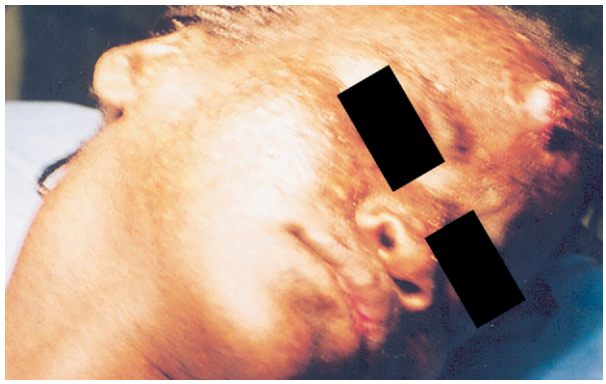

FIGURE 2. Case 4, patient 4. Girl is now 13 years old, recurrent forehead SCC with neck nodes metastasis.

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In August 2000, now 16 years old, she presented with a fungating forehead tumor 6 × 5 cm, where a tumor had been excised 18 months previously (Fig 3). Histopathologic examination showed recurrent SCC with some areas of BCC and intradermal nevus. Wide surgical excision was done with closure by advancement of local tissue flaps.

In October 2000, she presented with a recurrent fungating forehead tumor now 3.5 × 2 cm. Tumor excision was done along with resection of the outer cortical bone. Radiation therapy was withheld be-

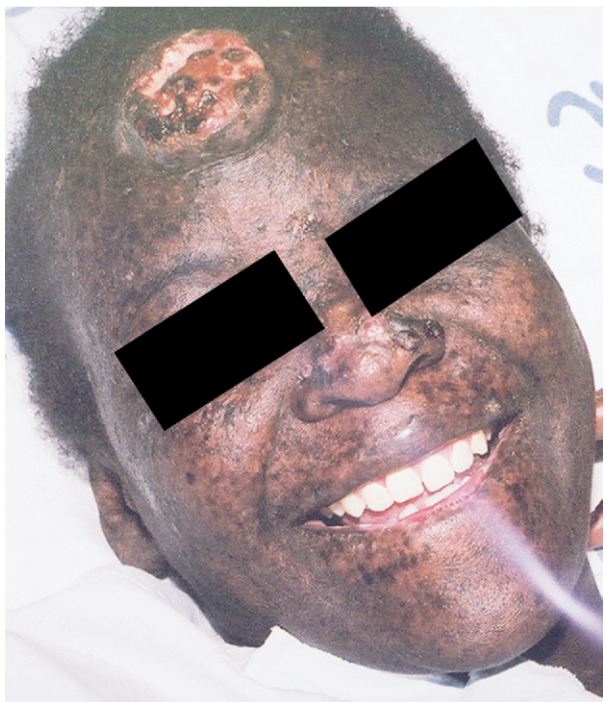

FIGURE 3. Case 4, patient 4. A 12-year-old girl has SCC over the forehead. Note healing site over nose postexcision SCC.

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cause of skin sensitivity in patients with XP and possible induction of malignancy in the noncancerous areas.

In November 2000, there was recurrent forehead tumor with palpable right cervical and preauricular lymph nodes. Excision of tumor and lymph nodes was done.

In August 2001, now 17 years old, she presented with a recurrent, fungating forehead tumor 2 cm in diameter and another fungating tumor 5 cm in diameter over the right parotid region extending onto the submandibular region. Incision biopsy confirmed recurrent SCC. This was now 2 years after excision of a right cheek SCC. Wide surgical excision was done with satisfactory surgical reconstruction using a right deltopectoral flap (Fig 4).

In September 2001, radiation therapy was instituted on the forehead site (4000 cGy in 10 treatments using 21 MeV electrons) and excellent response was achieved.

In January 2002, now 18 years, the patient died at home without any recurrence. She still had satisfactory vision and mild photophobia at the time of her death. An autopsy to ascertain cause of death and any possible metastatic disease was denied.

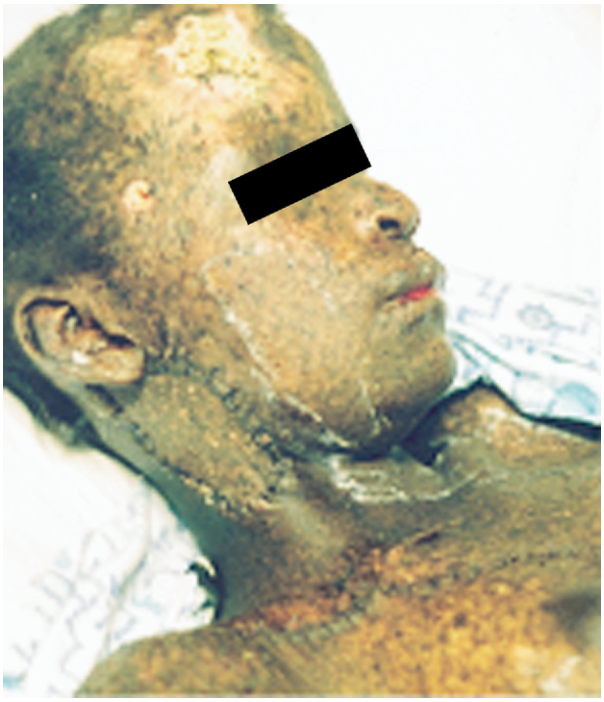

FIGURE 4. Case 4, patient 4. The girl is now 17 years old. Note severe XP all over the body and exposed forehead cortical bone post irradiation for recurrent SCC. Deltopectoral flap used for reconstruction after excision of right parotid and submandibular metastatic SCC.

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CASE 5

Patient 5, a 3-year-old girl, presented in June 1987 with severe XP, severe skin photosensitivity, and blindness. Examination showed a fleshy exophytic tumor 3 cm in diameter on the dorsum of the tongue and another tumor 2.5 cm in diameter on the right lateral border of the tongue. Tumors had been present for the past 10 months. Excision biopsy confirmed both tumors as SCC with margins free of tumor.

In December 1987, 6 months later, she presented with the following tumors: *1*) a tumor on the tip of the tongue 1 cm in diameter; *2*) right upper lip tumor 2 cm in diameter; and *3*) middle right cheek tumor 1.5 cm in diameter. Excision biopsies were done satisfactorily and confirmed all the tumors to be SCC. She was lost to follow-up after 8 months without any recurrence or new tumors developing.

CASE 6

Patient 6, a 5-year-old boy, presented in January 1985 with severe XP, severe skin photosensitivity, and blindness. He had the following tumors: *1*) multiple facial tumors with an average diameter of 2.5 cm; and *2*) 2 pedunculated tumors on the dorsum of the tongue, 2 cm and 1 cm in diameter, respectively. Excision biopsies confirmed the tumors to be SCC. He was lost to follow-up after 3 months without any recurrence or new tumors developing.

CASE 7

Patient 7, a 6-year-old girl, presented in May 1990 with severe XP, severe skin photosensitivity, and blindness (Fig 5). She had the following tumors: *1*) tip of the tongue 4 cm in diameter; and *2*) middle of the lower lip 3 cm in diameter. Excision biopsy confirmed SCC.

CASE 8

Patient 8, a 14-year-old girl, presented in December 1996 with severe XP, severe skin photosensitivity, and photophobia. She had multiple exophytic facial tumors with an average of 1 cm in diameter. She also had a tumor 1.5 cm in diameter on the tip of the tongue. Excision biopsies confirmed all the tumors as SCC.

CASE 9

Patient 9, an 18-year-old girl, presented in April 2005 with mild XP, mild skin photosensitivity, and no photophobia (Fig 6). She had an extensive tumor extending from the right lateral canthus involving the surface of the globe and the surrounding orbital contents. Incision biopsy showed that the tumor was an SCC. Right

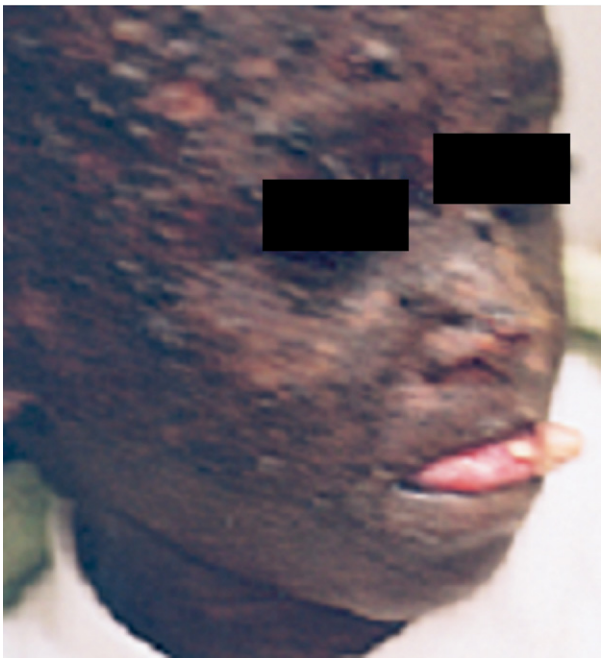

FIGURE 5. Case 7, patient 7. A 6-year-old girl with severe XP, tongue, and cutaneous SCC. Note early SCC over right supraorbital margin.

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orbital exenteration was carried out and histopathologic examination confirmed SCC. She is still under follow-up, 24 months after surgery, without recurrence or new tumor developing.

CASE 10

Patient 10, a 2-year-old boy, presented in June 2005 with mild XP, no skin photosensitivity, no photophobia, and satisfactory vision. He had a tumor 2.5 cm in diameter on the dorsum surface of the tongue. Excision biopsy confirmed SCC. He was lost to follow-up after 8 months without recurrence or new tumor developing.

CASE 11

Patient 11, a 3-year-old girl, presented in January 2006 with mild XP, mild skin photosensitivity, and normal vision. She had a tumor 1.5 cm in diameter on the dorsum of the tongue. Excision biopsy confirmed SCC. Follow-up was possible for 6 months without recurrence or new tumors developing.

CASE 12

Patient 12, a 4-year-old girl, presented in February 2006 with severe XP, severe skin photosensitivity, and severe photophobia. She had the following tumors: *1*) tip of tongue 1 cm in diameter; and *2*) right lateral border of tongue 2 cm in diameter. Excision biopsy confirmed tip of tongue tumor as SCC and lateral border of tongue tumor as FS. She was lost to follow-up after 8 months without recurrence or any new tumors developing.

Table 1 summarizes the clinical characteristics and management of the patients with XP. Table 2 shows the distribution of the patients by gender, cutaneous, ocular, and tongue lesions.

HISTOPATHOLOGIC EXAMINATION

SCC was the most common malignant lesion of the skin, lip, and tongue with tongue lesions being the most common (91.7%; n = 11). The tongue lesions were the main reason for initial presentation probably because they interfered with feeding. However, fibrosarcoma was found on the tip of the tongue of a 3-year-old girl with mild XP. Patient 5 later developed concurrently BCC, SCC, and intradermal nevus in the parotid/submandibular tumor. Ocular surface SCC was noted in an 18-year-old girl with mild XP.

Discussion

The incidence of XP is high in some countries: 15 to 20 per 1,000,000 in Libya, 10 to 25 per 1,000,000 in Egypt, 10 to 25 per 1,000,000 in Japan, Netherlands, and Israel.[13,14,16] XP is rare in our environment with only 12 patients seen over a 25-year period.

Consanguinity in the parents of the patients has been implicated as an etiologic factor. This has been reported to varying degrees of up to 92.8% in XP patients in Libya.[13] It also has been reported in studies

FIGURE 6. Case 9, patient 9. An 18-year-old female with mild XP and ocular surface SCC and involvement of adjacent orbital skin and orbital contents.

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Table 1. CLINICAL CHARACTERISTICS OF THE PATIENTS WITH XERODERMA PIGMENTOSUM

Case No.	Date of Attendance	Gender/ Age	Degree of Pigmentation	Skin Photosensitivity	Malignancy/Site	Treatment	Ocular Conditions	
							Blindness	Photophobia
1	June 1987	F/3	Severe	Mild pigmentation noted at age 3 months	SCC exophytic lesion 2 cm diameter mid dorsum tongue, lower lip	Wide excision	No	Mild
	Dec 1987	F/3	Severe	Severe	SCC tongue tip exophytic 1 cm diameter	Wide excision	No	Mild
	Oct 1988	F/4	Severe	Severe	SCC upper lip 0.5 cm diameter	Wide excision	No	Mild
					Right cheek lesion 1.5 cm diameter	Closure by tissue advancement		
	25 Aug 2000	F/16	Severe	Severe	Fungating, nodular forehead growth 60 mm × 50 mm, 6 months duration History of excision 18 months ago Histopathologic examination: SCC, BCC, intradermal nevus	Wide excision	No	Mild
	Sept 2000	F/16	Severe	Severe	SCC nodular forehead tumor, 3cm diameter	Wide excision	No	Mild
	12 Oct 2000	F/16	Severe	Severe	Recurrent forehead tumor, SCC, 3.5 cm × 2 cm	Excision and resection of outer cortical bone Irradiation	No	Mild
	Nov 2000	F/16	Severe	Severe	Metastatic SCC right submandibular nodes	Right neck dissection	No	Mild
	28 Nov 2000	F/16	Severe	Severe	Recurrent forehead tumor with metastatic SCC right cervical and right preauricular lymph node Metastasis squamous cell carcinoma	Excision of forehead tumor, preauricular cervical lymph nodes Irradiation withheld because of skin sensitivity	No	Mild
	27 Aug 2001	F/17	Severe	Severe	Recurrent tumor, right parotid/mastoid/ submandibular region	Tumor excised modified neck dissection Local flap advancement	No	Mild
	19 Sept 2001	F/17	Severe	Severe	Recurrent forehead tumor SCC	Radiation therapy 4,000 cGy in 10 treatments using 21 MeV Excellent response	No	Mild
	Sept 2001	F/17	Severe	Severe	Recurrent SCC forehead Right Right parotid submandibular tumor	Wide excision with deltopectoral flap reconstruction	No	Mild
	25 Jan 2002	F/18	Severe	Severe	Patient deceased at home	No recurrence	No	Mild

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Table 1. CLINICAL CHARACTERISTICS OF THE PATIENTS WITH XERODERMA PIGMENTOSUM (Cont'd)

Case No.	Date of Attendance	Gender/ Age	Degree of Pigmentation	Skin Photosensitivity	Malignancy/Site	Treatment	Ocular Conditions	
							Blindness	Photophobia
2	June 1987	F/3	Severe	Severe	SCC squamous cell carcinoma dorsum of tongue 2 nodules i) Exophytic 1cm diameter ii) 2.5 cm diameter	Excision	Yes	Severe
	Dec 1987	F/31/2	Severe	Severe	SCC i) Tongue tip 1 cm diameter ii) Upper lip tumor 2 cm diameter iii) Right cheek lesion 1 cm diameter	Excision	Yes	Severe
3	Jan 1985	M/5	Severe	Severe	SCC i) Multiple facial lesions average diameter 2.5 cm ii) 2 tongue lesions pedunculated, 1 cm diameter	Excision	Yes	Severe
4	Feb 1982	F/3	Severe	Severe	SCC	Excision	Yes	Severe
5	May 1983	M/9	Severe	Severe	SCC left cheek 4 cm diameter	Excision	Yes	Severe
6	Jun 2005	M/2	Mild	Mild	SCC dorsum of tongue 1.5 cm	Excision	Yes	Mild
7	Feb 1984	F/9	Severe	Severe	SCC i) Tongue tip ii) Upper lip iii) Right cheek, 4 cm diameter	Excision	Yes	Severe
	Dec 1985	F/10	Severe	Severe	Right alae nasi, 3.5 cm in diameter	Excision	Yes	Severe
8	May 1990	F/6	Severe	Severe	SCC i)Tongue tip ii) Lower lip	Excision	Yes	Severe
9	Dec 1996	F/14	Severe	Severe	SCC multiple exophytic facial and tongue lesions	Excision	Yes	Severe
10	Jan 2006	F/3	Mild	Mild	Fibrosarcoma tongue dorsum 1.5 cm diameter	Excision	Yes	Severe
11	Feb 2006	F/4	Severe	Severe	SCC dorsum tongue	Excision	Yes	Severe
12	Jan 2003	F/18	Mild	Mild	Ocular surface SCC	Excision	No	Mild

Abbreviations: BCC, basal cell carcinoma; SCC, squamous cell carcinoma; XP, xeroderma pigmentosum.

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Table 2. DISTRIBUTION OF PATIENTS BY GENDER, CUTANEOUS, OCULAR, AND TONGUE CONDITIONS

Gender	Patients (n)	Clinical Degree of XP		Cutaneous Conditions				Lip/Tongue Conditions		Ocular Conditions		
		Mild	Severe	Photosensitivity	SCC	Intradermal Nevus	BCC	SCC	Fibrosarcoma	Blindness	Photophobia Mild	Photophobia Severe
Male	4	1	3	4	4	0	0	4	0	4	1	3
Female	8	2	6	8	8	1	1	7	1	6	2	6
Total	12	3	9	12	12	1	1	11	1	10	3	9

Abbreviations: BCC, basal cell carcinoma; SCC, squamous cell carcinoma; XP, xeroderma pigmentosum.

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from Egypt, Pakistan, and Nigeria among some of the countries that have a high incidence of XP.[1,3,4] No history of consanguinity could be elicited in any of our patients.

There is, however, a varying degree of severity and the rate of progression of the disease even among siblings.[1,4,13] El-Hayek et al[17] described XP in 4 siblings in a consanguineous Pakistani family in which 1 had 3 different types of malignancies simultaneously and different rates of disease progression with death at 2 years, 2 years, 7 years, and 13 years, respectively. This variation has also been described in a Nigerian family of 3 consecutive siblings who presented with variable oculocutaneous manifestations of XP.[4] All these children are from the same families living in the same environment and exposed to the same etiologic factors. This supports the view that various phenotypes of this disease are in existence.

The female preponderance in the present case series is in agreement with other studies.[3,4] Some studies, however, show an equal male and female affliction.[13,18] History of onset of the signs and symptoms of XP in our case series began at 1 to 3 months after birth, initially mild but with the degree of pigmentation increasing over the years. The youngest patient recorded at presentation of the first skin malignancy is 18 months.[13] In our series the youngest patient who presented with cutaneous malignancy was 3 years old. Most series report an average age of 2 to 3 years at onset of first cutaneous malignancy.[3,13] In the present series the average age at presentation of the cutaneous malignancies was 4.6 years. This is a slightly older age group than in other series but considering the large size of the tumors one could suspect that the tumors must have been present for some time, but because of late presentation to hospital (a feature common in our environment) the tumors could have started much earlier than the age of presentation. In our series all but 1 of our patients were young children at the time of presentation. This is similar to other series that show a predominance of young children and also the fact that signs and symptoms of XP start in early childhood and worsen as the child grows.[3,4,8] In Japanese studies, however, XP has been noted in the 42 to 62 years age group.[16] This could be related to the developed health facilities, health seeking awareness, and the general longevity of the population.

There are few reports of fibrosarcoma in XP patients.[4] Only 1 case was found in our case series, and there also was 1 case of BCC. The patient with BCC also had SCC and intradermal nevus (case 4, patient 4). XP is a serious disease in the tropics because of its pronounced skin photosensitivity and the propensity for developing skin malignancies. Management is difficult because of the elaborate photoprotection that is required right from birth. Avoidance of sun exposure is not possible due to the nature of our tropical climate although some measures such as special glasses, clothes, and sunscreen creams can be instituted. These, unfortunately are too expensive in resource-poor settings and the virtual high sunshine all year round.[1,4,13]

The majority of our patients presented with the severe form of the disease probably because of the tropical nature of our climate (intense UV radiation) coupled with the lack of awareness of the disease and hence lack of any form of photoprotection against the high UV exposure. Sunlight exposure is implicated in the induction of cutaneous malignant melanoma as well as non melanoma malignancies (BCC, SCC); in patients with XP,[11] SCC was the malignant lesion noted on the facial skin and the tongue in our case series. The tongue SCC (91.7%, n =11) was higher than in most series, for instance, 1.6% (n = 13) in a series of 830 patients by Kraemer[18] and 0% to 20% in other series.[3,9,19] There seems to be an increase in the reports on intraoral involvement in XP as only 21 cases had been reported by 1981.[7] XP also predisposes to oral cancer.[20] XP patients younger than 20 years of age have an estimated 10,000 times greater frequency of carcinoma of the anterior tongue.[20] Carcinoma of the palate and gingiva are also known to occur with increased frequency. The intraoral site could be presumed to be shielded from direct UV radiation and thus less prone to malignant transformation. However, exposure of the tongue tip and dor-

sum surface to UV radiation during normal tongue function in areas of high UV exposure could be an etiologic factor. It has also been suggested that gross anemia in some of these patients, especially from resource poor settings, where most reports on tongue involvement are coming from, could lead to atrophic changes on the lingual mucosa rendering it susceptible to malignant transformation.[7] A similar high oral involvement in XP was noted in Libya and Sudan where there is also intense UV radiation.[7,9,13]

No MM was noted in our patients. MM has been reported in patients with XP presenting as single or multiple lesions and also simultaneously with SCC.[1] Multiplicity of different tumors increases with age and has been reported in adults mostly.[1] Patient 4 developed multiple different tumors later on in life, BCC with SCC. Multiplicity and the development of histologically different types of tumors can also occur in children younger than 6 years.[1] This was noted in our series where 1 patient had cutaneous SCC along with FS of the tip of the tongue.

Exposure to sunlight alone may not explain the severity and rate of disease progression as variance has been noted in siblings in a common environment characterized by the same high sunshine.[4] Studies have shown, however, that there is correlation of the clinical manifestations and gene mutations even among patients of the same complementation group.[4] It was not possible to do complementation studies in our patients. XP patients younger than 20 years of age with the severe form have a higher than 1,000-fold risk of developing early cutaneous malignancies.[2,3,18] This was confirmed in our series in which most patients had severe XP and were younger than 5 years of age when they developed facial skin and tongue SCC. Cutaneous SCC was 100% in our patients as opposed to other studies that show 60%[19] and 16.7%.[3] SCC lesions also were noted on the lips.

Photophobia is the earliest ocular symptom and is seen more commonly in young patients than in adults.[3,9] All our patients had marked photophobia; even those who were blind mentioned photophobia before loss of vision.

Ocular neoplasms like SCC, BCC, and MM are reported frequently from the conjunctiva, cornea, and eyelids.[3] Kraemer et al[18] and Goyal et al[19] reported that 11% and 20% of their cases had ocular malignancies, respectively. This contrasts with 8.3% (n =1) in our case series; only 1 patient had ocular surface SCC. Blindness from severe XP at the time of presentation was common among our patients, 83.3% (n = 10). This is unlike other reported series where it was rare.[3] This might be related to lack of understanding of ocular involvement in our patients as their reasons for presentation to the hospital was for skin and tongue lesions mainly.

Management of XP in our environment is complicated by the difficulty of instituting photoprotection by way of special glasses, clothes, and sunscreen creams as they are too expensive for the majority of our patients. Avoidance of sun exposure is impossible due to the tropical nature of our climate.

The current treatment modalities for XP are prophylactic avoidance of sun exposure, chemotherapy, radiotherapy, surgical excision of premalignant and malignant tumors, resurfacing with skin grafts, and dermabrasion. No effective treatment has been found. Surgical excision was our treatment of choice for the multiple extensive facial and tongue lesions, due to delay in seeking medical attention. Skin grafting for cosmesis was not done as there was intense extensive pigmentation all over the body with no unaffected area to harvest the skin graft from. Dermabrasion also was not considered because of the severe nature of the XP. Dermatome shaving and dermabrasion has been found useful as a prophylactic measure in XP. This treatment relies on re-epithelialization by the cells derived from those lying deep in the skin adnexa that have received less exposure to UV radiation. The cosmetic result is better with dermabrasion than with split skin grafting.[21]

Radiation therapy was withheld initially in 1 patient for fear of inducing further malignant transformation on the noncancerous areas. Radiosensitivity at both the clinical and cellular level in an XP patient assigned to genetic complementation group C has been reported and there is still need to resolve the extent of radiosensitivity in XP.[22] It was, however, used on 1 patient successfully who had repeated recurrences on the forehead after surgical excision (patient 4). Delay in presentation resulted in the extensive lesions seen in most of our patients. This may be attributed to poor health education and awareness of the disease.

Early recognition of XP will assist in instituting preventive measures and early detection of related ocular and cutaneous lesions to improve quality of life and longevity. The majority of patients with XP do not survive beyond the third decade.[4,9] This is even shorter in resource-poor settings where comprehensive photoprotection is difficult with the added disadvantage of intense tropical UV radiation. This is the case in the present series. Most of our patients were lost to follow-up within 6 to 12 months and presumed dead from local and metastatic disease. Our longest survivor with follow-up died at 18 years.

This study shows that XP is rare among black Africans and tends to present in the severe form with tongue, lip, and cutaneous SCC, early blindness, and early death.

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