

Xeroderma Pigmentosum with Multiple Cutaneous Malignancies: A Series of Four Cases

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ABSTRACT

Xeroderma Pigmentosum (XP) is an autosomal recessive disease. The pathogenesis lies in the Nucleotide Excision Repair (NER) genetic defect leading to various cutaneous and other malignancies developing at a very young age alarming the identification of precursor lesions and to adapt preventive measures. The DNA damage that occurs in exposure to UV rays in sunlight, gets repaired by a set of XP genes. The function of each gene works in a coordinated manner and the defective nucleotide gets excised from the DNA strand and a correct base pair is inserted. But in XP, the DNA repair mechanism defect leads to clinical manifestations involving the skin, eye and central nervous system. Here we report four cases of XP patients, out of them, two were siblings with a history of consanguineous marriage of their parents. All the patients were highly sensitive to sunlight and developed freckles, hyper and hypopigmentation, dryness, atrophy and even cutaneous malignancies. Ophthalmological examination revealed corneal dryness, scarring and partial blindness in two cases. Neurological examination revealed suboptimal intelligence and disturbed gait in two cases. The clinical diagnosis of XP was established and biopsy from the skin lesions revealed pyogenic granuloma, squamous cell carcinoma and basal cell carcinoma. Unfortunately, XP has no cure. But with rigorous sun protection, preventive measures and regular follow-up, they have not developed any new malignancies. As these paediatric patients are prone to develop malignancy with sun exposure, avoidance of UV light exposure is the preventive measure and children are encouraged to attend evening schools. Early identification of the disease and precancerous lesions may decrease the morbidity of XP patients.

Keywords: Basal cell carcinoma, Nucleotide excision repair defect, Squamous cell carcinoma, Xeroderma pigmentosum

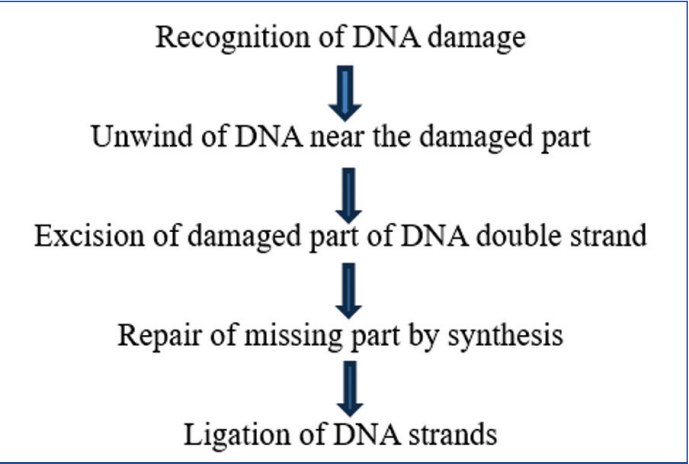
INTRODUCTION

The XP is an autosomal recessive genetic disorder that affects the skin, eyes, and nervous system. In 1874, Sir Moriz Kaposi diagnosed this hereditary disease [1]. The pathogenesis stems from mutations involving nucleotide repair genes. In a physiological state, the DNA damage that happens due to ultraviolet rays, gets repaired by a set of genes. Researchers highlighted the involvement of eight genes- XPA, XPB, XPC, XPD, XPE, XPF, XPG and XPV. Each gene involved in XP contributes a specific function that works in coordination with the others and the damaged part of DNA double-strand gets repaired [Table/Fig-1]. Mutation in any gene leads to failure in its role and clinical manifestation differs in each case [Table/Fig-2] [2]. Diminished DNA repair capabilities in the cells exposed to the UVB range of sunlight explain the photosensitivity and carcinogenesis observed in XP cases.

The XPA, XPC and XPV mutations are the most common and they constitute 75% of cases globally. XPA gene also known as Damage

Physiological function	Gene	Mutation
Damage recognition	XPC, XPE	Failure in damage verification
Unwinding of DNA strand	XPA, XPB, XPG	Failure in unwinding
Incision at the mutation site	XPG, XPF	Failure in incision
Excision of the damaged part	XPG	Failure in excision
Gap filling in the strand	XPV/DNA polymerase	Unable to synthesise new strand
Ligation	Ligase 1	Failure in translesion DNA synthesis (a DNA repair mechanism)

[Table/Fig-2]: Role of XP genes and their mutations.



[Table/Fig-1]: Steps in Nucleotide Excision Repair (NER) pathway.

Binding Protein 1 (DDB1) located in chromosome 9q22 assists the XPC gene in the unwinding of the damaged DNA. Mutation in the XPA gene clinically manifests as cutaneous and neurological abnormalities [1]. In the United States, XPC located in chromosome 3p25 mutation is common involving the skin only. XPV gene, the polymerase located on chromosome 6p21 plays a vital role in post-replication repair. However, the XPV gene is not directly involved in the NER pathway. 30% of XP have XPV mutation [2]. Rest gene mutations are relatively rare. Some are associated with Cockayne Syndrome and Trichothiodystrophy [3].

Epidemiological data revealed a maximum number of cases in Japan with an incidence 45/million followed by 2.3/million in Western Europe [2]. Though many case reports on XP from India are reported, incidence data is not available. States like Karnataka reported many cases within families where consanguineous marriages were observed [4].

Cutaneous features present as early as the first month with persistent photosensitivity. The patients may have poor physical development and short stature. There is hyperpigmentation, and hypopigmentation involving the face, neck, back, hand, and chest. Ongoing solar damage leads to premature aging of the skin, xerosis, atrophy, and

diffuse freckling. These changes further lead to premalignant lesions like actinic keratosis, and dysplasia followed by cutaneous malignancies like squamous cell carcinoma, basal cell carcinoma, malignant melanoma, and rarely Merkel cell carcinoma [5-10].

A review of the literature has shown that patients with XP have a 10,000-fold increased risk of developing Squamous Cell Carcinoma (SCC) and Basal Cell Carcinoma (BCC), and a 2,000-fold increased risk of developing melanoma compared with the general population of the same age group. These patients are also at increased risk of developing Central Nervous System (CNS) neoplasms [11].

Involvement of ocular adnexa like eyelids, cornea, and conjunctiva is most common leading to photophobia, keratitis, and corneal opacity in severe cases [12]. These patients usually have low Intelligence Quotient (IQ) due to neurological deficits in 20-30% of cases [13]. Gradually, sensorineural deafness, ataxia, areflexia and spasticity develop and lead to loss of neurons and cortical atrophy. There is also an increased risk of malignancies 1000-fold in the anterior segment of the eye and 50-fold in CNS tumours [1,2]. As these cases are more prone to develop cutaneous and systemic malignancies at a very young age and these precancerous lesions require histopathological examination for the diagnosis, identification of these patients is vital to protect them and prevent further new cutaneous malignancies to develop.

CASE SERIES

Case 1

Case 1 was a 12-year-old girl belonging to a low socio-economic status visited us with a chief complaint of an ulcerated plaque near her nose for three weeks, a nodule in the left cheek and back developed within two months. On physical examination, the patient had hypopigmented and hyperpigmented macules over her body involving face, back and both hands. The skin was dry and mottled. The skin changes started since she was two-year-old after exposure to sun. The plaque near the left ala of nose was measuring 2 cm×1 cm with surface ulceration, haemorrhagic uneven base and crusting margin at one end [Table/Fig-3a-c]. The back nodule was ulcerated measuring 2.5 cm×2 cm. With these clinical features, the case was diagnosed as XP with a differential diagnosis of Bloom's syndrome and Cockayne syndrome. Her parents had a consanguineous marriage. Ophthalmological examination showed conjunctival redness with B/L visual acuity 6/6. The patient didn't have any neurological deficits as screened by MRI and IQ test. To investigate the nature of plaque near nose and nodule in the back, scrape cytology followed by biopsy were done. Scrape cytoscans from the plaque revealed dysplastic squamous cells. Histopathology revealed malignant squamous cells showing keratinisation, infiltration into the dermis with epithelial atypia and atypical mitotic figures. The ulcerated plaque lesion was diagnosed as squamous cell carcinoma, well-differentiated [Table/Fig-3d]. The back nodule was completely excised and histopathology showed lobular arrangement of small, ectatic capillaries within an oedematous, inflamed stroma and diagnosed as pyogenic granuloma [Table/Fig-3e]. The patient was treated with 5% Imiquimod cream to apply locally over the nasal ulcerated site twice weekly for 12 weeks and was advised to avoid sun exposure, use sun-glasses and gloves to cover the exposed part of body while going out. After two years of follow-up, there were no new lesions after careful adaptation of precautionary measures.

Case 2

The second case was a nine-year-old boy. He was an offspring of consanguineous marriage and he was the brother of Case 1. The boy presented to us with nodule over scalp and on upper chest wall. On general examination, He had freckles, seborrheic keratosis and hypopigmented and hyperpigmented macules over all exposed parts of the body, but the neck was spared [Table/Fig-4a,b]. All these skin lesions started when the boy was 2.6-year-old and gradually spread

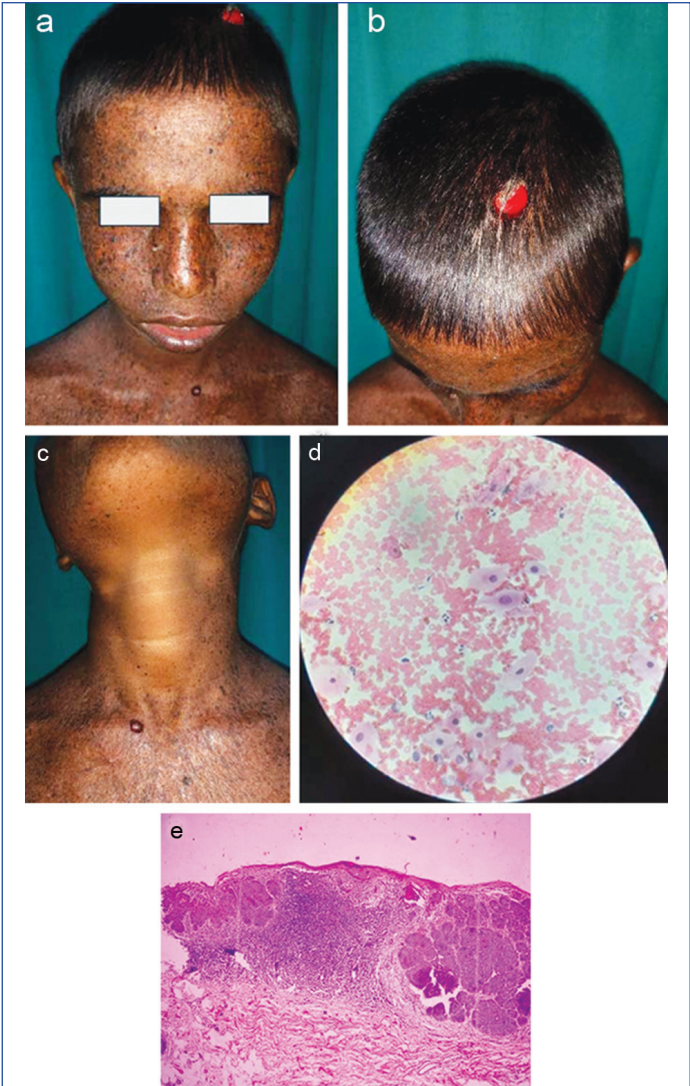
all over the body. The scalp nodule was haemorrhagic measuring 2 cm×1.5 cm. It developed two months back and gradually increased in size with intermittent bleeding with trauma [Table/Fig-4c]. There was a nodular lesion with smooth surface in the upper chest wall measuring 0.5 cm×0.5 cm. B/L eyes had corneal opacity with a vision of 6/18 and 6/12 in right and left eye, respectively. He was having normal growth and development, but mild mental retardation was noticed. Clinically, the patient was diagnosed as XP with squamous cell carcinoma of scalp and nodular basal cell carcinoma in the chest wall. The scalp nodule was excised and histopathology revealed features of squamous cell carcinoma, well-differentiated [Table/Fig-4d]. Biopsy from the chest wall nodule showed lobules of basaloid cells with peripheral nuclear palisading and fibromyxoid stroma. The diagnosis was established as nodular basal cell carcinoma [Table/Fig-4e]. The patient was referred to onco-surgery department for further evaluation and was advised not to get exposed to sun and to apply 5% imiquimod cream for 12 weeks along with sunscreen. After a follow-up of three years, the patient had not developed any new skin lesions. However, the visual defects couldn't be treated completely.



[Table/Fig-3]: Girl has an ulcerated plaque near nose: a,b) and a nodule in the back; c) Histopathology from the ulcerated plaque showed features of squamous cell carcinoma, Well-differentiated (H&E, 100X); d) and nodule showed features of pyogenic granuloma; e) (H&E, 400X).

Case 3

Case 3 was an 11-year-old boy of a tribal population and he had freckles and macules over both the hands, face, neck and back. The skin lesion started when he was only 1.8-year-old. He came to

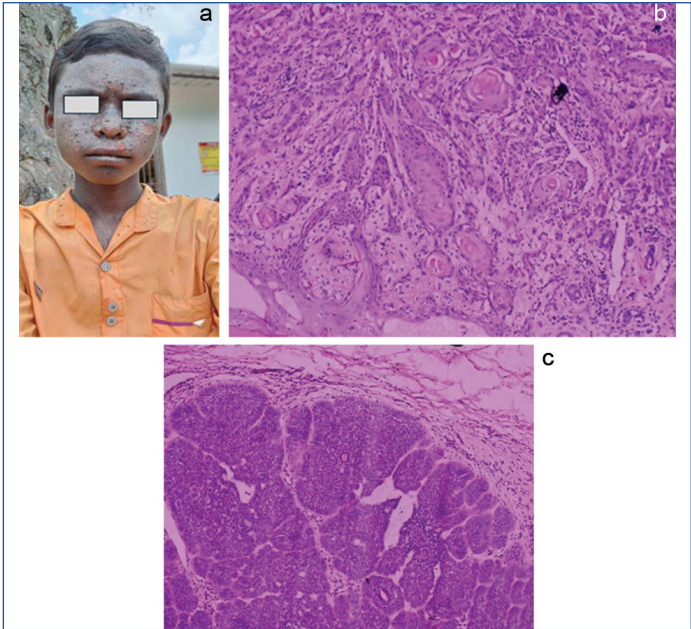


[Table/Fig-4]: A 9-year old boy showed a nodule over scalp and corneal opacity of both eyes: a,b). Neck is spared and nodule was present in the lower neck; c). FNA from the scalp nodule showed atypical keratinocytes scattered over a haemorrhagic background; d) (H&E stain, 400X). Histopathology from chest wall nodule showed clusters of basaloid cells with palisading and minimal atypia; e) (H&E stain, 100X).

dermatology OPD with a complaint of an ulcerated nodule near left nasal bridge and an ulcerated plaque in the left upper lip. The nodule was measuring 0.5 cm×0.5 cm. It had surface ulceration, rolled out margins and uneven base. The plaque was of size 2.5 cm×2 cm with uneven base [Table/Fig-5a]. The patient had disturbed gait with suboptimal intelligence. Ophthalmological examinations were within normal limits. With a clinical diagnosis of XP, FNAC and biopsy of said lesions were carried out. FNA smears from the nodular lesion near nose revealed tightly packed uniform looking basaloid cells with peripheral palisading. Histopathological findings were of basal cell carcinoma with a depth of invasion 1.6 mm without any lympho-vascular involvement and peripheral margins were free [Table/Fig-5b]. Histopathological examination of ulcerated plaque in the upper lip revealed pleomorphic squamous cells with invasion and good number of keratin pearls and diagnosed as squamous cell carcinoma, well differentiated type [Table/Fig-5c]. The patient was referred to oncology department for further management and advised the patient to apply local 5% Imiquimod cream twice weekly for 16 weeks. After two years of follow-up, he had no history of recurrence or metastasis.

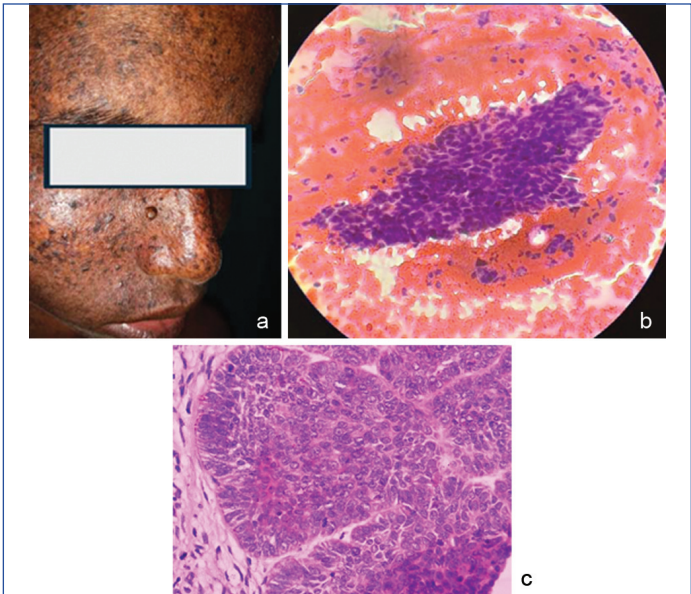
Case 4

The last case was a boy of nine-year-old without any history of consanguineous marriage of his parents. His skin was dry and had a mottled appearance. The patient had developed photosensitivity since last six years with thin, brittle hair. He had a nodule over the bridge of his nose measuring 0.6 cm×0.5 cm with smooth surface



[Table/Fig-5]: Boy had double malignancies clinically presenting as an ulcer with rolled out margins in the nasal bridge and a plaque in the upper lip: a). Biopsy from the nose was basal cell carcinoma with peripheral palisading, cleft artifact; b) (H&E, 400X). Histopathology from the upper lip was infiltrating Squamous cell carcinoma; c) (H&E, 100X).

since last two months [Table/Fig-6a]. His right eye had partial corneal opacity with a visual acuity 6/18. Neurological examination was normal. With a clinical diagnosis of XP, pathological evaluation of the nodule was done. Both FNAC and biopsy from the nodular lesion showed basaloid cells in lobules with peripheral palisading and retraction artifact along with a fibromyxoid stroma [Table/Fig-6b,c]. Final diagnosis was XP with nodular basal cell carcinoma. Close monitoring, local application of 5% Imiquimod cream for 12 weeks and follow-up were carried out with preventive measures as the patient had minimal complications. [Table/Fig-7] presents clinical summary of cases.



[Table/Fig-6]: Boy had a nodule in the nose with partial visual loss: a) FNA; b) (H&E, 400X) and histopathology; c) showed basaloid cell clusters with peripheral palisading, minimal atypia and mitosis (H&E, 400X).

Variables	Patient 1	Patient 2	Patient 3	Patient 4
Age	12 years	9 years	11 years	9 years
Sex	Female	Male	Male	Male
Consanguineous marriage history of parents	Yes	Yes	No	No
Living standards	Low	Low	Low	Low

Start of skin lesions	2 years	2.6 years	1.8 years	3 years
Skin lesions	Freckles Nodules over left cheek and back Ulcerated plaque over nose	Freckles Papule Seborrheic keratosis	Poikiloderma Telangiectasia Freckling and hypopigmented macules over both fore arms	Photosensitivity Mottled pigments Xerosis Thin hair
Site of malignancy	Nose, upper back	Scalp, chest wall	Nose, upper lip	Nasal bridge
Site of biopsy taken	Nose, upper back	Scalp, chest wall	Nose, upper lip	Bridge of the nose
Histopathology diagnosis	SCC, Pyogenic granuloma	SCC, nodular BCC	BCC, SCC	Nodular BCC
Ophthalmological findings	Conjunctival congestion	Corneal opacity, partial blindness	Normal	Right eye partial blindness
Neurological findings	Normal	Mild mental retardation	Disturbed gait, subnormal intelligence	Normal
Management	Imiquimod cream Artificial tear	Imiquimod cream Artificial tear UV sunglasses with side shield	Imiquimod cream Protective measures advised	Imiquimod cream Protective measures advised
Follow-up	No new skin lesions after 2 years	No new skin lesions after 3 years of follow-up	No recurrence of malignancy within 2 years	No new skin lesions within 1.5 years

[Table/Fig-7]: Presents a clinical summary of the cases.

DISCUSSION

The XP is a rare disorder and begins in early childhood. The mutation that involves the particular gene manifests differently. As it is an autosomal recessive transmission, the disease occurs more commonly in consanguineous marriages. Jaouad IC et al., study found an increased number of XP cases in consanguineous marriages (15.25%) [14]. In this case series also, two cases (50%) were siblings and had consanguineous parents.

The XP patients are born with normal skin. To start with, photosensitivity is observed as early as six months of age. Gradually, by the age of two years, poikiloderma, skin atrophy, xerosis, telangiectasia, and hypo and hyperpigmentation patches develop. The DNA repair genes defect leads to a deficiency of fibroblast function and UV damage cannot be repaired effectively [15]. Premalignant skin changes like actinic keratosis progresses to cutaneous malignancies like SCC, BCC, malignant melanoma, and Merkel cell carcinoma [16]. In our case series, all four cases (100%) had cutaneous malignancies. One boy had both SCC and BCC at the age of only 11 years. Vora R et al., reported 18-year-old girl having XP with melanoma and SCC [17].

Melanin is considered a UV-protective pigment. It effectively repairs the UV rays-induced DNA damage with an intact NER mechanism. This explains the increased incidence of cutaneous malignancies in the light complexion population than dark as the melanin content is less. But, when the NER mechanism is defective, the UV rays penetrate and damage the DNA and the melanin can't repair this defect. Studies have shown that XP patients have similar incidences of carcinoma in both light and dark complexion nullifying the protective role of melanin pigment [18].

The XP may have neurological and ophthalmological defects on the basis of genetic mutation of eight genes. These patients should be differentiated from XP/Cockayne Syndrome and trichothiodystrophy on the basis of clinical presentation. Cockayne syndrome patients are classically 'Cachectic dwarf' appearance with thinning of skin and hair, cataracts, ataxia, and retinopathy. Trichothiodystrophy patients

have ichthyosis, brittle hair, micrognathia, and protruding ears-like features [19,20]. Tamhankar PM et al., performed a mutational analysis on 13 cases of XP from the Indian population and concluded that XPA gene mutation is associated with moderate to severe mental retardation in 60% of cases [21]. Nearly 30-40% of XP cases have a different spectrum of neurological manifestations [22]. In our case series, 1/4 (25%) have low IQ. Spasticity and seizures were not observed. Gulanikar A et al., also reported 10% neurological involvement in XP patients [23].

The XP patients are more prone to UV radiation affecting the anterior parts of the eye like the cornea, conjunctiva, and eyelids as the lens acts as a barrier and protects the posterior parts [24]. Many studies have shown that 90% of XP cases have ocular manifestations in the form of photophobia, blepharospasm, conjunctival xerosis, pinguicula, pterygium, corneal ulceration and neovascularisation, pannus formation, etc., [25]. Genetic mutations in XPC, XPE, and XPV genes are more commonly associated with ocular involvement [26]. Lopes-Cardoso C et al., reported a 41-year-old man, XP with conjunctival congestion, perioral scars, dental scars, and periodontal problems [27]. In our cases, two out of four had partial blindness.

Due to NER defect, though cutaneous malignancies are more common in XP, other internal organ tumours were also reported. Bencharef H et al., reported a case of XP with Acute Myeloid Leukaemia (AML) [28]. Multiple skin cancers can also arise in a patient. Out of 4 cases, one 11-year-old boy had both SCC and BCC over his face. Frequent biopsies and excision of malignant lesions involving the nose, lips, eye, and ear are of cosmetic importance and may be an inciting factor for depression low self-esteem, and social withdrawal behaviour. For confirmation of XP, skin fibroblasts are exposed to UV rays, and the DNA synthesis pattern in damaged cells is observed [3].

As it is a genetic defect, there is no cure. Cutaneous lesions can be reduced by adopting proper preventive measures. Patients are advised to be fully covered and use UV-protected sunglasses with side shields. The children are encouraged to attend night school. Regular dermatological and ophthalmic visits are recommended. Besides these, regular vitamin D supplementation is done to avoid its deficiency as they are protected from sunlight. Malignant tumours are excised and treated with conventional chemotherapy. The topical application of Imiquimod (5-Fluorouracil) is promising for both premalignant and excised malignant lesions. We have treated all four cases with topical Imiquimod. None of them presented with new lesions within a follow-up period of three years. Cemiplimab, an immune checkpoint inhibitor targeting PD-1, has been approved for locally advanced and metastatic cutaneous squamous cell carcinoma. Though its role is not clear, it can be used as a drug of choice in locally advanced, unresectable, and metastasized squamous cell carcinoma.

As authors had a newly established medical college without molecular laboratory due to financial constraint and the patients belong to low socio-economic status, They could not afford molecular testing for the genetic defect.

CONCLUSION(S)

The XP is a genetic disease with no cure. Due to frequent DNA defects without an efficient repair mechanism, cutaneous and other malignancies are more frequent in these patients occurring at an early phase of life. The XP associated cutaneous malignancies need histopathological confirmation and further management. Clinicians should be vigilant enough in these cases and preventive measures can increase the quality of lives. The treatment is challenging and it needs a multidisciplinary approach. Adapting preventive measures and regular follow-up may decrease morbidity.

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