

# Nasal basal cell carcinoma in xeroderma pigmentosum - diagnosis, workup, management, and reconstruction



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## ABSTRACT

Xeroderma pigmentosum (XP) is genetic autosomal recessive disorder with photosensitivity on exposure to sun and also presents with various malignancies such as squamous cell carcinoma, basal cell carcinoma, and malignant melanoma. A 22-year-old female visited our outpatient clinic with 18 months history of pigmented nodule on her nose. She was diagnosed with XP since she was 2 years old. Physical examination revealed a waxy, blackish-brown nodule on her nose. Incisional biopsy was performed. The histological findings revealed multiple, palisaded basophilic tumor masses surrounded by prominent infiltrating lymphocytes confirming our clinical diagnosis. She was advised sun protection with the use of protective clothing and glasses. XP leads to chances of various malignancies. Early diagnosis, proper clinical examination, histopathological examination for the diagnosis of condition is must to come to proper diagnosis. Instructions to the patients to take proper protection from sun and wearing protective clothing must be given.

**Key words:** Basal cell carcinoma; Histopathological; Incisional biopsy; Sun protection; Xeroderma pigmentosum

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## INTRODUCTION

Xeroderma pigmentosum (XP) is a rare autosomal recessive disorder, characterized by failure of deoxyribonucleic acid (DNA) nucleotide excisional repair after sun-induced damage from ultraviolet (UV) B light (a spectrum of 280–320 nm).<sup>1,2</sup> It results in dry pigmented skin.<sup>1,2</sup> Patients develop photosensitivity at the early age of 1–2 years. It increases the chances of various skin cancers such as basal cell carcinomas (BCCs), malignant melanomas, and squamous cell carcinomas (SCCs) than the normal population.<sup>3,4</sup> Disease causing mutations have been identified in 8 different genes in patients with XP. 7 of these genes (XPA, XPB, XPC, XPD, XPE, XPF, and

XPG) are involved in nucleotide excision repair and the eighth gene is involved in mutation of damaged DNA.<sup>5,6</sup> Together with T regulatory cells, myeloid-derived suppressor cells, tumor-associated macrophages, and CD163+ M2 macrophages have recently been shown to contribute to establishing the tumor microenvironment in skin cancer.<sup>7,8</sup>

## CASE REPORT

A 22-year-old female visited our outpatient clinic with 18 months history of pigmented nodule on her nose. She had photosensitivity since she was 2 years old and was diagnosed with XP. On her initial visit, physical

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examination revealed a waxy, blackish-brown nodule on her nose. The size of the nodule was approximately 20 × 10 mm in diameter. Besides, she had multiple hypo- and hyper-pigmented macules all over her body, especially on sun-exposed area. An incisional biopsy of the nose lesion was performed. Histopathological examination of the biopsied specimens showed features of BCC.

Based on the above findings, patient diagnosed as having BCC with XP and excised the tumor with a 2-mm surgical margin from the lesion. The histological findings revealed multiple, palisaded basophilic tumor masses surrounded by prominent infiltrating lymphocytes confirming our clinical diagnosis (As in Figure 1). The procedure of surgery was- Peritumoral incision was taken. Blunt dissection was done, lesion was separated from the surrounding tissue and was removed in toto. Defect was reconstructed using forehead rotation flap (supratrochlear based) (as shown

in Figure 2 and 3). Donor site was repaired using STSG (from neck and neck defect was closed primarily). Hemostasis was achieved nicely. Primary closure was done. Incision was closed in layers and dressing was done. Figure 4 in our case report is of post recovery.

A contrast-enhanced computed tomography of the paranasal sinuses and neck was performed. There was no abnormality detected on the imaging. Investigations were within normal limits. Chest X-ray was normal. She was advised sun protection with the use of protective clothing and glasses.

## DISCUSSION

XP is autosomal recessive disorder characterized by extreme sensitivity to sunlight, resulting in sunburn, pigment changes in the skin, and a greatly elevated incidence of skin cancers. It manifests few months after birth. Estimated incidences vary from 1 in 20,000



Figure 1: Lesion over nose



Figure 3: Patient with flap



Figure 2: Rotational flap applied over nose



Figure 4: Post recovery

in Japan to 1 in 250,000 in the United States (USA), and approximately 2.3/million live births in Western Europe.<sup>2</sup> The median age of onset of non-melanoma skin cancers reported in patients with XP is 8 years, compared to 60 years in the healthy population.<sup>9</sup> The two most common types of cancer found in XP patients are BCC and SCC, mainly occurring on the face, head, and neck. Melanomas occur in one-fourth of cases, and one-third of these occur in the head and neck.<sup>10</sup> Early detection of these malignancies is necessary because they are fast growing, metastasize early and lead to death. Case reports have been reported in patients with XP having cancers.

In the study by Aisyah and Hidayati 5-year-old girl complained of brown and black spots on the entire body since the age of 7 months. Histopathology examination results in their study of left infraoculi region was malignant SCC and the nasal region was malignant BCC.<sup>2</sup> In our study also, BCC was found in the nose region.

In the study of Furudate et al., 26-year-old Japanese had BCC along with XP. The histological findings revealed multiple, palisaded basophilic tumor masses surrounded by prominent infiltrating lymphocytes.<sup>11</sup> In the study of Grampurohit et al., a case of XP in an 18-year-old male presenting with multiple cutaneous malignancies: SCC, malignant melanoma, and pigmented BCC. BCC was seen in the inner canthus of the eye, SCC was seen in the lower lip, mass from the neck had metastatic melanoma. All findings were confirmed on histopathological examination.<sup>9</sup>

In the study of Halkud et al., 11 cases of XP were noted six males and five females. SCC of the lids were seen in seven cases while BCC was seen in 8 cases and limbal and conjunctival SCC was seen in one case.<sup>12</sup> In study conducted by Mulimani et al., a 9-year-old child with XP scheduled for excision of BCC under general anesthesia.<sup>13</sup>

Many risk factors are known to exacerbate the cutaneous features resulting in skin cancers and early death. These risk factors include sunny weathers, outdoor living, fair skin, smoking, poor availability of diagnostic facilities, delayed diagnosis, and poor protection from sunlight.<sup>14</sup> For prevention sunscreens are useful. Sunscreens with a sun protection factor of 15 or higher should be used. They should be applied at least 30 min before going out in the sun. UV-protected glasses should be worn. In this case, the treatment XP for this patient were natrium fusidic 2% cream, sunscreen SPF 30, moisturizer, chlorpheniramine maleate. Regular visits to a dermatologist and ophthalmologist are

vital for early detection and management. Due to the high likelihood of psychological stress and social withdrawal, a psychiatrist should also be involved as well as a genetic counselor. Patients must also be given Vitamin D supplements to compensate for strict sun avoidance.

## CONCLUSION

Early diagnosis, proper clinical examination, histopathological examination for the diagnosis of condition is must to come to proper diagnosis. Instructions to the patients to take proper protection from sun and wearing protective clothing must be given.

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**Authors Contribution:**

**SV-** Concept and design of the study, prepared first draft of manuscript, and interpretation; **PW-** Interpreted the results; reviewed the literature and manuscript preparation; **GP-** Concept, coordination, preparation of manuscript and revision of the manuscript

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