

## Keratoacanthoma in Xeroderma Pigmentosum: An Entity Beyond Guess

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### Abstract

Xeroderma pigmentosum (XP) is a rare autosomal recessive disorder, characterized by photosensitivity, pigmentary changes, premature skin aging and increase in risk of developing malignant neoplasms of both skin and eyes. Keratoacanthoma (KA) is a rapidly growing benign skin tumor, occurring primarily in elderly light skinned individuals. Here, we report the occurrence of KA of the nose with an unusual morphology in a 12 year old boy with XP where, such an association of the disease in young age is exceedingly rare.

**Keywords:** Xeroderma, Keratoacanthoma, Photosensitivity, Neoplasia

### Introduction

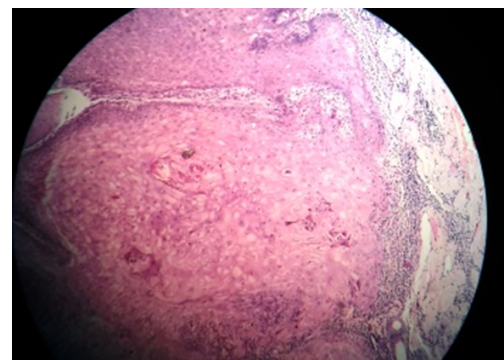
Xeroderma pigmentosum is a progressive, degenerative genodermatosis. Clinical manifestations include sensitivity to ultraviolet radiation resulting in inflammation and neoplasia in sun-exposed areas of the skin, mucous membranes, ocular surfaces and occasionally neurologic degeneration [1]. The basic defect lies in nucleotide excision repair, causing deficient repair of DNA damaged by UV radiation [2]. XP patients have a greater than 1000-fold increase in the incidence of sunlight-associated skin cancer [1]. Keratoacanthoma usually occurs on the sun-exposed areas in middle age and elderly individuals. It may be viewed as an aborted squamous cell carcinoma but rarely, may evolve into an overt malignancy.

### Case Report

A 12 year old boy, clinically diagnosed as a case of XP was brought to our department with complaints of a slowly enlarging asymptomatic raised lesion on dorsum of nose since one month. He had an asymptomatic skin lesion over right leg below knee, which was excised and diagnosed as dermatofibroma by histopathology one year back. History of first degree consanguinity was present for his parents. No other family members had XP. On examination, freckled pigmentation was present over face, ears, chest and conjunctiva. He had a firm, hyperpigmented plaque of size 3 × 2 cm on dorsum of nose, the surface of which showed a central crater with erythema (Figure 1). The lesion had minimal crusting and peripheral scaling. His scholastic performance was poor compared to the peer group. Ophthalmology



**Figure 1:** A 3 × 2 cm plaque with central crater, peripheral scaling and crusting on dorsum of nose.



**Figure 2:** Lobules of uniform squamous cells arising from epidermis and extending to dermis and neutrophilic microabscess. [H&E stain, 40x magnification].

and neurology evaluations were normal. Based on the above features, differential diagnosis of actinic keratosis, discoid lupus erythematosus, basal cell carcinoma and squamous cell carcinoma were considered for the lesion on nose.

Excision biopsy was done. Histopathology revealed lobules of uniform squamous cells arising from epidermis and extending to dermis. Cells in centre of lesion were large polygonal with pale pink cytoplasm and those in periphery were basaloid with neutrophilic microabscesses found within squamous cells (Figure 2). These findings were suggestive of Keratoacanthoma which was really unexpected for the lesion.

Child was advised to avoid sun exposure and to comply with regular sunscreens and sunglasses.

### Discussion

Xeroderma pigmentosum was first described in 1874 by Hebra and Kaposi [1]. In 1882, Kaposi coined the term Xeroderma Pigmentosum referring to its characteristic dry pigmented skin [2]. Upto 60% of persons

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with XP will eventually develop skin cancer, in many cases multiple tumours [3]. Actinic keratoses, warty papillomas, keratoacanthomas, fibromas, neurofibromas, angiofibromas and angiomyomas are the benign tumors reported [2]. The malignant neoplasias related to XP are basal cell carcinoma, squamous cell carcinoma, malignant melanoma, fibrosarcoma and angiosarcoma [4,5]. Though there have been a few reports of KA in adults with XP in literature, the exact incidence is not known. The mean age for KA in general population is 45 years [2]. Our case stands unique in virtue of the very early presentation of KA in this child.

Keratoacanthoma although common, remains an under reported tumor of the skin. It is a rapidly growing cutaneous neoplasia presumably arising from hair follicles, appearing as a dome-shaped nodule with a central keratin-filled crater which later degenerates into an involuting keratinous mass [2,6]. Chemicals, viruses like HPV, sunlight, trauma and altered immunity has been implicated as the causative factors amongst which, exposure to sunlight stands first. 95% of the solitary lesions are found on sun exposed areas of face, head and extremities [2].

In our case, the site of lesion and rapidity in progression favored the possibility of KA while, the age of onset and morphology mislead the diagnosis initially.

A presentation of this tumour in XP at a very young age was even reported earlier [1]. Unlike in our case, morphology of the tumour showing a well-defined dome shaped and rough mass on nose in the child was typical for a clinical diagnosis of KA in the prior report [1]. A plaque with central indentation is commonly a feature of DLE, actinic keratosis and cutaneous leishmaniasis. Hence, several differential diagnoses were thought of at the outset in our case. Histopathological features enabled us in exactly sketching the diagnosis as KA. Actinic keratosis, which is the most common tumour in patients with XP, was ruled out here because of a preserved granular layer, absence of parakeratosis, basal cell atypia and solar elastosis of dermis. Dearth of pleomorphic cells and keratinized squames in laminated layers ruled out squamous cell carcinoma as well. Anastomosing nests and cords of proliferating basaloid cells arising from basal layer of epidermis with palisading of nuclei distinctive of basal cell carcinoma were also absent [7].

Surgical excision is the preferred option in management of KA followed by modalities including intralesional injection of chemotherapeutic agents like methotrexate, 5-fluorouracil and Interferon alpha, lasers, cryotherapy, radiotherapy and photodynamic therapy [8]. Prophylactic treatment with oral isotretinoin can reduce the incidence of skin cancer in XP.

## Conclusion

Possibility of a Keratoacanthoma should be borne in mind for any patient with Xeroderma Pigmentosum having a skin tumor especially in sun exposed site, irrespective of the age. Histopathology has immense role in confirming the diagnosis, because clinical presentation at instances, may be perplexing as in our case. Regular medical follow up in these patients is crucial for an early diagnosis and treatment of tumors at a curable stage before they spread out or turn malignant.

## References

1. Al-Kamel MA (2016) Keratoacanthoma of the nose coexisted with xeroderma pigmentosum in a Yemeni child: A rare case. *Our Dermatol Online* 7: 419-421
2. Saawarn N, Saawarn S, Shahikanth MC, Chaithanya NCSK (2011) Keratoacanthoma with xeroderma pigmentosum. *International Journal of Dental Clinics* 3: 120-122.
3. Naik SM, Shenoy AM, Nanjundappa A, Halkud R, Chavan P, et al. (2013) Cutaneous malignancies in xeroderma pigmentosum: Earlier management improves survival. *Indian J Otolaryngol Head Neck Surg* 65: 162-167.
4. Shetty R, Girish BS, Baleel R, Permi HS, Makannavar P, et al. (2014) Xeroderma pigmentosa with multiple cutaneous malignancies: A rare case report and review of literature. *NUJHS* 3: 2249-71110.
5. Chowdhury RK, Padhi T, Das GS (2005) Keratoacanthoma of the conjunctiva complicating xeroderma pigmentosum. *Indian J Dermatol Venereol Leprol* 71: 430-431.
6. Schwartz RA (1979) The keratoacanthoma: A review. *J Surg Oncol* 12: 305-317.
7. Hussain I, Soni M, Khan BK, Khan MD (2011) Basal cell carcinoma presentation, histopathological features and correlation with clinical behaviour. *Pak J Ophthalmol* 27: 3-6.
8. Jeon HC, Choi M, Paik SH, Ahn CH, Park HS, et al. (2011) Treatment of keratoacanthoma with 5% imiquimod cream and review of the previous report. *Ann Dermatol* 23: 357-361.