



Albinism in India; Social Stigma and Undertreated Entity- A Review

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Abstract

Background: Albinism is autosomal recessive pigmentary disorder, characterized by lack of protective melanin, as a result of which albinos are highly susceptible to sun-induced damage to skin.¹ Persistent photosensitivity leads to development of various premalignant and malignant skin lesions. Awareness regarding strict sun protection and early detection of malignant lesions is necessary to prevent further complications.

Case Presentation: A 40-year-old male, who was a known case of Oculocutaneous albinism (OCA) presented to us with multiple yellowish thick papules and crusted lesion on sun exposed area. He was diagnosed to have Oculocutaneous albinism with actinic keratosis, with Bowen's disease (in situ squamous cell carcinomas). The case is reported to highlight social awareness and preventive aspects in the management of albinos.

Conclusion: Albino patients are more susceptible to develop malignancy; high index of suspicion is necessary for early diagnosis and treatment of premalignant lesion. In management physician should counsel patient and his family about nature of disease, pattern of inheritance and need for strict sun protection measures.

Keywords: Albinism; Actinic keratosis; Bowen's disease; Social stigma.

Introduction

Albinism is inability to synthesize melanin in the skin and the eyes. There are two types of albinism; ocular albinism and Oculocutaneous albinism (OCA), Oculocutaneous albinism (OCA) is the most common inherited disorder of pigmentation, with an estimated frequency of 1: 20,000 in most populations.¹

Skin manifestation of Oculocutaneous albinism is sun burns, premalignant skin lesions, melanoma and non melanoma skin cancers and most is actinic keratoses predisposing to squamous cell carcinoma.² Previous studies has explained

correlation between exposure to sun and the incidence of the above cutaneous problems in Oculocutaneous albinism.¹

In India social stigma is associated with any pigmentary disorder, thus albino with premalignant lesions present in later stage to medical facilities, which increases the incidence of advanced malignancy.

Case Presentation

A 40 year old male born of non consanguineous marriage presented to dermatology department with complaints of yellowish thick rough papules

present on sun exposed area since 4-5 years and 1 month back he noticed sudden increase in size of pre-existing lesion present on forehead which was associated with pain and intermittent bleeding. He also gave a history of photosensitivity and presence of white hair since birth.

On examination Patient had typical features of cutaneous albinism in the form of totally depigmented skin with white hairs. Ocular examination was within normal limits. He had 4-5 keratotic papular and plaque lesions of 2mm to 1cm in size on vertex of scalp [Figure 1] with background erythema (actinic keratosis). Besides this, he had single crusted lesion with



Figure 1

seosanguinous discharge size 2 x 3 cm on forehead. [Figure 2]

No family history of similar complaints was present.

Routine investigations were within normal limits. Skin biopsy showed part of epidermis shows full thickness atypia of cells with large pleomorphic nuclei and loss of polarity of cells. Dermis showed solar elastosis and inflammatory cell infiltrate.

On the basis of these clinical findings and biopsy report a diagnosis of Oculocutaneous albinism with actinic keratosis with Bowen's disease was made.



Figure 2

Discussion

Albinism is inherited pigmentation disorder with impaired melanin synthesis, resulting in universal absence of eumelanin in the skin and hair follicles. When the eyes are also affected, it is called Oculocutaneous albinism (OCA), eye involvement can vary from complete blindness to nearly undetectable visual defect, strabismus, and nystagmus.³ Melanocyte provides important defense mechanism against UV rays. Therefore albinos are more prone to develop deleterious

effects of UV light on their skin such as melanoma and non-melanoma skin cancers.^{4,5}

Excessive sun exposure can result in various premalignant skin lesions like actinic keratosis, actinic cheilitis which progress to invasive malignancy. These lesions are commonly found on sites of sun-exposed skin such as the face, bald scalp, and dorsum of the hand.^{6,7} Actinic keratosis are irregular rough, scaly brown to skin colored hyperkeratosis papules or plaques. Microscopic examination of actinic keratosis shows dysplastic

keratinocytes which is initial step of UVB induced cascade of events. These altered keratinocyte may remain stable for years together but further sun exposure can bring about clonal expansion of activated keratinocyte.⁸ If left untreated the risk of progression of sunlight keratosis to invasive SCC can be as high as 20% per year.⁹

In addition photosensitivity is present in almost every albinism patient, thus constant local inflammation derived free radicals leads to DNA damage and defect in repair mechanism.¹⁰ Various malignancy of epithelium and adenexal structure is reported in Indian albino patients.¹¹ Bowens disease is carcinoma in situ, an epithelial neoplasm; and 3-5% cases of can progress to invasive malignancy.¹²

The ideal objectives of cancer management for any cancer should be reduction of the incidence, early detection, and prompt treatment of the disease when it occurs. Albino patient should take universal precautions against sunlight exposure in the form of protective clothing to cover as much of the skin as possible, and the use of broad spectrum sunscreen, lifestyle modification such as minimizing of outdoor activities during peak sunlight hours from early childhood.⁹

In developing countries like India public health measures are targeted to solve basic needs thus preventive measures to minimize sunlight-induced damage are unsuccessful. Due to illiteracy, knowledge for basic understanding of problem and associated precaution is lacking.¹³ In addition Indian population with pigmentary disturbance suffer from social discrimination because of superstitious beliefs and the stigma associated with it.¹⁴ Also these patients have less chances of getting a job at interview leading to unemployment. Therefore albino patients report to medical facilities often in later stage, which ultimately lead to delayed management and complication of premalignant lesion.

Conclusion

Regular examination of all albinos for early detection and treatment of the various malignant

lesions. Patients should be educated for preventive sun protection measures and early signs of any new developing lesions. Psychological and genetic counseling should be included as a part of treatment.

References

1. Lookingbill DP, Lookingbill GL, Leppard B. Actinic damage and skin cancer in albinos in northern Tanzania. *J Am Acad Dermatol* 1995;32:653-8.
2. Schulze KE, Rapini RP, Duvic M. Malignant melanoma in oculocutaneous albinism. *Arch Dermatol* 1989;125:1583-5.
3. King RA, Summers CG. Albinism. *Dermatologic clinics*. 1988;6:217-28.
4. Kromberg JG, Castle D, Zwane EM, Jenkins T. Albinism and skin cancer in Southern Africa. *Clinical genetics*. 1989;36:43-52.
5. Chatterjee K, Rasool F, Chaudhuri A, Chatterjee G, Sehgal VN, Singh N. Basal cell carcinoma, oculo-cutaneous albinism and actinic keratosis in a native Indian. *Indian journal of dermatology*. 2013;58:377.
6. Rosen RH, Studniber H. Solar keratoses: Analysis in a dermatological practice in Australia. *Australas J Dermatol* 2003;44:34-9.
7. Schwartz RA, Bridges TM, Butani AK, Ehrlich A. Actinic keratoses: an occupational and environmental disorder. *Eur Acad Dermatol Venerol* 2008;22:606-15
8. Cockerell CJ. Pathology and pathobiology of the actinic (solar) keratosis. *Br J Dermatol* 2003;149:34-6.
9. Chiller KG, Washington C, Sober AJ, Koh HK. Cancer of the skin. In: Kasper DL, Braunwald E, Fauci AS, Hauser SL, Longo DL, Jameson JL, eds. *Harrison's Principles of Internal Medicine*. 16th ed. New York: McGraw-Hill 2005, pp. 497-503

10. L. Feller, M. Altini, and J. Lemmer, "Inflammation in the context of oral cancer," *Oral Oncology*, vol.49, no.9, pp.887–892, 2013.
11. Hanumanthappa H. Eccrine gland adenocarcinoma in albinism. *Indian Journal of Dermatology, Venereology, and Leprology*.1994;60:110.
12. Kao GF. Carcinoma arising in Bowen's disease. *Arch Dermatol*.1986;122:1124–6.
13. Ali I, Wani WA, Saleem K. Cancer scenario in India with future perspectives. *Cancer therapy*. 2011;8:56-70.
14. Shah H, Mehta A, Astik B. Clinical and sociodemographic study of vitiligo. *Indian Journal of Dermatology, Venereology, and Leprology*. 2008 Nov 1;74(6):701.