International Journal of Scientific Reports
Bhargava S et al. Int J Sci Rep. 2018 Feb;4(2):40-43
http://www.sci-rep.com
pISSN 2454-2156 | eISSN 2454-2164

DOI: http://dx.doi.org/10.18203/issn.2454-2156.IntJSciRep20180397

Case Report

Premalignant and malignant changes of skin in a patient with oculocutaneous albinism: multiple actinic keratosis and squamous cell carcinoma

Shashank Bhargava*, Ujjwal Kumar, Richa Rokde

Department of Dermatology, R. D. Gardi Medical College, Ujjain, Madhya Pradesh, India

Received: 07 December 2017
Accepted: 11 January 2018

***Correspondence:**
Dr. Shashank Bhargava,
E-mail: shashank2811@gmail.com

ABSTRACT

Pigmentation of skin is a feature which is governed by multiple factors including the number of melanocytes, their metabolic activity of the melanocytes, the melanogenic activity of the melanosomes and lastly the morphology and differentiation of the melanosomes "Squamous cell carcinoma" (SCC) of sun-exposed skin is the most frequently observed malignancy among Albinos. It is ultimately due to lack of Eumelanin which guards against both the sunlight as well as oxidative stress-induced DNA damage. A 41 year old albino male patient presented with multiple asymptomatic raised skin lesions of different morphology and dimensions over scalp, neck, behind right ear and back with duration of 7 years. The initial lesion developed as a small papule over scalp which gradually progressed to present size and later appeared before the right ear, neck and lastly over the back. On examination, the initial lesion over scalp was verrucous plaque with adherent crusting, while other lesions were indurated, non-tender ulcers with irregular margins and punched out edges. There are few whitish raised scaly papules and plaques over the back. Routine investigations were within normal limits. Histopathology findings from the neck revealed squamous epithelium with tumour cells infiltrating the underlying stroma. Tumour cells showed pleomorphism, increased N:C ratio, hyperchromatic nuclei, prominent nucleoli and keratin pearls. Histopathology findings from the scaly lesion over the back revealed mild hyperkeratosis with dysplasia of the basal keratinocytes and prominent solar elastosis in the superficial dermis. Based on clinical and histopathogical findings, diagnosis of multiple SCC with actinic keratosis was made. Patient was referred to oncosurgeon for further management. Early detection and prompt treatment of the disease is required to reduce the spread to other parts of the body along with photo-protection all throughout life.

Keywords: Oculocutaneous albinism, Squamous cell carcinoma, Actinic keratosis

INTRODUCTION

Fitzpatrick categorized various skin color into types I-VI depending on the pigmentation of skin in increasing order.[1] Pigmentation of skin is a variable feature which is governed by multiple factors including the number of melanocytes, their metabolic activity of the melanocytes, the melanogenic activity of the melanosomes and lastly the morphology and differentiation of the melanosomes. The predominant types of melanin namely eumelanin or pheomelanin also determine the pigmentation of skin and in turn the appearance. Eumelanin has a protective role from sunlight mainly ultraviolet radiations while pheomelanin does not have this quality. Melanocortin-1 receptor (MC1R) on the melanocytes and its ligand, α-melanocyte stimulating hormone (αMSH), activity of tyrosinase in the basal layer, tyrosinase related protein 1 (TRP1), and transport proteins are the factors which

ultimately determine melanin synthesis.[2] Migration of precursor melanocytes to skin and eye is under a number of genes which control the differentiation and proliferation of neural Crest cells. The degree of pigmentation of the skin is said to correlate inversely with the risk of sun-induced skin cancers.[3] Oculocutaneous albinism (OCA) is a rare autosomal recessive inherited pigmentary disorder of skin and hair associated with ophthalmological anomalies. They have normal number and distribution of melanocytes. It is due to impaired melanin biosynthesis, defects in melanosome biogenesis or dysregulation of intracellular transport proteins required for melanin production. Ocular change can manifest in reduced visual acuity and nystagmus.[4] Squamous cell carcinoma (SCC) of sun-exposed skin is the most frequently observed malignancy among Albinos. It is ultimately due to lack of protective effect of Eumelanin which guards against both the sunlight as well as oxidative stress-induced DNA damage. The risk of occurrence of SCC is about 1000-fold higher in black albinos in sub-Saharan Africa than the general population.

CASE REPORT

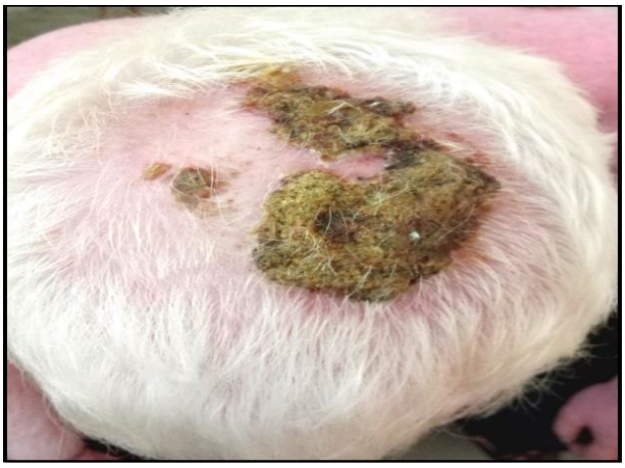

Figure 1: Multiple vegetative plaques over the scalp with few scaly lesions around them.

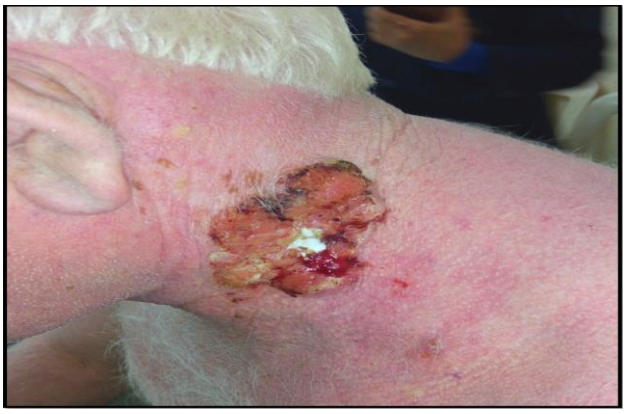

Figure 2: An ulcer over the neck with irregular border and slight bleeding.

A 41 year old albino male patient presented at the dermatology OPD with multiple asymptomatic raised skin lesions of different morphology and dimensions over scalp, neck, behind right ear and back with duration of 7 years. The initial lesion developed as a small ulcer over scalp which gradually progressed to present size and later appeared before the right ear, neck and lastly over the back. He had photosensitivity and decreased visual acuity. On examination, the initial lesion over scalp was verrucous plaque with adherent crusting, while other lesions were indurated, non-tender ulcers with irregular margins and punched out edges, gradually increasing in size (Figure 1-3). Right cervical lymph nodes were enlarged. There are few whitish raised scaly papules and plaques over the back. Routine investigations like Haemogram, random blood sugar level, RFT and LFT were within normal limits.

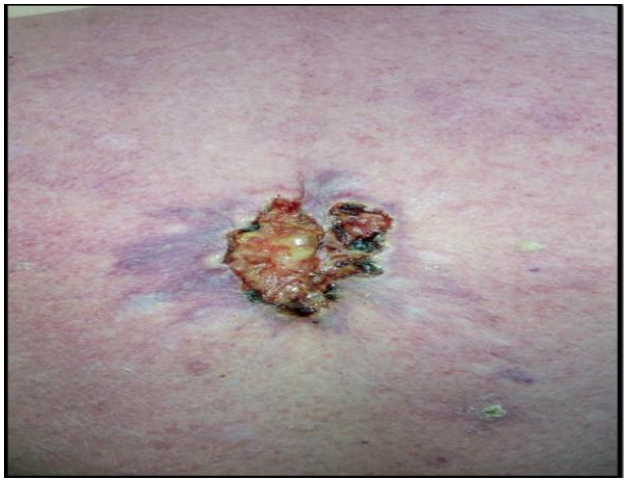

Figure 3: An ulcer with few scaly lesions of actinic keratosis over back.

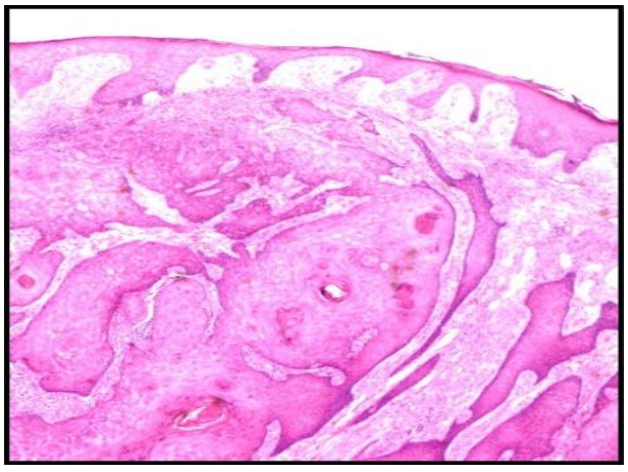

Figure 4: Histopathology findings revealed squamous epithelium with tumour cells infiltrating the underlying stroma. Tumour cells showed pleomorphism, increased N:C ratio, hyperchromatic nuclei, prominent nucleoli, moderate amount of eosinophillic cytoplasm and keratin pearls.

Histopathology findings from the scalp revealed squamous epithelium with tumour cells infiltrating the underlying stroma. Tumour cells showed pleomorphism, increased N:C ratio, hyperchromatic nuclei, prominent nucleoli, moderate amount of eosinophillic cytoplasm and keratin pearls (Figure 4). Histopathology findings from the scaly lesion over the back revealed mild hyperkeratosis with dysplasia of the basal keratinocytes and formation of small buds extending into the papillary dermis with prominent solar elastosis in the superficial dermis. The dysplastic changes primarily affect the epidermis between follicles (Figure 5). On the basis of clinical and histopathogical findings, diagnosis of multiple squamous cell carcinoma with actinic keratosis was made. Patient was referred to oncosurgeon for further management.

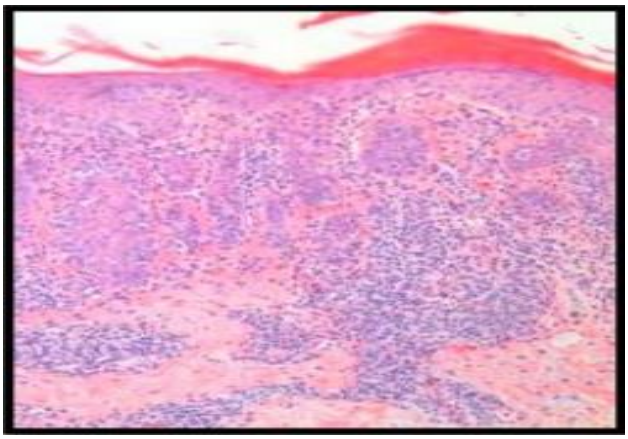

Figure 5: Histopathology findings from the scaly lesion over the back revealed mild hyperkeratosis with dysplasia of the basal keratinocytes and formation of small buds extending into the papillary dermis with prominent solar elastosis in the superficial dermis.

DISCUSSION

OCA predisposes to squamous cell carcinoma of the skin (SCC), particularly of the sun-exposed head and neck.[5] SCC is more frequent and runs a more aggressive course. OCA type 1 (OCA1), the second most common type of OCA occurs with a frequency of about 1/40,000 worldwide which is characterized by loss of function of the enzyme tyrosinase (TYR) as a result of a mutation in the TYR gene. OCA1A and OCA1B have completely nonfunctional and partial functional activity TYR gene respectively.[6] OCA2 is the most common form of albinism worldwide, it affects Blacks more commonly than Whites. It is due to mutations in the OCA2 gene (formerly known as the P gene) that encodes the p protein. Transporting proteins to the melanosome, stabilizing the melanosomal protein complex, and in regulating melanosomal pH are controlled by p protein.[7,8] Other types of OCA are OCA3, OCA4 and OCA5 which are not seen very commonly. Recently OCA5 was detected in members of a Pakistan family due to consanguinity.[9]

Eumelanin (brown melanin) provides protection both against sunlight and against oxidative stress-induced DNA damage. In light-skinned persons, the extent of the sunlight-induced DNA damage exceeds the capacity of cellular DNA repair mechanisms with increases risk of malignant transformation.[6] Hence effective functioning cellular DNA repair mechanisms are more important in preventing SCC than the amount of melanin pigment in sun-exposed skin and thats the reason for xeroderma pigmentosa to occur in both dark and light skinned individuals due to defect in DNA repair mechanisms. During synthesis of pheomelanin, reactive oxygen species (ROS) are generated which are carcinogenic. Therefore reduction in eumelanin responsible for photoprotection and elevation of pheomelanin-derived ROS are culprits for malignancies in albinos.[4] Albinos acquire SCC of skin de novo or from transformation of premalignant lesions like actinic keratosis which is due to damage of basal cell keratinocytes due to intense and prolong sun exposure. Genomic instability due to mutation in p53 Tumor suppressor gene predisposes the initially transformed keratinocytes to additional genetic alterations leading to clonal expansion of keratinocytes which subsequently lead to SCC of skin. Smaller and frequent exposures to sunlight are more likely to cause malignant transformation than greater but infrequent exposures, because each exposure has the potential to cause a genetic change.[10] Sunlight-induced local inflammation in the skin is another reason to enhance proliferation of basal keratinocytes.

CONCLUSION

Early detection and prompt treatment of the disease is required to reduce the spread to other parts of the body. Minimizing outdoor activities, universal precautions for sunlight and use of sunscreen right from early childhood, continued throughout the life and also after the surgery can help and protect Albinos from skin malignancies.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Fitzpatrick TB. The validity and practicality of sun-reactive skin types I through VI. Arch Dermatol. 1988;124:869-71.
2. Feller L, Masilana A, Khammissa RA, Altini M, Jadwat Y, Lemmer J. Melanin: the biophysiology of oral melanocytes and physiological oral pigmentation. Head Face Med. 2014;10:8.
3. Manga P, Kerr R, Ramsay M, Kromberg JG. Biology and genetics of oculocutaneous albinism and vitiligo - common pigmentation disorders in southern Africa. S Afr Med J. 2013;103:984-8.

4. de Vijlder HC, de Vijlder JJ, Neumann HA. Oculocutaneous albinism and skin cancer risk. J Eur Acad Dermatol Venereol. 2013;27:433-4.
5. Mapurisa G, Masamba L. Locally advanced skin cancer in an albino, a treatment dilemma. Malawi Med J. 2010;22:122–3.
6. Wood NH, Khammissa R, Meyerov R, Lemmer J, Feller L. Actinic Cheilitis: A Case Report and a Review of the Literature. Eur J Dent. 2011;5:101–6.
7. Nikolaou V, Stratigos AJ, Tsao H. Hereditary Nonmelanoma Skin Cancer. Semin Cutan Med Surg. 2012;31:204–10.
8. Hawkes JE, Cassidy PB, Manga P, Boissy RE, Goldgar D, Cannon-Albright L, et al. Report of a novel OCA2 gene mutation and an investigation of OCA2 variants on melanoma risk in a familial melanoma pedigree. J Dermatol Sci. 2013;69:30-7.
9. Kausar T, Bhatti MA, Ali M, Shaikh RS, Ahmed ZM. OCA5, a novel locus for non-syndromic oculocutaneous albinism, maps to chromosome 4q24. Clin Genet. 2013;84:91-3.
10. Andreassi L. UV exposure as a risk factor for skin cancer. Expert Rev Dermatol. 2011;6:445-54.

Cite this article as: Bhargava S, Kumar U, Rokde R. Premalignant and malignant changes of skin in a patient with oculocutaneous albinism: multiple actinic keratosis and squamous cell carcinoma. Int J Sci Rep 2018;4(2):40-3.