Abstract: Albinism is inherited as an autosomal recessive disorder, characterized by lack of skin pigment melanin, as a result of which albinos are highly susceptible to sun-induced damage to skin. Previous studies have documented a high frequency of actinic keratoses and skin cancers in albinism patients. Here we report a 45 year old male patient a known case of albinism presented with complaints of non-healing ulcer over right leg for 1 year duration. Biopsy of the lesion revealed squamous cell carcinoma moderately differentiated. Patient has a similar history of lesion over right ear for which he was operated which revealed similar histology. The case is reported to highlight preventive aspects in the management of albinos.

Keyword: Albinism, Squamous cell carcinoma, Actinic keratoses

INTRODUCTION: Albinism is a common inherited disorder which is acquired through autosomal recessive pattern. It is a premalignant condition associated with absence of melanin pigments in the skin due to absence or defect in the enzyme tyrosinase which is essential for melanin synthesis which is a photosensitive pigment that protects the skin from the harmful effects of UV radiation. The estimated incidence is 1 in 20,000 in most parts of the world, highest among cuna Indians around 6.3 /1000. In India the incidence being 1 in 17,000. The cutaneous problems seen with albinism are sun burns, basal cell carcinoma, malignant melanoma, dysplastic nevus syndrome and, most important and most common of all, actinic keratoses predisposing to squamous cell carcinoma. Of these the most common being squamous cell carcinoma however the recurrent nature is a rare entity.

CASE REPORT: A 45 year old male patient presented with complaints of non-healing ulcer in the right leg for 1 year duration. Patient has a significant past history of similar lesion in his right ear for which he underwent surgery 20 years back. Patient underwent surgery for similar lesion( chronic non healing ulcer) over left knee joint , four years back. Histopathological examination revealed squamous cell carcinoma. Patient also has history of keratotic growths over the trunk and limbs, of one and a half years’ duration. He also gave a history of photosensitivity and presence of white hair since birth. His family history is significant, as multiple family members (two elder brothers and one elder sister) suffering from similar skin condition (Albinism). Out of these three siblings, only one (first elder brother) developed skin cancer and treated with surgery; but he died of some cardiac problem later. On physical examination, he had typical features of albinism in the form of totally depigmented skin with white hairs. His iris was non-pigmented and the pupillary area looked red. He had multiple keratotic papular and plaque lesions of 2mm to 2cm in size that were round to oval in shape, surrounded by a zone of erythema (actinic keratoses). A 2 x 2 cm lesion with ulcerated base and scalloped edge over the medial aspect of the right leg 6 cms above the ankle joint, similar lesion of 4 x 4 cm ulcerated lesion with excavated margins and irregular margins over the left leg 5 cms above the left ankle joint. A nodular lesion with central umbilication arising from the skin over the epigastic region. Besides this, he had ulcerated and crusted lesions all over the body which were predominantly over the trunk and limbs.

On the basis of these clinical findings, a diagnosis of albinism with actinic keratoses with multiple squamous cell carcinomas and cutaneous horns was made. Dermatologist opinion was obtained. Diagnosis of albinism with multiple squamous cell carcinoma and keratoacanthoma was made. Biopsy from the skin lesion taken and sent for histopathological examination.
The base line investigations showed hemoglobin of 12 gm%. His total leukocyte count, differential leukocyte count, and platelet counts were within normal limits. His blood sugar, liver and renal function tests were with in normal limits. Biopsy from the lesion showed moderately differentiated squamous cell carcinoma of the skin. His metastatic work-up with CT scan of chest and abdomen turned to be negative. He was treated with chemotherapy consists of cisplatin and 5-FU after metastatic workup turned to be negative. Patient was deferred RT and topical 5-FU in view of multiplicity of the lesions.

**DISCUSSION:**

Among the most common inherited disorder of general hypopigmentation, with an estimated frequency of 1: 20,000, in most populations. There are several types of albinism. Tyrosinase-related OCA (OCA1), one of the two most common types of albinism, produced by loss of function of the melanocytic enzyme tyrosinase, resulting from mutations of the tyrosinase gene. Null mutations result in total loss of function and, therefore, some pigment formation is seen (OCA1B). A typical feature of OCA1 is ‘white hair’ at birth.

From the clinical features, it was obvious that the patient fell under the subtype OCA1A or classic tyrosinase-negative OCA resulting in the characteristic ‘albino’ phenotype. In albinos lack of melanin, a photo protective pigment against the harmful UV radiation, predisposes to actinic keratoses and, thereby, to squamous cell carcinomas. Males develop actinic keratoses more frequently than females. They usually occur on sun exposed areas. Actinic keratoses is a clinical manifestation of UV radiation (most commonly UV-B radiation) induced neoplastic transformation of keratinocytes.

The UV-B radiation causes thymidine dimer formation in DNA and RNA, resulting in mutations causing neoplastic changes in keratinocytes. The two important sites of mutations taking place in actinic keratoses formation are in telomerase and the tumor suppressor gene p53, located on chromosome 17p132. These mutant DNA cells, resistant to apoptotic death, undergo clonal expansion and accumulate genetic injury, resulting eventually in neoplastic transformation. Studies have shown that up to 60% of the squamous cell carcinomas begin as actinic keratoses and that there is histologic evidence of contiguous actinic keratoses in 97% of the squamous cell carcinoma lesions that arise on sun damaged skin. The likelihood of a fully developed squamous cell carcinoma evolving from a given actinic keratosis has been estimated to occur at a rate of 0.075- 0.096% per lesion per year. Thus, an affected individual, with an average number of 7.7 actinic keratoses on his skin, could expect to develop squamous cell carcinoma at a rate of 10.2% over 10 years. Other sources give even higher estimates, with rates of 13-20% of such individuals developing squamous cell carcinoma over a 10-year period and with the albinos being even more vulnerable, due to lack of photo protective pigment. The five-year rate of recurrence of primary cutaneous lesions is 8 percent, and the five-year rate of metastasis is 5 percent. Large lesions (>2 cm in diameter) recur at a rate of 15 percent, which is twice that of smaller lesions, and they metastasize at a rate of 30 percent, three times that of smaller lesions. Squamous-cell carcinomas of the lip and ear are also aggressive lesions, with rates of recurrence and metastasis ranging from 10 to 25 percent. Squamous-cell carcinomas arising in injured or chronically diseased skin are associated with a risk of metastasis that approaches 40 percent.

Other clinical features associated with recurrence and metastasis include rapid growth and local recurrence of the tumor, as well as immunosuppression. A complete history taking and physical examination are indispensable. Information should be elicited about sun exposure beginning in childhood, occupational exposure to ultraviolet light or carcinogenic chemicals, previous radiation treatment, and potential causes of immunosuppression. If the patient has a history of skin cancer, the type, location, and dates of treatment should be noted. A total-body examination of the skin is the only screening test available for cutaneous squamous-cell carcinoma. Since early disease is highly curable and metastatic disease carries a grim prognosis, prompt detection is potentially lifesaving. Particularly among patients who have previously had skin tumors, screening is necessary to monitor for recurrence or persistence of the tumors and for the presence of new lesions. There is a 30 percent risk of having a second primary squamous-cell carcinoma within five years after treatment for the first tumor. Our patient had recurrence within 5 years. Since Mohs’ surgery are more costly but can offer significantly lower rates of recurrence and metastasis for patients with high-risk tumors. Treatment of nodal disease may involve radiation, lymph-node dissection, or both. Fractionated radiation treatment may be preferred for patients who are unable to tolerate surgery or who have inoperable tumors and may provide favorable functional and cosmetic results. Photodynamic therapy may also be used as a treatment modality for patients who are unable to tolerate surgery or other methods of therapy to treat aggressive or recurrent lesions. The use of systemic therapy for advanced and recurrent disease is well established with questionable outcome however the use of cisplatin with SFU or other agents like bleomycin remains a corner stone in the management of advanced and recurrent disease.

**REFERENCES:**


