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Squamous cell Carcinoma of the Eye in a Xeroderma pigmentosum Child

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Abstract: Xeroderma pigmentosum (xp) is a rare autosomal recessive disorder, characterized by photosensitivity, pigmentary changes, premature skin aging and marked increase in risk of developing malignant tumor development. We report a case of squamous cell carcinoma of the eye in a xeroderma pigmentosum child.
Keywords: ocular manifestation, morning glory syndrome.

INTRODUCTION

Xeroderma pigmentosum is a hereditary autosomal recessive disorder, characterized by mucocutaneous and ocular hypersensitivity to ultraviolet radiation. Herba and Kaposi first described XP in 1974 [1]. Xeroderma pigmentosum occurs with an estimated frequency of 1: 25000 in United States and Europe. In Japan, it is 1 case per 40,000 populations [2]. In general the signs and symptoms of Xeroderma Pigmentosum starts from the age of 1-2 years. The disease begins with photosensitivity and burning sensation after normal sun exposure in 60% cases. Cutaneous manifestation include dryness of skin, pigmentation, freckling and telangiectasis. Occular abnormalities include photophobia, ectropion, conjunctival infection, keratitis with incidence of tumors like squamous cell carcinoma, melanoma and epithelioma. There is 1000 fold increased risk of skin malignancy on sun exposed sites [3]. Chronic sun exposure causes marked alterations in the skin leading to keratosis, telangiectasia, atrophy and development of malignant tumors like squamous cell carcinomas, basal cell carcinoma, and malignant melanoma [4].

CASE REPORT

A 9-year-old boy referred to our office by a dermatologist. On admission, the chief complaint was burning sensation and photophobia both eyes. His history revealed the gradual onset of dark spots on the face at the age of 5 years, and then they spread all over his body. No positive family history with similar condition was found. The child is the third issue of his parents, other siblings are healthy. General physical examination found multiple blackish spots spread all over the body with a dry and rough skin. On ophthalmic examination, the child had blepharospasm rendering the passive opening of the eyes difficult. Eyelids were pigmented and the conjunctiva was congested both eyes. On the right eye, there was a bullous lesion of whitish appearance at inferior bulbar conjunctiva. The lesion was excised under general anesthesia. Histopathological examination of the excised tissue revealed features of a well differentiated squamous cell carcinoma. Before discharging the child, her parents were given thorough explanations about the disease and the necessity to avoid sun exposure and the wearing of tinted glasses as well as clothes that covers almost the whole body. A regular follow up during one year noticed no particularity.

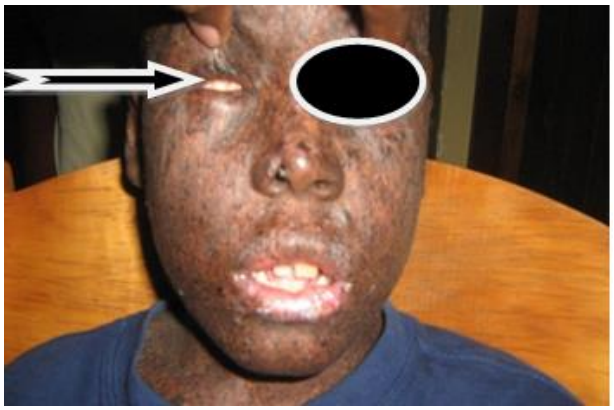

Fig-1: Photograph of the child with right eye squamous cell carcinoma

DISCUSSION

Squamous cell carcinoma of conjunctiva is an invasive tumor showing keratinocytic differentiation [5]; it occurs chiefly at limbus and spread over surface

and into fornices rarely penetrate the globe. Patient with xeroderma pigmentosum have a severe sensitivity to all sources of ultraviolet (UV) radiation, especially sunlight, and they developed serious sunburns with onset of poikiloderma in light-exposed skin [6]. Although the etiology of squamous cell carcinoma is not well known, exposition to ultraviolet radiation has been suggested as a possible risk factor. Squamous cell carcinoma is common in patients who are genetically more susceptible to sunlight, particularly those with Xeroderma pigmentosum [7]. A variety of tumors like basal cell carcinoma, squamous cell carcinoma, angiosarcoma and keratoacanthoma have been reported with XP with rare association with melanoma and fibrosarcoma. Ocular involvement is seen in 80% of the cases [8]. Our patient hailed from a poor rural family where the main activity is farming under bright and burning sunlight. The long exposure to sunlight is prejudicial to the health of people with xeroderma pigmentosum. Xeroderma pigmentosum is transmitted in most cases as an autosomal recessive manner but in our case, the pedigree did not reveal a family history.

Commonly, later in childhood dysplastic and neoplastic lesions occur with the development of actinic keratosis, keratocanthoma, basal cell carcinoma, squamous cell carcinoma and malignant melanoma [9].

Clinical management of XP includes avoidance of sunlight, minimizing UV and cigarette smoke exposure, early excision of skin lesions, and genetic counseling.

CONCLUSION

Patients with xeroderma pigmentosum are very sensitive to sunlight and are very likely to develop malignancies like squamous cell carcinoma. They deserve a cautious protection against direct sunlight. Early medical follow up can help to diagnose tumors at a curable stage before they spread out.

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