CASE REPORT

A THREE YEAR STUDY OF SKIN CANCER IN A CASE WITH XERODERMA PIGMENTOSUM

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INTRODUCTION: Xeroderma pigmentosa was first described by Hebra and Kaposis.[1] It's a rare disorder transmitted by autosomal recessive manner.[2,3] Xeroderma characterized by dry, pigmented skin lesions resultant of severe sensitivity to UV radiation from sun exposure.[4,5,6] Main defect is inability to repair the DNA damage.[5] The prevalence is at 1: 1,000,000, the effects on skin is cumulative and irreversible.[1] There is 1000 fold increase in development of skin cancers, precancerous lesions of mouth and eye.[7,8] We are here presenting a case of xeroderma pigmentosa with skin cancer and its management and follow up.

CASE PROFILE: The study involved a family of three brothers and one sister. All three brothers are normal, sister suffers from xeroderma pigmentosum. The parents are normal with such history on the mother's side only. These three siblings are 11, 8 and 6 years of age, sister is elderly, she is 14 years of age. Three years back the eldest developed a mole which was excised. Subsequently she developed squamous cell carcinoma of fore head near the nose bridge. There are many precancerous lesions on the face and other areas of the exposed skin are about 6 year's duration as multiple skin lesions and keratotic patches. Since this condition is of very rare occurrence especially in India and the incidence of skin cancers is much earlier in these cases as compared to the usual ages at presentation of cancers, hence this rare familial disorder is being presented. The other relatives of the patient are being followed very closely to observe for subsequent skin disorders.

The girl has under gone wide excision and a forehead flap reconstruction based on supra orbital vessels. Surgery was conducted three years ago and patient was followed up since then, with regular monthly intervals. She was on sun protection measures and regular follow up. There is no evidence of recurrence and new cancers.

DISCUSSION: Xeroderma pigmentosum is a rare autosomal recessive neurocutaneous disorder caused by inability to repair DNA damage produced by ultraviolet radiations.[4,2,3,5,6] A variety of defects happen but mainly there is proneness to skin cancers of all types and most sufferers succumb to metastatic disease at early age.[7,8,9] Surgical excision is the principal treatment for these skin cancers.[5,10] Lasers play is an important role in the treatment of premalignant lesions on superficial areas with co2 laser silk touch ablation mode.[9,10,11]

There are three stages of xeroderma pigmentosa,[2,8,12,13] the first stage, exposure to sun at the age of 6 months, develops dry, scaly, pigmented lesions histologically as hyperkeratosis and increased melanin pigment.

The second stage, characterized by poikloderrma with dry skin, telangiectasia, mottled hyper and hypo pigmentation and hyperkeratosis.

The third stage, where complications manifest. Squamous cell carcinoma, malignant melanoma, basal cell carcinoma and fibrosarcoma skin are common.[2,6,9,12,13,14]

The radiation treatment induced damage unlike UV damage is very much reparable and hence radiotherapy has been used where surgery is not feasible.[15] The type of radiation has to be customized to the individual and external beam, electrons or mould therapy may be required.

The goal of treatment is to protect from sun light exposure by sun screen lotions and protective covers all over the body. The treatment is customized according to the stage of decease and individualized.[1,10,11]

CONCLUSION: The xeroderma pigmentosa is rare autosomal recessive disorder presenting with dry scaly pigmented skin lesions with increased incidence of skin cancers especially melanoma and squamous cell carcinoma. Treatment is surgical excision and lasers. Skin protection from sun exposure is the main preventive measure. Our case presented with squamous cell carcinoma and we treated the case as per the guidelines and took preventive measures.

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