

Xeroderma Pigmentosum with Multiple Malignant Lesions, Present in Siblings Linking it to Genetic Aspect of the Disease and Possible Treatments

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Department of Radiation Oncology, Gujarat Cancer and

Corresponding author: Radiation Oncology

Tel:8347281991; E-mail:dipenmistry57555@gmail.com

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Abstract

Being a Geno dermatosis, photosensitive disorder Xeroderma Pigmentosum (XP) is a rare autosomal recessive disorder. Patient with XP commonly presented with an interactable and dermatological disease. Patients with XP are prone to cutaneous, mucosal malignancies and ophthalmic complications. Reason being inability to repair the Ultra Violet (UV) radiation dispensed damage to Deoxyribonucleic Acid (DNA), genetic nature of disease is revealed. Not only genetic but environmental factor plays big role in course of disease. In XP cutaneous and mucosal Basal Cell Carcinoma (BCC) and Squamous Cell Carcinoma (SCC) or melanoma is most common.

Keywords: Xeroderma pigmentosum; Deoxyribonucleic Acid (DNA); Basal cell carcinoma; Squamous cell carcinoma

Introduction

XP characterized with sever sunburns, development of many freckles at early age, rough surfaced wart like growths, cutaneous and mucosal malignancies, blistering, telangiectasia's, ophthalmic manifestations, limited growth of hair on chest and legs and dry skin. 45% to 60% patients of XP were observed to be developing malignant skin neoplasm in different studies [1]. Compare to normal controls Robbins has shown that incident of internal neoplasm in XP patient was 10-20 times higher. Angiosarcoma and Fibrosarcomas have also been reported. In XP eye tissues are also exposed to UV and cause lesion mainly in anterior ocular segment, such as conjunctival xerosis, corneal drying, decrease of lacrimation, corneal neovascularization,

Younger brother a 9-year old male child presented initially with swellings on lip, scalp, face and multiple hyper pigmented skin lesions over whole body as a diagnosed case of Xeroderma pigmentosa. Further investigation revealed squamous cell carcinoma right side of cheek, pyogenic granuloma in lip lesion and no evidence of malignancy from scalp lesion then further taken chemotherapy for response computed tomography done which shows lesions on skin of cheek on both side and nose with soft tissue thickening on tip of tongue but pathologically no evidence of malignancy on tip of tongue. Then taken multiple chemotherapy regimen. For response assessment computed tomography was done which shows increase in size of previous lesion. Another regimen of chemotherapy taken as further line of treatment. Further workup done computed tomography shows additional lesions over the bilateral parietal region of scalp. For further management he was referred for radiotherapy. Radiotherapy was given for scalp lesion.

Elder sister a 13-year female child presented with a lesion over tongue, lower lip and as described above multiple hyper pigmented skin lesions as diagnosed case of Xeroderma pigmentosa. Punch biopsy from tongue and lip shows well differentiated squamous cell carcinoma. As further investigated computed tomography and magnetic resonant imagine was done which shows the lesion on the left lateral border of tongue then taken three cycles of chemotherapy. Lesion was clinically completely responded. After 1 month she developed another new lesion on scalp. Computed tomography brain was done which shows lesion on right parietal occipital lesion. Wide local excision was done which shows benign lesion suggestive of telangiectasia. Referred for radiotherapy for primary lesion management. Curative radiotherapy was given for tongue lesion. After 2 month she developed ulcer over left side cheek. Biopsy was taken which shows squamous cell carcinoma. After that she developed multiple swelling over face, forehead and scalp. Biopsy was done from left inner canthus shows basal cell carcinoma so further investigated. Computed tomography and magnetic resonant imagine was done which shows lesion involving skin and subcutaneous plane of right premolar region. So as further treatment 3 cycles of chemotherapy were given (Table 1).

	history	Malignancy	Genetic disorder	Prognosis	Treatment
Case 1	Present	Squamous cell carcinoma tongue	Xeroderma pigment's	Poor	Chemotherapy Radiotherapy
Case 2	Present	Squamous cell carcinoma scalp, cheek	Xeroderma pigment's	Poor	Chemotherapy Radiotherapy

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