Case Report of Xeroderma Pigmentosum

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Abstract

Background: Xeroderma pigmentosum (XP) is a rare autosomal recessive disease which occurs due to an abnormality of nucleotide excision repair caused by a mutation to any of eight genes (XPA-G and XPV). Epithelial neoplasms, severe ocular involvement, and neurological degeneration are some of the clinical manifestations of this disease. One of the most common malignancies is Squamous Cell Carcinoma.

Case Report: We report a case of a 80 years old patient which was admitted at the dermatologic department presenting a large hemorrhagic nodule in the shoulder and three ulcerated skin lesions located at the face and neck. He also presented freckles, telengiactesia, xerosis, actinic keratosis, erythema and lentiginous macules all over the body. The patient had ocular invasive SCC ten years ago, which was resolved by surgery, and a large hypotrophic scar in the lower back due to a massive SCC.

Conclusions: In general patients with Xeroderma Pigmentosum have shorter life because of recurrent malignancies and their interior organ involvement. It is important to have proper follow up and evaluation of the patient for early diagnosis and treatment of malignancies.