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Clinicopathologic features of ophthalmic neoplasms arising in the setting of Xeroderma pigmentosum

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Background: Patients with Xeroderma Pigmentosum (XP) are strongly predisposed to the development of numerous cutaneous cancers. However, the extent of ocular pathology in these patients has not been adequately studied.

Methods: We retrieved formalin-fixed paraffin-embedded material from tumors involving the ocular surface and ocular adnexa from 6 patients who met clinical criteria for XP diagnosis. Clinical information was obtained by retrospective chart review. Histopathological evaluation was performed, as well as immunohistochemistry in all available cases using antibodies directed against the most common mutated proteins in XP patients (XPA, XPC, and XPD).

Results: Patients included 4 males, 2 females with a mean age of 20.8 years (range 10 – 31 years) who met clinical criteria for XP and were found to have a total of 13 neoplasms involving the ocular surface and adnexal skin; 6 squamous cell carcinoma (SCC), 3 conjunctival intraepithelial neoplasia (CIN), 2 malignant melanoma (MM), 1 basal cell carcinoma (BCC) and 1 atypical fibroxanthoma (AFX). Complete XPD loss was present in two tumors from one patient suggesting a germline defect, and the invasive component of a squamous carcinoma from a second patient suggesting a somatic alteration. No clear pattern of loss for XPA or XPC was evident.

Conclusions: Our study outlines our early experience with pathology of ocular neoplasms in XP patients. XPC immunoreactivity in all tumors suggests that XPC genetic alterations may not be a common feature in our population. Immunohistochemistry for XPA and XPD may be more useful in the study of invasive tumors compared to in situ carcinoma. These findings deserve further exploration with genetic studies and additional patients.

Biography

Maria Jose Suarez currently pursuing Postdoctoral Research Fellow in Ophthalmic Pathology at Johns Hopkins University.

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