



GENETICS AND GENODERMATOSES

INTERNAL MALIGNANCIES IN PATIENTS WITH XERODERMA PIGMENTOSUM

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Introduction: Xeroderma pigmentosum (XP) genes have been incriminated as a predisposing factor to internal cancers. We herein describe 6 cases of XP with internal malignancies since 2014.

Case reports:

Patient 1 (XP-V) smoker and outdoor worker with no UV protection, developed since the age of 19, 3 squamous cell carcinomas (SCC) and at the age of 45, a lung cancer.

Patients 2 and 3 (XP-C) had a leiomyosarcoma of uterus at the ages of 20 and 28. Whereas, patient 2 never developed skin cancer. Patient 3 had since the age of 9, 21 basal cell carcinomas (BCC), 7 SCC and 4 melanomas.

Patient 4 (XP-C), with a history of 7 BCC and 1 SCC since the age of six, developed a thyroid cancer at age 21.

Patient 5 (XP-C), with has an excellent photoprotection and never developed skin cancer, died of an acute leukemia at the age of 9.

Patient 6 (XP-C) with history of 13 BCC, 1 SCC, 1 MM and poor photoprotection, since age six, developed a thyroid cancer at age 16.

Discussion: During the last 25 years, more than 600 cases of XP were treated in Tunisia but only very rare cases of internal tumors were reported. The reason may be the short life expectancy of these patients. If the mechanism of cutaneous carcinogenesis in XP is relatively fast (especially in XP-A and C), internal carcinogenesis is still obscure and seems to be a much slower process. It doesn't seem to involve UV-induced mutations but probably environmental carcinogens that cause DNA damage. The various levels of sun exposure of our patients comfort this hypothesis. Different types of internal cancer have been reported sporadically over XP. They are independent of exposure to sunlight. From our study, the internal cancers are more common in the XP-C (5 cases).

