Journal of Information Systems Engineering and Management
2025, 10(52s)
e-ISSN: 2468-4376
https://www.jisem-journal.com/

Research Article

Case Report of Xeroderma Pigmentosum of A 67- Year- Old Patient

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ARTICLE INFO

Received: 08 Mar 2025

Revised: 11 May 2025

Accepted: 20 May 2025

ABSTRACT

Introduction: Xeroderma pigmentosum (XP) is a rare autosomal recessive genodermatosis characterized by severe photosensitivity, skin pigmentary changes, malignant tumor development, and occasionally progressive neurologic degeneration. The median age of death in patients with XP without neurodegeneration is about 37 years old. The median age of death in patients with XP with neurodegeneration is younger at about 29 years old.

Case Report: A 67-year-old man presented to the Clinic of Dermatology to diagnose a painful nodular hemorrhagic growth on the right shoulder, an eroded hyperpigmented plaque in the right retro auricular region and 3 ulcerated crusted lesions with elevated borders at the frontal and temporal regions. On examination, the patient had diffuse freckle-like pigmentation, xerosis, actinic damage in sun-exposed areas, multiple Actinic Keratoses (AK) and a large scar in the lumbar region, resulting from the excision of a Squamous Cell Carcinoma (SCC) located on that area. He denied early blistering in his childhood, also refereed that due to its condition he had always worked indoor. He also reported being diagnosed with XP at a young age and, since then has been advised by a dermatologist to avoid sun exposure and to undergo regular examinations for any suspicious lesions suggestive for Carcinoma lesions in his body. His neurologic examination was normal and lymph node examination was negative. Dermoscopy of the lesions was performed showing characteristic features for Basal Cell Carcinoma (BCC), SCC and Actinic Keratoses. The lesions were removed surgically and the biopsies confirmed the clinical and dermoscopic diagnoses.

Conclusion: Early detection and proper treatment of XP can prevent the onset of common malignancies and increase the life expectancy of the patients.

Keywords: Basal Cell Carcinoma, Dermosscopy, Squamous Cell Carcinoma, Xeroderma pigmentosum.

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare autosomal recessive genodermatosis characterized by severe photosensitivity, skin pigmentary changes, malignant tumor development, and occasionally progressive neurologic degeneration[1,2]. There are 8 different genetic types of Xeroderma Pigmentosum (XP-A to XP- and XP-V). Certain XP subtypes (A, B, C, D, G, F) are associated with progressive neurologic degeneration[2], while patients with XP-V may have delayed onset of clinical manifestations, lack of neurologic findings and variable severity. The median age of death in patients with XP without neurodegeneration is about 37 years old. The median age of death in patients with XP with neurodegeneration is younger at about 29 years old. While the most common cause of death amongst patients with XP is metastatic Malignant Melanoma or Invasive Squamous Cell Carcinoma (SCC), the second most common cause is due to neurodegeneration[3,4].

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CASE REPORT

A 67-year-old man presented to the Clinic of Dermatology to diagnose a painful nodular hemorrhagic growth on the right shoulder (fig. 1), an eroded hyperpigmented plaque in the right retro auricular region (fig. 2) and 3 ulcerated crusted lesions with elevated borders at the frontal and temporal regions (figure 3).

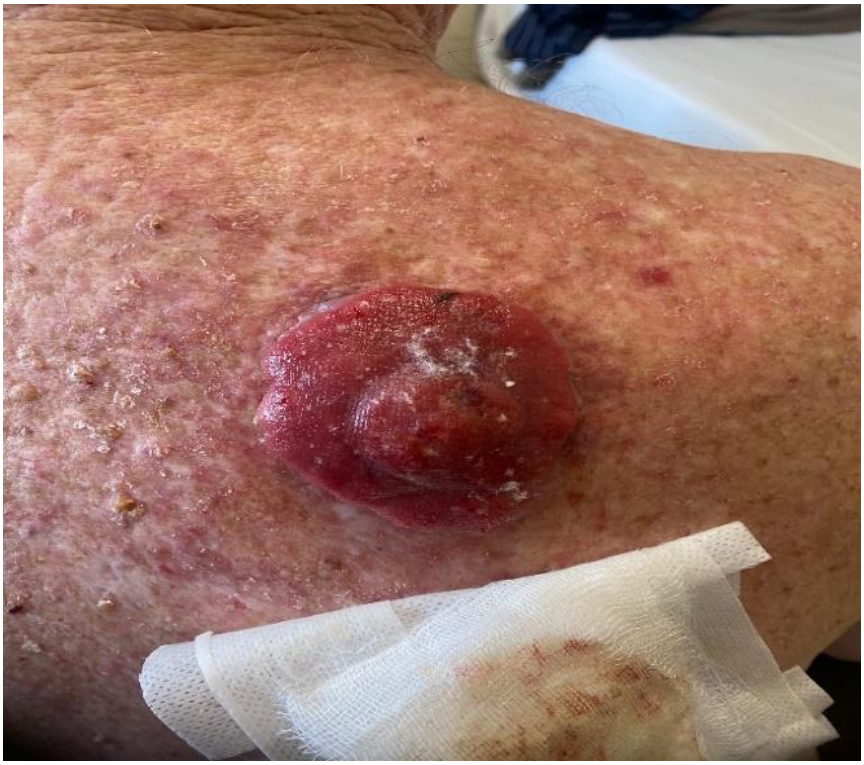

Figure 1: Large nodular hemorrhagic growth located in the right shoulder.

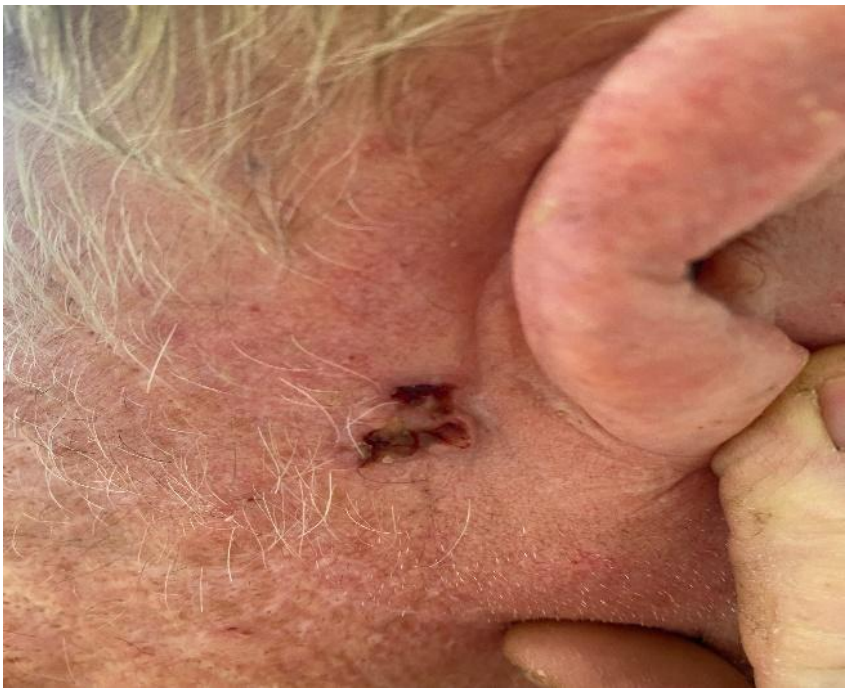

Figure 2: Eroded, hyperpigmented plaque in the right retro auricular region.

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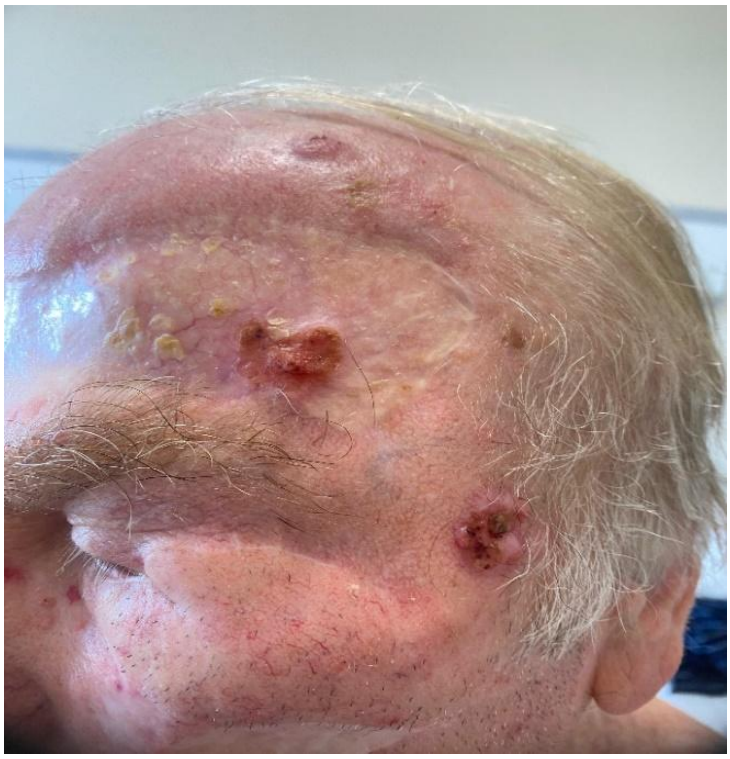

Figure 3: Multiple BCC located in the frontal and temporal region, large scar at the right eye due to a removal of a previous SCC of the conjuctiva.

On examination, the patient had diffuse freckle-like pigmentation, xerosis, actinic damage in sun-exposed areas, multiple Actinic Keratoses and a large scar in the lumbar region, resulting from the excision of a SCC located on that area (figure 4).

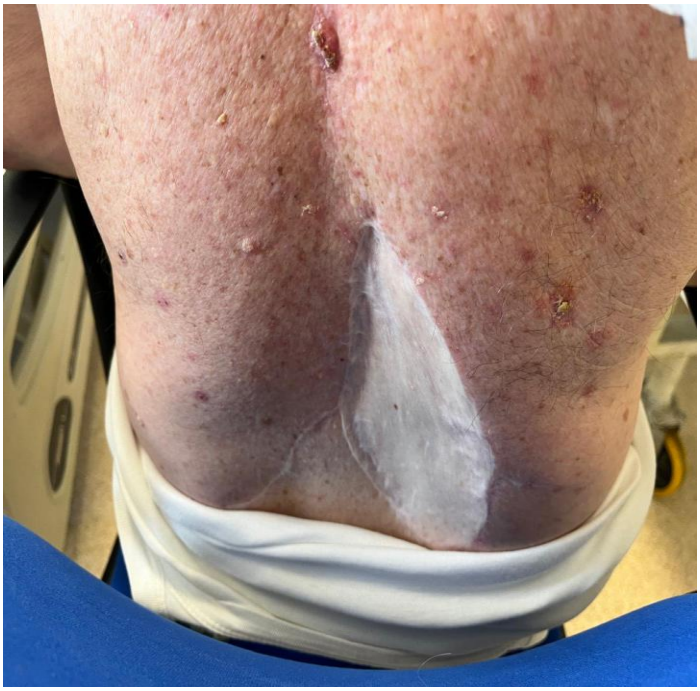

Figure 4: Multiple Actinic Keratoses, Xerosis of the skin and large scar at the lumbar region due to a previous removal of a large SCC.

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He indicated that he had ocular involvement of the XP, manifested with photophobia, conjunctival xerosis, blepharospasm and at the age of 39 years old, he was diagnosed with an invasive Squamous Cell Carcinoma of the conjunctiva of the left eye (figure 3). That led to the total removal of his eye Biopsies at his previous visits found 12 Basal Cell Carcinomas, 5 Squamous Cell Carcinoma but never a Melanoma. He denied early blistering in his childhood, also refereed that due to its condition he had always worked indoor. He also reported being diagnosed with XP at a young age and, since then has been advised by a dermatologist to avoid sun exposure and to undergo regular examinations for any suspicious lesions suggestive for carcinoma in his body. His neurologic examination was normal and lymph node examination was negative. Dermoscopy of the lesions was performed showing characteristic criteria for several BCC-s and a SCC of the nodus in the right shoulder (figure 5,6).

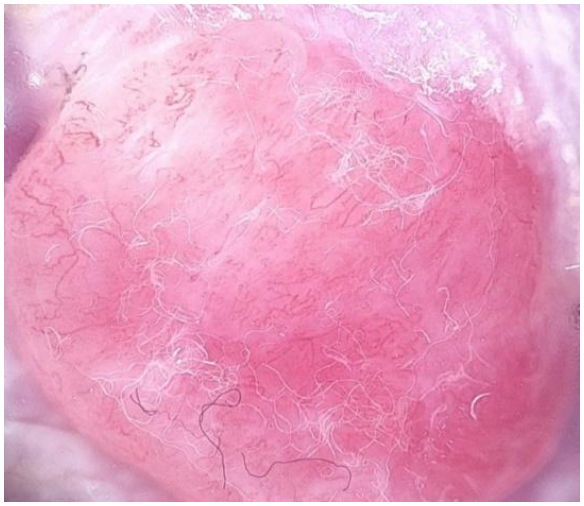

Figure 5: Dermoscopy of the large hemorrhagic SCC in the right shoulder

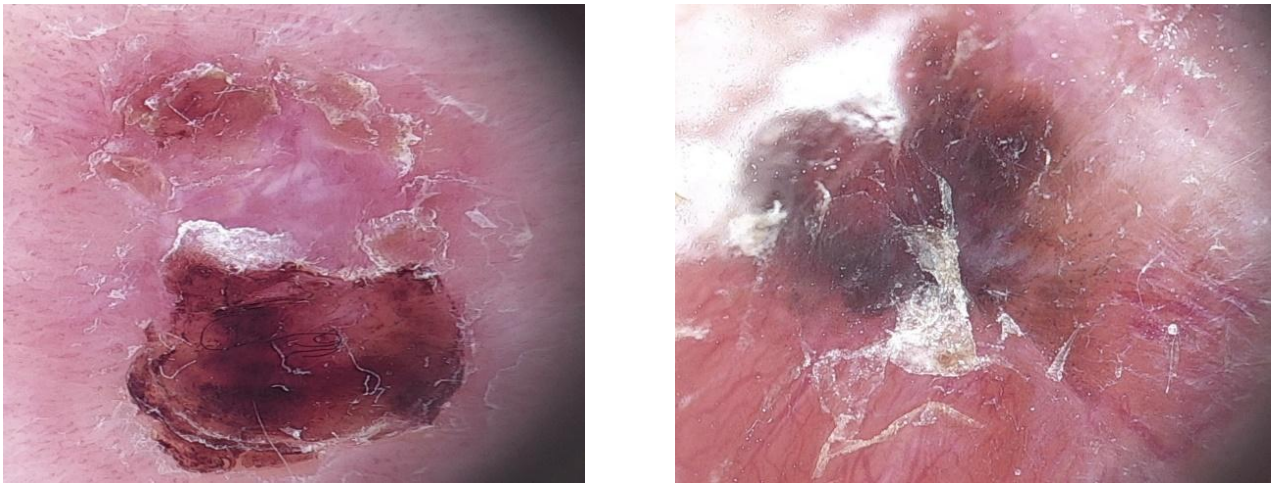

Figure 6: Dermoscopy of BCC in the retroauricular anf frontal region.

The lesions were removed surgically and the biopsies confirmed the clinical and dermoscopic diagnoses.

DISCUSSION

The main factor that makes this case noteworthy for presentation is the age of the patient. Statistics show that the median age of death in patients with XP without neurodegeneration is about 37 years old. The median age of death in patients with XP with neurodegeneration is younger at about 29 years old. The median age for the development of their first non-melanoma skin cancer is about 9[5,6]. Patients may develop dozens to hundreds of non-melanoma skin cancers per year. Patients with XP-V typically do not sunburn or present with extreme sunlight sensitivity. They do

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not exhibit exaggerating burn and can even tan, also they tend to be spared the neurologic findings and often have a better prognosis and longer life expectancy. Our patient is 67 years old and, although he reports having been diagnosed with XP-V since childhood and has undergone surgery for several SCC-s and BCC-s of varying sizes – one of which resulted with in the loss of an eye, he has not yet developed any major life- threatening complications, thanks to his continuous monitoring and awareness of the disease. Given the significantly increased risk of developing malignant tumors, patients with XP should have regular medical education[7]. The most effective method to avoid and decrease the number of malignant tumors is strict sun avoidance and protection. If XP patients are diagnosed in early life and stringent UVA protection is consistently maintained, the amount of DNA damage and subsequent skin cancers and ocular damage may be minimized[8]. At the time the diagnoses is made, dermatologists and family doctors must educate the patient/family about UV protection and how to live with this condition.

CONCLUSION

Early detection and proper treatment of XP can prevent the onset of common malignancies and increase the life expectancy of the patients.

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