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(CASE REPORT)

A rare occurrence of Hodgkin lymphoma in a Xeroderma pigmentosum patient: A case report and literature review

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Abstract

Xeroderma pigmentosum (XP) is a rare genetic disorder characterized by extreme sensitivity to ultraviolet (UV) radiation, resulting in an increased risk of skin cancer and other UV-induced malignancies. Reports of hematological malignancies occurring in patients with XP are found in the literature, but they seem to be quite rare. This report describes a 29-year-old male patient with XP who developed Hodgkin lymphoma. The potential increased risk of lymphoma in XP patients is highlighted in this paper; however, further studies are needed to better understand the underlying mechanisms of lymphomagenesis in XP patients and to develop optimal treatment strategies for hematological and other internal malignancies in this setting.

Keywords: Xeroderma pigmentosum; NER; Internal malignancies; Hodgkin lymphoma

1. Introduction

Xeroderma pigmentosum (XP) is a rare genetic disorder that affects the body's ability to repair damage induced by ultraviolet (UV) rays. The primary cause of this disorder lies in mutations affecting genes involved in the nucleotide excision repair (NER) pathway, which is responsible for repairing DNA damage caused by UV radiation. It is well-documented that XP patients are susceptible to various types of skin cancers, including basal cell carcinoma, squamous cell carcinoma, and melanoma. However, there is limited documentation in the scientific literature regarding the development of internal malignancies, such as hematological malignancies, especially Hodgkin lymphoma, and such occurrences are notably uncommon (1).

We hignlight the case of a 29-year-old Moroccan patient diagnosed with Xeroderma pigmentosum who developed Hodgkin lymphoma.

2. Case report

A 29-year-old Moroccan man, one of 2 siblings born to consanguineous parents, was diagnosed with Xeroderma pigmentosum at the age of 1 year, based on consanguinity and family history. He has been followed up in our establishment since the age of 6 and has undergone 6 surgeries for cutaneous carcinoma.

The patient was admitted to our department due to fever, weight loss, and night sweats persisting for the past month. General examination identified a febrile patient with lymphadenopathy grouped in clusters in the cervical, axillary, and bilateral inguinal regions, without hepatomegaly or splenomegaly. Dermatological examination revealed a poikilodermic lesion on the face and limbs, multiple solar lentigines, and freckles on the face and scalp. There was a

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nodular lesion on the lower left eyelid and destruction of the nasal cartilage with obstruction of the left nostril (Figure 1). On the scalp, there were four centimeter-sized pigmented nodular lesions with a pearly border.

The complete blood count showed normochromic normocytic anemia with lymphopenia and elevated LDH levels. Peripheral blood smear and bone marrow aspiration revealed inflammatory changes without any dysplastic features or blasts. Tuberculosis screening yielded negative results. The Positron Emission Tomography (PET) scan demonstrated diffuse nodal hypermetabolism. Lymph node biopsy revealed a morphological pattern consistent with classical Hodgkin lymphoma, with the tumor cells expressing CD20, CD15, CD30, and weakly expressing PAX5. They do not express CD3 or LMP1.

The patient was referred to a hematological establishment for specialized management. Additionally, he underwent enucleation of the left eye due to the rapid progression of the nodular lesion on the left eyelid, with histological findings indicating infiltrating squamous cell carcinoma

3. Discussion

Xeroderma pigmentosum (XP) is an infrequent autosomal recessive condition distinguished by a deficiency in nucleotide excision repair (NER) mechanisms, leading to the inability to repair DNA damage induced by ultraviolet (UV) radiation and other genotoxic agents. There are eight complementation groups of XP, each corresponding to mutations in specific genes involved in NER, including XPA…XPG, and XP dominant, with the XPC group being the most prevalent in the North African population (2).

Xeroderma pigmentosum patients exhibit extreme sensitivity to sunlight exposure due to their impaired NER function, resulting in an increased frequency of somatic mutations and a significantly higher incidence of skin cancers, such as squamous cell carcinoma and melanoma, at an early age, as was the case with our patient (3) (4). Moreover, NER dysfunction predisposes XP patients to develop tumors in non-sun-exposed tissues, including internal organs like neurological and hematological tissues (1).

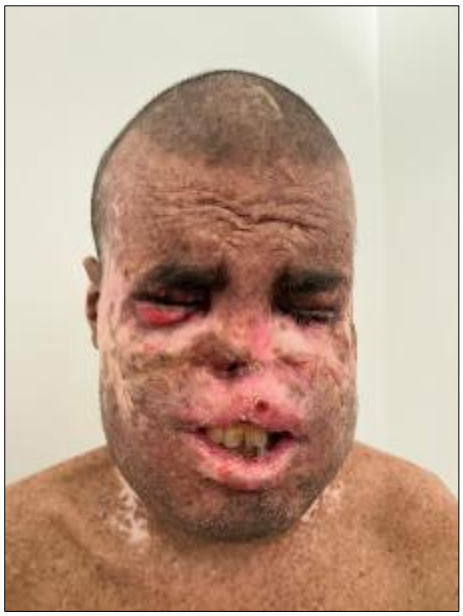

Figure 1 Multiple solar lentigines, and freckles on the face, nodular lesion on the lower left eyelid and destruction of the nasal cartilage with obstruction of the left nostril

In a combined cohort of 4 cohorts of individuals with xeroderma pigmentosum (XP), the relative risk of developing internal tumors was found to be 34 times higher with a comparatively younger age distribution compared to the general population. Among these tumors, the most elevated risks were associated with tumors of the central nervous system, hematological neoplasms, and gynecological tumors. Out of a total of 434 patients in the combined XP cohort, 11% developed internal malignancies, yielding 50 cases, of which 21 were hematological neoplasms. Among these individuals, those with XPC mutations constituted 54% of the total patients who developed internal malignancies (5).

In a French cohort study spanning from 1982 to 2022, out of 181 cases of XP examined, 117 originated from North Africa and were classified under the XPC group. Among these 117 individuals, 31% (43 patients) were found to have developed internal malignancies. Among these patients, the cohort listed 23 hematological neoplasms (53%) with 3 cases of Hodgkin lymphoma (6).

Another cohort study involving 117 XP patients conducted at the National Institutes of Health (NIH) from 1971 to 2018 revealed that 65 individuals were identified with mutations in the XPC gene. Among these, four patients of North African descent developed hematologic neoplasms, including high-grade lymphoma (7).

Management of hematologic neoplasms in XP patients presents unique challenges, as there are currently no consensus guidelines for treatment. Chemotherapy, the mainstay of treatment for internal tumors in the general population, poses significant risks in XP patients due to their impaired DNA repair mechanisms leading to myelosuppression and multiple organ failure (8).

4. Conclusion

Xeroderma pigmentosum patients exhibit susceptibility not only to external neoplasms but also to internal tumors, particularly in the Maghrebin region where the XPC subtype is prevalent. It is essential that physicians treating XP patients be aware of this strong predisposition, especially since XP patients now live longer due to better sun protection and improved knowledge of the disease. The management of these patients remains a challenge, and studies focusing on XP patients developing hematological malignancies are necessary to better understand the most appropriate strategies and precautions for this specific case.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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