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(CASE REPORT)

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A rare association between Xeroderma pigmentosum and diffuse large B-cell lymphoma: A case report

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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. Diffuse large B-cell lymphoma (DLBCL) is the most common type of non-Hodgkin's lymphoma (NHL). It belongs to a group of cancers that affect the lymphatic system. Reports of DLBCL occurring in patients with XP are found in the literature but it seems to be quite rare.

This report described a 31-year-old female patient with XP who developed DLBCL. 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 DLBCL in this setting.

Keywords: Lymphoma; Diffuse lymphoma large B-cell; Xeroderma pigmentosum; DLBCL; Case report

1. Introduction

Xeroderma pigmentosum (XP) is a rare genetic disorder that affects the body stability to repair damage caused by ultraviolet (UV) radiation from the sun. Mutations in genes involved in the nucleotide excision repair (NER) pathway, which is responsible for the DNA reparation after damage caused by UV radiation, is the main cause of this disease [1, 2, 6,7]. It is well known that XP patients are significantly at risk of developing skin cancers, including melanoma, basal cell carcinoma, and squamous cell carcinoma.

On the contrary, few reports can be found in the literature on XP patients developing lymphoma which is a cancer of the lymphatic system. So, the association between XP and hematologic malignancies such as diffuse large B-cell lymphoma (DLBCL) is reported but remains rare [3,4,5,6]. DLBCL is a type of non-Hodgkin's lymphoma that occurs when abnormal B-cells grow out of control and form tumors in the lymphatic system due to genetic mutations in the B-cells. Some studies suggest that the mutations causing XP may also affect the NER pathway in B-cells, leading to increased DNA damage and genomic instability. This genomic instability may then increase the risk of developing lymphoma [7].

This report described a patient with XP who developed DLBCL.

The subject provided informed consent to participate in this study as it was conducted according to the Helsinki Declaration. No regulatory approval is required for non-interventional studies according to the Moroccan law.

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2. Case presentation

2.1. Patient information

This paper presents a 31-year-old Moroccan female who was admitted at the department of clinical hematology at the University Hospital of Marrakesh, Morocco in 2020 for fever and declaring night sweats and a recent significant weight loss. Concerning the patient's medical history, she was diagnosed with XP at the age of two years old. She had a XP family history as her sister also had this condition. She had multiple basal cell carcinomas history as well.

2.2. Examinations and assessments

Physical examination revealed lymphadenopathy characterized by a mass in the right thigh causing a limp. The evolution was marked by an ulceration of the mass.

A biopsy of the mass was performed .The immune histochemical staining was positive for CD20, CD79a, CD 3 and BCL2 and negative for pancytokeratin, CD5, CD10, and BCL6, and consistent with a germinal center B-cell subtype. A bone marrow biopsy was prescribed too and was negative.

An initial blood test showed hemoglobin at 11g/dL , white blood cell at 12 thou/uL and a platelet count at 702 200 a VS at 76 mm, a lactate dehydrogenase level at 520 U/L. On MRI, a lesion in the right hemi-pelvis centered on the sacroiliac region was found measuring 106mm with uretero-vascular and uretero-hydronephrosis involvement up stream. Bilateral external and internal iliac lymphnodes were flowing a long with a fracture of the femoral neck with displacement in varus. Diffuse abnormalities in bone signal were noted as well in the iliac and femoral diaphysis.

As part of the extension assessment, a CT thoracic –abdominal-pelvic was prescribed and showed multiple mediastinal lymphnodes. The largest measured two cm. We found a large right pleural effusion and homogeneous hepatomegaly. Also , we observed a sacral and right iliac tumor process infiltrating the surrounding marrow areas, and the largest right external iliac lymphnode measured 15.6 mm.

2.3. Diagnostic and intervention

Mass biopsy revealed DLBCL by malignant tumor proliferation with large undifferentiated necrotic cells with numerous mitoses. The patient was treated with low dose of R-CHOP chemotherapy (rituximab, cyclophosphamide, doxorubicin, vincristine, and prednisone).

2.4. Outcome

After several of chemotherapy, the patient died due to complications of the DLBCL.

3. Discussion

The exact mechanisms linking XP and lymphoma are not fully understood yet, but researchers reported cases of individuals with XP developing various types of lymphoma, including Hodgkin's lymphoma, non-Hodgkin's lymphoma, and primary cutaneous B-cell lymphoma. According to them, genetic mutations causing XP may also predispose individuals to an increased risk of developing lymphoma [1,2, 8, 9,11].Also, XP patients may have a weakened immune system due to the chronic inflammation and the increased oxidative stress. The immune system plays a fundamental role in recognizing and eliminating cancer cells. Thus, XP could indirectly contribute to lymphoma development [10,11]. On top of that, some studies suggest that chronic UV exposure may contribute to lymphoma development in XP patients as UV radiation can damage DNA and suppress immune function [6]. Our case might go in line with these hypotheses as our patient developed a DLBCL without any specific lymphoma risk factor except XP.

On another note, the Epstein-Barr virus (EBV) could be one of the explanations of an association between XP and DLBCL. EBV is an ubiquitous virus that infects B-cells and is implicated in the pathogenesis of DLBCL. Some studies show that XP patients are at an increased risk of developing EBV [12]. However, EBV was not reported in our case.

It must be noted that XP patients with DLBCL face great challenges in their diseases management due to their increased sensitivity to radiation and chemotherapy as shown in our case. Indeed, radiation therapy may be contraindicated in XP patients due to the high risk of skin cancer probability, and some chemotherapy agents may exacerbate the DNA repair defects in XP cells .Compared to non-XP patients with DLBCL, they tend to get worse outcomes, although the exact reasons for this are unclear .Thus, even though the association between XP and lymphoma is rare, clinicians should be

aware of the possible hematologic malignancies in XP patients and consider prompt evaluation and biopsy of any suspicious masses or lymphnodes [3, 13].

4. Conclusion

This case highlights the risk of lymphoma in patients with XP. Long-term follow-up with a multidisciplinary team is important to manage the increased risk of skin cancer and any other malignancies in XP patients such as DLBCL. Indeed, early DLBCL diagnosis with an appropriate chemotherapy protocolis crucial for improving outcomes in XP patients with DLBCL.

More research is needed to better understand the link between XP and lymphoma. Understanding the genetic and molecular mechanisms underlying lymphomagenesis is central to develop more effective treatments and protocols for XP patients with DLBCL.

Compliance with ethical standards

Acknowledgments

Pr. TAZI, Dr. BENHALIMA , participated in design and development of the case report, managing and treating the patient, writing of the case report and final approval of the submitted version after critical review.

Disclosure of conflict of interest

All the authors: Yasmina BENHALIMA, Illias TAZI and Fatimzahra LAHLIMI report no conflict of interest in relation to the subject matter.

Statement of informed consent

Informed consent was obtained. The patient understands that her name and initials will not be published and has given her consent for clinical information to be reported in a case report.

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