



CERVICAL LYMPHADENOPATHY REVEALING METASTATIC EWING SARCOMA IN A PATIENT WITH XERODERMA PIGMENTOSUM: A CASE REPORT

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ABSTRACT

Background: Ewing sarcoma is an aggressive malignant neoplasm that predominantly affects children, adolescents, and young adults. It most commonly arises in bone or soft tissue and is characterized by specific molecular alterations involving FET and ETS family genes. Cervical lymph node metastasis is an uncommon presentation. Xeroderma pigmentosum (XP) is a rare inherited disorder associated with defective repair of ultraviolet-induced DNA damage and an increased risk of cutaneous malignancies. A causal association between XP and Ewing sarcoma has not been established.

Case presentation: We report the case of a 23-year-old woman with a history of xeroderma pigmentosum under medical follow-up who presented with cervical lymphadenopathy. A biopsy was performed and established the diagnosis of Ewing sarcoma, according to the available clinical information. The patient was subsequently referred to the oncology department for further evaluation and management. Detailed clinical findings, the primary tumor site, molecular characterization, staging results, and subsequent oncological treatment should be documented from the patient's medical records.

Conclusion: This case highlights the importance of investigating persistent or atypical cervical lymphadenopathy and obtaining histopathological confirmation before treatment. The coexistence of XP and Ewing sarcoma is clinically noteworthy but does not establish a causal relationship between the two conditions.

Keywords: Ewing sarcoma; Xeroderma pigmentosum; Cervical lymphadenopathy; Metastatic disease; Biopsy; Case report.

1 INTRODUCTION

Ewing sarcoma is an aggressive malignant tumor that predominantly affects children, adolescents, and young adults. It most frequently originates in bone, particularly the pelvis and long bones, but may also arise in soft tissues. Clinical manifestations depend on the anatomical location of the tumor and may include localized pain, swelling, and functional impairment.

Conventional Ewing sarcoma is characterized by a fusion involving a FET-family gene, most commonly EWSR1, and an ETS-family transcription factor, frequently FLI1. The diagnosis requires integration of clinical presentation, imaging, histopathological findings, immunohistochemistry, and molecular testing where available [1,2].

The most common metastatic sites are the lungs, bones, and bone marrow. Lymph node involvement is less frequently reported, making cervical lymphadenopathy an unusual presentation that may complicate the initial diagnostic assessment.

Xeroderma pigmentosum is a rare inherited disorder characterized by impaired repair or bypass of ultraviolet-induced DNA damage. The disease is associated with marked photosensitivity, pigmentary abnormalities, and a substantially increased risk of cutaneous malignancies, particularly basal cell carcinoma, cutaneous squamous cell carcinoma, and melanoma [3,4].

Although XP is associated with an increased susceptibility to certain malignancies, a causal relationship between XP and Ewing sarcoma has not been established. The coexistence of these two conditions should therefore be interpreted cautiously.

We report the case of a 23-year-old woman with XP in whom cervical lymphadenopathy led to the diagnosis of Ewing sarcoma. This case emphasizes the importance of tissue diagnosis, appropriate staging, and multidisciplinary oncological assessment in patients presenting with unusual cervical lymphadenopathy.

2 CASE PRESENTATION

2.1 Patient history

A 23-year-old woman with a known history of xeroderma pigmentosum under medical follow-up was evaluated for cervical lymphadenopathy. Her underlying genetic condition was an important element of her medical history because of the increased risk of malignancy associated with XP.

The duration of the cervical swelling, associated symptoms, previous cutaneous malignancies, family history, and other relevant medical conditions should be documented according to the patient's clinical records.

2.2 Clinical examination

The clinical examination focused on the cervical lymphadenopathy and the identification of a possible primary lesion. The precise cervical level, laterality, dimensions, consistency, mobility, tenderness, and relationship to surrounding structures should be reported from the original examination.

A complete head and neck examination is important in this setting to assess the upper aerodigestive tract and identify any possible primary lesion. Dermatological examination is also relevant in a patient with XP because of the increased risk of cutaneous malignancies.

The actual examination findings should be inserted here without extrapolation from illustrative or previously published cases.

2.3 Imaging investigations

Radiological assessment of cervical lymphadenopathy is essential for characterizing the lesion, evaluating its relationship to adjacent structures, and planning an appropriate biopsy.

Ultrasound and contrast-enhanced computed tomography may help determine the number, dimensions, morphology, and anatomical distribution of abnormal cervical lymph nodes. Additional imaging should be selected according to the suspected primary tumor and the requirements of systemic staging.

In a patient ultimately diagnosed with Ewing sarcoma, staging is necessary to determine the primary tumor site and assess the presence of distant disease. The actual imaging findings, including the primary tumor location and the presence or absence of other metastatic sites, should be described using the original radiology reports.

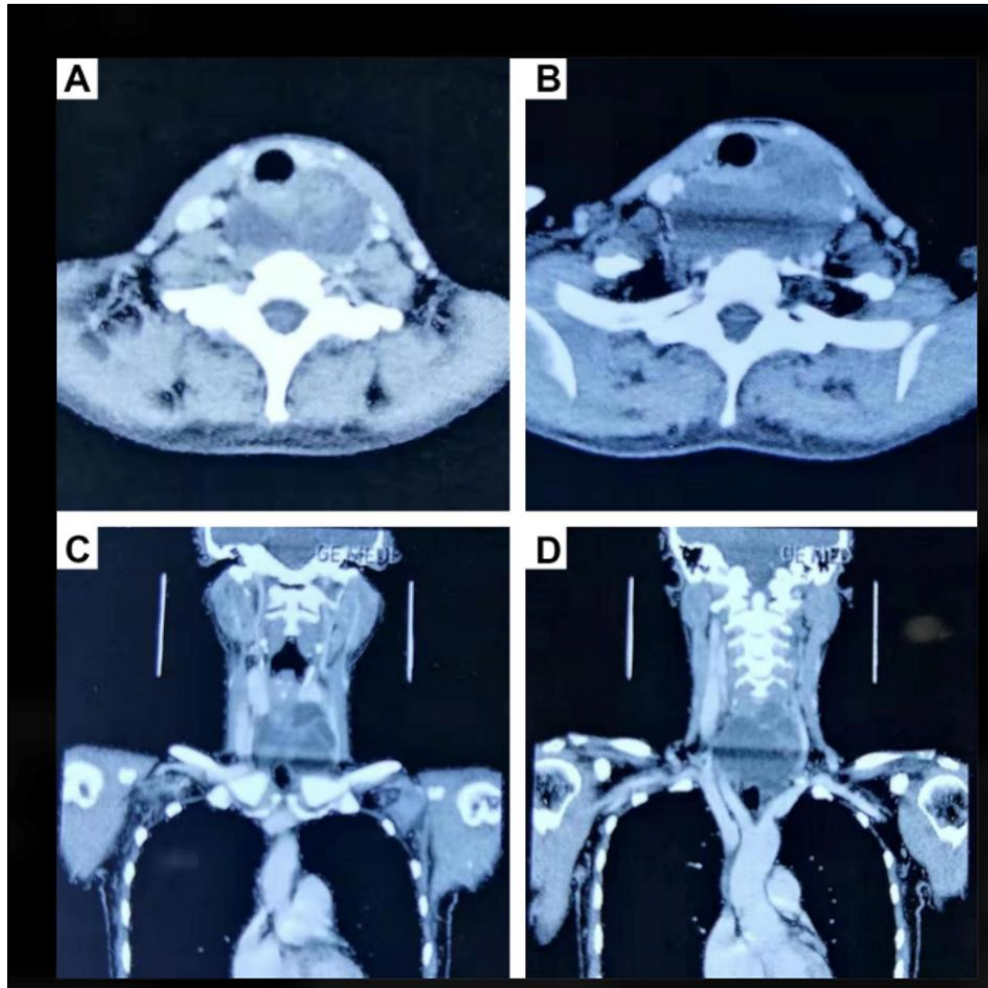


Figure 1: Representative pretreatment cervical CT imaging of Ewing sarcoma.

2.4 Histopathological diagnosis

A biopsy was performed to establish the nature of the cervical lesion. According to the available clinical information, the pathological diagnosis was Ewing sarcoma, and the patient was referred to oncology following the diagnostic procedure.

The original pathology report should be used to describe the histological appearance and immunohistochemical profile. Conventional Ewing sarcoma typically consists of relatively uniform small round cells with scant cytoplasm. CD99 expression is frequently strong and membranous, while NKX2.2 may provide additional diagnostic support.

These immunohistochemical findings are not independently specific. Molecular confirmation of a characteristic FET::ETS fusion, most frequently EWSR1::FLI1, is important where available to distinguish Ewing sarcoma from other small round cell neoplasms [1,2].

The exact immunohistochemical and molecular findings in this patient should be reported only if documented in the original laboratory results.

2.5 Oncological referral and management

Following biopsy, the patient was referred to the oncology department for further evaluation and treatment planning.

The management of Ewing sarcoma generally requires a multidisciplinary approach involving medical oncology, radiology, pathology, and the relevant surgical and radiation oncology teams. Treatment decisions depend on the primary tumor site, disease extent, resectability, and the patient's clinical condition.

The subsequent investigations and treatment received by this patient should be documented from the oncology records. The available information does not establish whether systemic chemotherapy, surgery, or radiotherapy was subsequently administered.

3 DISCUSSION

3.1 Cervical lymphadenopathy as a presenting manifestation

Cervical lymphadenopathy has a broad differential diagnosis, including infectious, inflammatory, hematological, and metastatic malignant conditions. Persistent or suspicious lymphadenopathy requires a systematic diagnostic approach, particularly when clinical findings raise concern for malignancy.

In patients with XP, a history of previous or suspected cutaneous malignancy may influence the initial differential diagnosis. Nevertheless, the underlying disorder should not lead clinicians to assume that every suspicious lesion represents a UV-associated skin cancer.

Tissue sampling is therefore essential when malignancy is suspected. In the present case, biopsy provided the diagnostic basis for referral to oncology. The precise anatomical distribution of the cervical disease and identification of the primary tumor are essential for interpreting the unusual nature of the presentation.

3.2 Diagnostic considerations in Ewing sarcoma

Ewing sarcoma belongs to a group of malignant small round cell tumors that may share overlapping morphological features. The differential diagnosis includes lymphoblastic lymphoma, rhabdomyosarcoma, neuroblastoma, poorly differentiated carcinoma, and other undifferentiated round cell sarcomas.

Immunohistochemistry is useful in narrowing the differential diagnosis, but individual markers should not be interpreted in isolation. Molecular characterization is particularly valuable in unusual anatomical presentations because it can support the diagnosis and distinguish conventional Ewing sarcoma from other tumors with overlapping features [1,2].

For this reason, the publication of the present case should include the actual histopathology report and any molecular testing performed. These results are important for ensuring that the diagnosis is accurately documented.

3.3 Xeroderma pigmentosum and malignancy

XP is associated with inherited abnormalities affecting the cellular response to ultraviolet-induced DNA damage. Patients typically require lifelong photoprotection and dermatological surveillance because of their substantially increased risk of cutaneous malignancies [3,4].

The occurrence of Ewing sarcoma in a patient with XP raises an interesting clinical observation, but the two conditions should not be assumed to share a causal mechanism. Ewing sarcoma is characterized by oncogenic gene fusions, whereas the established cancer susceptibility in XP is strongly linked to impaired processing of ultraviolet-induced DNA damage.

A single case cannot establish an epidemiological association or demonstrate that XP predisposes patients to Ewing sarcoma. Any discussion of a possible relationship must therefore remain appropriately cautious.

3.4 Treatment considerations

Ewing sarcoma is generally managed using systemic multiagent chemotherapy combined with local control of the primary tumor. Depending on the clinical circumstances, local treatment may involve surgery, radiotherapy, or both. The choice of treatment depends on tumor location, disease extent, response to systemic therapy, and the feasibility of achieving local control [1,2].

Cervical lymph node involvement presents additional considerations because treatment must account for the anatomical relationships of the involved nodes and the overall distribution of metastatic disease. However, the presence of cervical lymphadenopathy does not, by itself, establish an indication for neck dissection.

The treatment strategy should be determined by a multidisciplinary sarcoma team. In the present case, the documented management at the time of reporting consisted of biopsy followed by referral to oncology. Further therapeutic details should be incorporated only after verification of the patient's subsequent clinical course.

3.5 Clinical implications

This case emphasizes several practical principles. First, persistent cervical lymphadenopathy should be investigated systematically rather than attributed automatically to a patient's pre-existing medical condition. Second, histopathological assessment is central to establishing the diagnosis of an unusual malignant lesion. Third, suspected Ewing sarcoma requires appropriate pathological characterization and systemic staging to guide treatment.

Finally, patients with XP require continued specialist surveillance, but the appearance of a malignancy outside the best-established cancer spectrum of XP should be evaluated according to the same principles of accurate diagnosis and multidisciplinary care.

4 CONCLUSION

Cervical lymphadenopathy may present a diagnostic challenge when the underlying cause is an uncommon malignancy. This report describes a 23-year-old woman with xeroderma pigmentosum who underwent biopsy for cervical lymphadenopathy and was diagnosed with Ewing sarcoma before referral to oncology.

The case underscores the importance of tissue diagnosis, accurate pathological characterization, and appropriate oncological evaluation. Although the coexistence of XP and Ewing sarcoma is noteworthy, it does not establish a causal relationship between the two conditions.

The primary tumor site, molecular diagnostic findings, staging results, and subsequent treatment should be documented in the final manuscript to provide a complete and clinically interpretable case report.

STATEMENTS ON COMPLIANCE WITH ETHICAL STANDARDS

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Disclosure of Conflict of Interest

The authors declare no conflicts of interest regarding this manuscript.

Statement of Informed Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying clinical images. All reasonable measures were taken to protect the patient's identity and confidentiality

REFERENCES

- [1] National Cancer Institute. Ewing Sarcoma and Undifferentiated Small Round Cell Sarcomas of Bone and Soft Tissue Treatment (PDQ®)–Health Professional Version. Available at: <https://www.cancer.gov/types/bone/hp/ewing-treatment-pdq>
- [2] National Cancer Institute. Childhood Cancer Genomics (PDQ®). Available at: <https://www.cancer.gov/types/childhood-cancers/pediatric-genomics-hp-pdq>
- [3] Kraemer KH, DiGiovanna JJ. Xeroderma pigmentosum. In: GeneReviews®. University of Washington, Seattle. Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1397/>

- [4] National Cancer Institute. Xeroderma Pigmentosum. Available at: <https://www.cancer.gov/>
- [5] Published case report of cervical extraosseous Ewing sarcoma, including representative CT imaging. Available at: <https://pmc.ncbi.nlm.nih.gov/articles/PMC10904673/>