Case Report

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Small Round Blue Cell Sarcoma of the Left Humerus in a Pregnant Patient: Clinical Course of a Rare Aggressive Soft Tissue Tumor

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Abstract

Small round blue cell tumors (SRBCT) represent a heterogeneous group of aggressive tumors with overlapping histological features, often requiring complex immunohistochemical and molecular studies for accurate diagnosis. Their occurrence during pregnancy poses significant diagnostic and therapeutic challenges, particularly when located in rare anatomical sites such as the humerus. We present the case of a 21-year-old pregnant woman at 31 weeks of gestation who was admitted with progressive pain and swelling in the left upper extremity. MRI revealed a large, intramedullary soft tissue mass in the mid-diaphysis of the left humerus with extension to adjacent musculature and neurovascular structures. Initial histopathological evaluation identified a poorly differentiated SRBCT. Despite extensive immunohistochemical work-up, a definitive diagnosis, necessitating advanced molecular analysis, remained elusive. The patient underwent cesarean delivery at 33 weeks due to obstetric indications. Subsequent PET-CT imaging demonstrated multiple hypermetabolic lesions consistent with metastatic disease. Chemotherapy with the VAC regimen was initiated following postpartum stabilization. This case underscores the complexity of diagnosing SRBCT during pregnancy and highlights the importance of multidisciplinary collaboration. Early recognition, thorough histopathological and molecular evaluation, and individualized oncologic and obstetric management are critical for optimizing maternal and fetal outcomes.

Keywords: Humerus, perinatal oncology, pregnancy, small round blue cell tumor, soft tissue sarcoma

Introduction

Small Round Blue Cell Sarcoma, particularly Ewing's sarcoma, presents significant clinical management challenges in pregnant patients due to its rarity and the complexities of treatment during gestation. Diagnosis often occurs late, with nonspecific symptoms, as evidenced by patients presenting with fractures or localized masses (1, 2). Management typically involves a multidisciplinary approach, including preoperative chemotherapy and surgical intervention, with considerations for fetal safety (2-4). Prognostic factors indicate that maternal survival rates can be favorable, with one-year survival at 100% for bone sarcomas, although complications such as metastasis and local compression symptoms are common (4). Case studies highlight the necessity for individualized treatment plans, balancing maternal health and fetal outcomes, as seen in cases where cesarean sections were performed to mitigate risks(1). Overall, the prognosis remains variable, necessitating further research to optimize management strategies for this unique patient population (4).

Case Report

A 21-year-old primigravida woman at 31 weeks of gestation presented with progressive pain and swelling in the left upper extremity. Her medical history was unremarkable, and there was no known family history of malignancy or genetic disorders. The pregnancy proceeded without complications until the emergence of musculoskeletal symptoms. Due to breech presentation and arrest of labor, a cesarean section was performed at 33 weeks of gestation. The patient reported no history of tobacco, alcohol, or substance use.

The initial complaint involved localized pain and swelling in the mid-portion of the left arm. Physical examination revealed soft tissue fullness, marked tenderness over the humerus, and restricted range of motion due to pain. No constitutional symptoms such as fever, weight loss, or fatigue were noted.

Magnetic resonance imaging demonstrated a destructive, intramedullary mass in the mid-diaphysis of the left humerus, measuring up to 85×50 mm. The lesion exhibited cortical disruption and extension into surrounding musculature and

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adjacent neurovascular structures (Figure-1). Tru-cut biopsy revealed features consistent with an undifferentiated small round blue cell sarcoma. Immunohistochemical analysis showed diffuse positivity for vimentin, actin, S100, synaptophysin, and a Ki-67 proliferation index of 35%. Markers including CD99, desmin, myogenin, and others were negative. A definitive diagnosis could not be established despite comprehensive immunophenotyping, and further molecular testing (FISH for CIC and EWSR1 rearrangements) was recommended.

Postpartum fluorodeoxyglucose (FDG) PET-CT imaging (Figure-2) revealed multiple hypermetabolic metastatic lesions in the lungs, thoracic and cervical vertebrae (including T10 and sacrum), and contralateral humerus. Following delivery, the patient was initiated on chemotherapy with a VAC regimen (vincristine, actinomycin D, cyclophosphamide).

As expected, the tumor exhibited high metabolic activity and locally invasive behavior. Unexpectedly, disseminated metastatic disease was present at the time of diagnosis, suggesting a more rapid progression possibly influenced by diagnostic delays associated with pregnancy. The prognosis is currently guarded, owing to the tumor's aggressive biological nature, undifferentiated histopathology, and the extent of metastatic involvement. The patient remains under multidisciplinary oncological care, with molecular analysis ongoing to refine therapeutic planning.

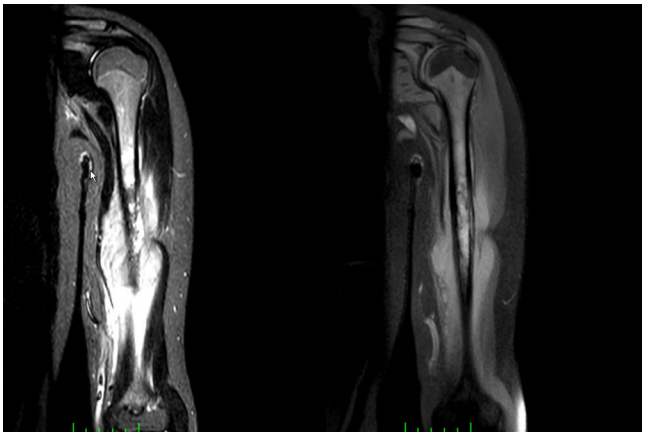

Figure 1. The lesion exhibited cortical disruption and extension into surrounding musculature and adjacent neurovascular structures

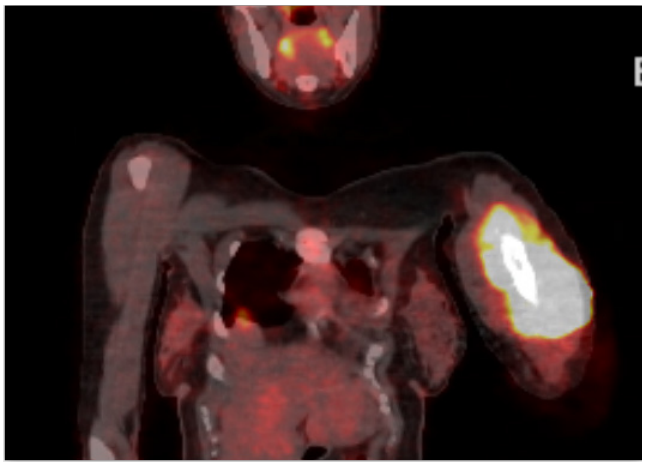

Figure 2. Postpartum fluorodeoxyglucose (FDG) PET-CT imaging

Discussion

This case report presents a 21-year-old pregnant female patient who presented with pain in the left arm, specifically the shoulder region. Upon initial assessment, an MRI revealed a mass-like lesion located in the humeral midshaft, which was associated with an intramedullary extension. The lesion appeared to involve adjacent muscle structures, particularly the medial head of the triceps and the brachialis, with significant diffusion restriction. Further radiologic studies, including a CT and PET scan, confirmed the suspicion of metastasis, highlighting the presence of a hypermetabolic lesion in the left humerus, suggesting a primary tumor with metastatic spread (Figure 2). Additionally, the biopsy report raised concerns about a small blue round cell tumor, with possibly Ewing's sarcoma being a differential diagnosis.

The unique aspect of this case is the diagnosis in a pregnant patient, which is uncommon for soft tissue sarcomas like Ewing's sarcoma, particularly during the third trimester. The diagnostic process involved multiple imaging modalities, including MRI, CT, PET/CT, and histopathology, contributing to a multi-dimensional view of the lesion (Figure 1,2). Of particular note is the failure to achieve a definitive diagnosis from immunohistochemistry, with initial results pointing to the possibility of a neurogenic or myogenic sarcoma. However, including specialized methods such as FISH (Fluorescence in situ Hybridization) and additional genetic and molecular testing is crucial to reaching a more conclusive diagnosis (5). The pathology report indicated that the tumor exhibited aggressive local infiltration into surrounding muscular tissues, which could result in further complications if left untreated. This local infiltration, coupled with metastases seen in the lungs and vertebrae, presents a serious concern regarding disease progression and necessitates urgent management (6).

One of the significant challenges in this case was the complex interplay between the tumor's progression and the pregnancy. Ewing's sarcoma, while a relatively rare diagnosis in young adults, becomes particularly challenging to manage during pregnancy due to the limited options for chemotherapy and radiation therapy (7). Given the patient's status as 31 weeks pregnant, the oncological management needed to be balanced with the safety of both the mother and the fetus. The presence of a primary lesion with suspected metastasis further complicates this situation, as delaying chemotherapy could increase the risk of widespread disease progression, while immediate treatment might endanger the pregnancy.

The use of chemotherapy in pregnant women is controversial, particularly when the drugs used in traditional chemotherapy regimens (such as VAC, Vincristine, Dactinomycin, and Cyclophosphamide) are teratogenic in nature (8). In this case, chemotherapy was initiated with appropriate pre-treatment precautions, including anesthesia and careful monitoring, highlighting the importance

of multidisciplinary coordination between oncology, obstetrics, and anesthesiology teams. Another notable point is the recommendation for further molecular and cytogenetic testing, which is essential for confirming the diagnosis of Ewing's sarcoma and evaluating possible genetic mutations, such as EWSR1 gene translocations, which are characteristic of this tumor type (9). These genetic markers would help refine the diagnosis, determine prognosis, and guide treatment decisions (10). Moreover, such testing is imperative to distinguish between other small round blue cell tumors, such as neuroblastoma or lymphoma, which may have overlapping clinical and histopathological features.

Integrating molecular genetics into clinical practice is increasingly critical in treating cancers, especially in rare or unusual cases like this one. Given that conventional histopathology and immunohistochemistry could not definitively identify the tumor subtype, relying on genetic sequencing could provide the clarity needed to proceed with more targeted therapies. The patient's prognosis is heavily dependent on the stage of the disease at diagnosis, with metastatic involvement of the vertebrae and lungs raising concerns about a poor prognosis. Despite this, prompt initiation of chemotherapy, which is effective in treating Ewing's sarcoma, offers a chance for disease control. The patient's young age is a positive prognostic factor, as younger individuals tend to tolerate chemotherapy better and may experience more favorable responses. Furthermore, ongoing follow-up and monitoring through imaging and molecular markers will be necessary to assess the tumor's response to chemotherapy and to detect any potential recurrence or progression, particularly considering the presence of metastases. This case also brings forth ethical and psychological concerns regarding the management of a pregnant cancer patient. The psychological burden on the patient, who faces not only the threat to her own life but also the health of her unborn child, must be addressed. Offering appropriate counseling and psychological support is crucial in ensuring the patient's well-being and compliance with the proposed treatment regimen. In terms of ethics, the decision to proceed with chemotherapy, which poses a risk to fetal development, requires careful consideration. This decision-making process must involve the patient, her family, and a multidisciplinary team, with clear communication regarding the potential risks and benefits. The healthcare team must also ensure that the patient fully understands the implications of the treatment options available to her and the fetus.

Conclusion

This case emphasizes the complexity of diagnosing and managing soft tissue sarcomas, particularly Ewing's sarcoma, in pregnant patients. The use of advanced imaging, genetic testing, and chemotherapy offers promising avenues for treatment, but careful coordination among healthcare providers and consideration of the ethical implications are crucial for optimizing outcomes. Further research into safer treatment protocols for pregnant patients with cancer is needed to improve the prognosis for both the mother and the fetus. This case also highlights the need for continuous advancements in molecular diagnostics to refine the classification and treatment of rare tumors like Ewing's sarcoma.

Patient consent for publication: Written informed consent was obtained directly from the patient involved in this case.

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