CASE REPORT

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Alveolar Rhabdomyosarcoma of the Hand with Bone Marrow and Skeletal Metastases in an Adolescent: A Rare Case Report

P Raja[1], Govindan[2]

Assistant professor,professor

Department Of Pathology

Amala Institute of Medical Sciences, Thrissur

Email: rajapirai@gmail.com

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Abstract

Background: Alveolar rhabdomyosarcoma (ARMS) is a rare and aggressive soft tissue sarcoma that commonly affects adolescents and is associated with early metastasis and poor prognosis. Primary involvement of the hand is exceptionally uncommon, posing significant diagnostic and therapeutic challenges.

Case Presentation: A 15-year-old female presented with a progressively enlarging painful swelling of the right hand associated with restricted finger movements. Ultrasonography and magnetic resonance imaging revealed an aggressive soft tissue mass involving the fourth and fifth metacarpal region with cortical erosion. Histopathological examination and immunohistochemistry confirmed the diagnosis of alveolar rhabdomyosarcoma. Whole-body positron emission tomography-computed tomography and bone marrow examination demonstrated multiple skeletal and bone marrow metastases. The patient received multimodal treatment comprising multi-agent chemotherapy followed by second-line chemotherapy due to disease progression. Owing to extensive local disease, a right below-elbow amputation was performed. Despite aggressive management, follow-up imaging demonstrated persistent metabolically active disease with further metastatic progression, necessitating third-line chemotherapy and palliative supportive care.

Conclusion: This case highlights the rare occurrence of alveolar rhabdomyosarcoma arising from the hand with advanced metastatic disease. Early diagnosis using multimodal imaging, histopathology, immunohistochemistry, and comprehensive staging is essential for timely management. However, metastatic ARMS continues to have a poor prognosis despite intensive multimodal therapy, emphasizing the need for improved therapeutic strategies.

Keywords: Alveolar rhabdomyosarcoma, Hand tumour, Soft tissue sarcoma, Bone marrow metastasis, Skeletal metastasis, PET-CT, Below-elbow amputation, Adolescent, Case report

Introduction

Alveolar rhabdomyosarcoma (ARMS) is a rare and highly aggressive subtype of rhabdomyosarcoma, accounting for approximately 20–30% of all rhabdomyosarcomas, and predominantly affects adolescents and young adults. Compared with the embryonal subtype, ARMS demonstrates a more aggressive biological behavior, characterized by rapid local invasion, early hematogenous dissemination, and a significantly poorer prognosis. The tumour commonly metastasizes to the lungs, regional lymph nodes, bones, and bone marrow, making metastatic disease a major determinant of survival. Most cases are associated with PAX3–FOXO1 or PAX7–FOXO1 fusion genes, which are linked to increased tumour aggressiveness and adverse clinical outcomes [1,2].

Primary involvement of the hand is exceptionally uncommon, with only a limited number of cases reported in the literature. Because of its rarity and non-specific clinical presentation, lesions involving the hand are frequently misdiagnosed as benign soft tissue swellings, traumatic injuries, osteomyelitis, or inflammatory conditions, resulting in delayed diagnosis and advanced disease at presentation. Comprehensive evaluation using magnetic resonance imaging (MRI), positron emission tomography-computed tomography (PET-CT), histopathological examination, immunohistochemistry, and molecular testing is essential for accurate diagnosis, staging, and treatment planning. Immunohistochemical positivity for Desmin, MyoD1, and Myogenin remains the cornerstone for confirming skeletal muscle differentiation and distinguishing ARMS from other small round blue cell tumours [3,4].

The current standard treatment for ARMS involves a multimodal approach combining systemic chemotherapy, surgical resection, and radiotherapy. However, the prognosis remains poor in patients presenting with metastatic disease, particularly those with bone marrow or skeletal involvement, with substantially lower long-term survival rates than patients with localized disease. Recent advances in molecular diagnostics and targeted therapies have improved the understanding of tumour biology, but effective treatment options for advanced ARMS remain limited [5-9].

We report a rare case of alveolar rhabdomyosarcoma arising in the hand with extensive skeletal and bone marrow metastases in an adolescent, highlighting the diagnostic challenges, clinicopathological findings, multimodal management, and the importance of early recognition of this uncommon presentation. This case adds to the limited literature on hand ARMS and emphasizes the need for a multidisciplinary approach to optimize patient outcomes.

Case Presentation

Patient Information

A 15-year-old female presented with a progressively enlarging swelling over the right hand for approximately three months. She complained of pain, restricted finger movements, and difficulty performing routine daily activities. There was no history of trauma, fever, or significant constitutional symptoms. The patient had no known family history of malignancy.

Clinical Findings

Physical examination revealed a firm, tender swelling involving both the palmar and dorsal aspects of the right hand. The swelling was diffuse with restricted movements of the fourth and fifth digits. The overlying skin appeared stretched without ulceration during the initial presentation. The clinical differential diagnosis included infective and neoplastic soft tissue lesions.

Diagnostic Assessment

Ultrasonography

Ultrasonography of the right hand demonstrated a well-defined hypoechoic lesion measuring 5.4 × 4.2 × 2.4 cm, encasing the flexor tendons without internal calcification, necrosis, or significant vascularity. The findings were suggestive of a soft tissue lesion, and MRI was recommended for further evaluation.

MRI Findings

Magnetic resonance imaging revealed a 5.3 × 4.8 × 4.0 cm lobulated soft tissue mass centered over the fourth and fifth metacarpal region. The lesion involved the palmar and dorsal interosseous muscles with extension into adjacent tendon compartments. Cortical erosion and marrow edema involving the fourth and fifth metacarpals indicated an aggressive malignant soft tissue neoplasm with osseous involvement.

Histopathological Examination

Core needle biopsy demonstrated a poorly differentiated malignant small round cell tumour composed of tumour cells arranged in alveolar, papillary, nested, and dyscohesive patterns with invasion into skeletal muscle. Frequent atypical mitotic figures (5–6 per 10 high-power fields) were identified. The initial

differential diagnoses included alveolar rhabdomyosarcoma, Ewing sarcoma, and synovial sarcoma.

Immunohistochemistry

Immunohistochemical analysis showed diffuse positivity for Desmin, MyoD1, Myogenin, CD99, and BCL-2, with weak positivity for TLE-1. The tumour cells were negative for Synaptophysin, Smooth Muscle Actin (SMA), S100, FLI-1, and NKX2.2, confirming the diagnosis of alveolar rhabdomyosarcoma.

Bone Marrow Evaluation

Bone marrow aspiration and biopsy revealed metastatic tumour involvement. Initial marrow examination showed atypical malignant cells consistent with metastatic rhabdomyosarcoma, confirming advanced-stage disease with bone marrow metastasis.

PET-CT Findings

Whole-body PET-CT demonstrated an intensely FDG-avid soft tissue mass involving the right hand with erosion of the fourth and fifth metacarpal bones. Multiple metabolically active skeletal lesions involving the right humerus, L5 vertebral body, right iliac bone, sacrum, bilateral femora, and right knee were identified, consistent with metastatic disease. Follow-up PET-CT after chemotherapy demonstrated persistent metabolic activity of the primary lesion without significant metabolic response and the appearance of a new metabolically active right paravertebral soft tissue lesion, indicating disease progression.

Final Diagnosis

Alveolar rhabdomyosarcoma of the right hand with bone marrow involvement and multiple skeletal metastases (Stage IV metastatic disease).

Therapeutic Intervention

The patient received multimodal treatment under a multidisciplinary oncology team. She was initially treated with first-line multi-agent chemotherapy (IRS-IV/VAC-based protocol). Although bone marrow disease showed an initial response, follow-up imaging demonstrated persistent disease and progression of metastatic lesions. She subsequently received second-line chemotherapy with Cyclophosphamide and Topotecan. Owing to further disease progression despite salvage chemotherapy, the patient underwent surgical management followed by third-line chemotherapy and palliative supportive care.

Surgical Findings (Below-Elbow Amputation)

A right below-elbow amputation was performed on 31 March 2026 because of extensive local disease progression. Gross examination of the amputated forearm specimen measured approximately 25.5 × 16.5 × 7.5 cm. A large nodular tumour occupied both the palmar and dorsal aspects of the right hand and wrist. The overlying skin was tense and stretched with prominent dilated superficial blood vessels. On cut section, the tumour appeared grey-white, fleshy, and predominantly solid, with extensive infiltration of the surrounding soft tissues. The lesion involved the carpal and metacarpal bones, confirming extensive local invasion. Histopathological sampling included the skin resection margins (A1–A6), soft tissue margins (A7–A9), nerve bundle margin (A10), tumour tissue (A11–A15), involved bones (A16–A18), radius (A19), and ulna bone resection margin (A20).

Follow-up and Outcome

Despite aggressive multimodal treatment, including multi-agent chemotherapy and surgical intervention, the patient demonstrated progressive metastatic disease. Follow-up PET-CT showed persistent FDG uptake within the primary tumour, multiple skeletal metastases, and a newly developed right paravertebral metastatic lesion, indicating poor response to therapy. The patient continued under multidisciplinary oncology care with third-line chemotherapy and palliative supportive management because of persistent advanced metastatic disease.

Discussion

Alveolar rhabdomyosarcoma represents one of the most aggressive pediatric soft tissue sarcomas and accounts for approximately one-quarter of all rhabdomyosarcomas. Compared with embryonal

rhabdomyosarcoma, ARMS demonstrates earlier hematogenous dissemination, increased metastatic potential, and significantly poorer survival. The presence of PAX3–FOXO1 and PAX7–FOXO1 fusion genes has been consistently associated with aggressive tumor biology and poor prognosis. Recent studies have demonstrated that patients presenting with metastatic disease continue to have substantially lower overall survival despite advances in multimodal therapy [9–11].

The present case is unusual because the primary tumor originated in the hand, an exceptionally uncommon anatomical location. Most extremity rhabdomyosarcomas arise around the forearm, thigh, or lower leg, whereas hand involvement has rarely been described. Owing to its rarity, lesions involving the hand frequently mimic cellulitis, osteomyelitis, traumatic swelling, or benign soft tissue tumors, often resulting in delayed diagnosis. Similar diagnostic challenges have been described in recent reports of extremity rhabdomyosarcoma [12,13]. Magnetic resonance imaging played a pivotal role in evaluating local tumor extent, demonstrating aggressive soft tissue infiltration with metacarpal destruction. Whole-body PET-CT further identified widespread skeletal metastases, emphasizing the importance of comprehensive staging. Current international guidelines recommend MRI for local staging and PET-CT for detecting distant metastases because both modalities provide superior sensitivity compared with conventional imaging [4,10].

Histopathological examination remains the diagnostic gold standard. In our patient, immunohistochemistry demonstrated diffuse positivity for Desmin, MyoD1, and Myogenin, confirming skeletal muscle differentiation. These markers remain the most reliable immunohistochemical indicators for distinguishing ARMS from other malignant small round blue cell tumors such as Ewing sarcoma, lymphoma, neuroblastoma, and poorly differentiated synovial sarcoma [9,14]. Bone marrow metastasis is an uncommon but well-recognized manifestation of advanced ARMS and carries significant prognostic implications. Recent pediatric oncology studies have shown that simultaneous skeletal and bone marrow involvement is associated with markedly reduced event-free and overall survival because of extensive hematogenous dissemination [1,8]. The present patient demonstrated diffuse skeletal lesions together with bone marrow infiltration at diagnosis, indicating advanced-stage disease.

Current treatment strategies rely on multimodal therapy combining systemic chemotherapy, surgery, and radiotherapy. Despite aggressive chemotherapy followed by below-elbow amputation for local disease control, our patient demonstrated continued disease progression, reflecting the aggressive biological behavior of metastatic ARMS. Similar findings have been reported in recent literature, where metastatic ARMS remains associated with poor therapeutic response despite standardized treatment protocols [11,15]. Recent advances in molecular oncology have improved understanding of ARMS biology and have identified several potential therapeutic targets, including IGF-1R, FGFR4, PI3K/AKT/mTOR signaling pathways, and immune checkpoint inhibitors. Although these targeted therapies remain under clinical investigation, they represent promising future treatment options for patients with metastatic disease [6,11,16].

Summary

A rare case of alveolar rhabdomyosarcoma of the right hand in a girl aged 15 years,with bone marrow and multiple skeletal metastasis is presented here. MRI, histopathology, and immunohistochemistry results with support by PET-CT and MBE confirmed diagnosis. This case was a severe form of Ewing sarcoma that showed continuous local and metastatic progression in spite of multimodal treatment with multi-agent chemotherapy followed by below-elbow amputation. This report highlights the aggressive clinical behaviour of metastatic alveolar rhabdomyosarcoma, pitfalls in diagnosis due to its rare anatomical site and reinforces a multidisciplinary approach for optimal management of such patients.

Conclusion

To date, alveolar rhabdomyosarcoma of the hand has not been described in the literature probably due to its status as a very rare and aggressive malignancy. For accurate staging and timely intervention, early diagnosis through advanced imaging, histopathological examination, immunohistochemistry, and thorough metastatic workup is crucial. Bone marrow infiltration and distant skeletal metastases at presentation have an adverse prognosis even in the setting of intensive multimodal therapy. This case highlights the importance of prompt diagnosis of atypical soft tissue tumours, coordinated treatment between multiple specialities and further investigation into optimized management in patients with advanced alveolar rhabdomyosarcoma.

Declaration

Consent: Written informed consent was obtained from the patient. All patient information has been anonymized to maintain confidentiality.

Conflict of Interest: Nil

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