

## Pediatric extremity rhabdomyosarcoma—A diagnostic dilemma

Yogita Devi<sup>1</sup>, Meena Anand Pangarkar<sup>2</sup>, Radhika Praveen Pagey<sup>3</sup>

From <sup>1</sup>Junior Consultant, <sup>2</sup>Head and Consultant Oncopathologist, <sup>3</sup>Consultant Oncopathologist, Department of Pathology, National Cancer Institute, Nagpur, Maharashtra, India

### ABSTRACT

Rhabdomyosarcoma (RMS) is a common soft-tissue malignancy of childhood, accounting for approximately 7% of cancers in children. They arise from primitive mesenchymal cells of skeletal muscle differentiation and occur most commonly in the head-and-neck region. Despite aggressive approaches, the overall outcome remains poor. Here, we present the case of a 9-year-old girl who presented with complaints of swelling over the left forearm for 2 months. Magnetic resonance imaging revealed a well-defined lobulated mass in the flexor compartment of the left forearm, suggestive of soft-tissue neoplasm. A biopsy done of the mass showed a malignant round blue cell tumor. Immunohistochemistry led to the diagnosis of RMS. The patient was started on multimodal therapy and is doing well on follow-up. Extremity swelling in the pediatric age group with malignant round cell morphology can pose diagnostic difficulty. A detailed work-up is essential for an accurate diagnosis. This case report emphasizes the role of a multimodality approach to the diagnosis and treatment of pediatric RMS.

**Key words:** Malignant round cell tumor, Pediatric rhabdomyosarcoma, Rhabdomyosarcoma, Soft-tissue malignancy

Rhabdomyosarcoma (RMS) is a common soft-tissue malignancy of childhood and adolescence, accounting for approximately 7% of cancers in children [1]. They arise from primitive mesenchymal cells committed to skeletal muscle differentiation and occur most commonly in the head-and-neck region, genitourinary tract, retroperitoneum, and less commonly in the extremities [2]. The World Health Organization recognizes four morphological subtypes and disease classification by molecular studies has refined it into “Fusion positive” and “Fusion negative” RMS with fusion positivity imparting a poorer outcome [3,4]. Despite aggressive approaches, including surgery, combination chemotherapy, and radiotherapy, the overall outcome remains poor for locally advanced, recurrent, and metastatic disease [5].

We are presenting this case to reiterate the importance of immunohistochemistry (IHC) in the diagnosis of malignant round cell tumors, as well as, the role of multimodal therapy in locally advanced RMS.

### CASE REPORT

A 9-year-old girl presented with complaints of swelling over the left forearm for 2 months. She had no history of fever, weight loss, or trauma.


General examination and preliminary basic laboratory investigations were found to be within normal limits.

Magnetic resonance imaging (MRI) revealed a well-defined lobulated mass in the flexor compartment of the left forearm, measuring 13.4×4.1×3.5 cm inseparable from the adjacent forearm flexor muscles, closely abutting the left radius and ulna without any obvious cortical changes. It was also abutting the left radial vessels with a partial encasement, suggestive of soft-tissue neoplasm-Sarcoma/Nerve sheath tumor (Fig. 1). Computed tomography (CT)-guided biopsy done from the mass showed features of a malignant round blue cell tumor (Fig. 2).

The differential diagnosis of malignant round cell tumors in pediatric patients includes Ewing’s sarcoma, primitive neuroectodermal tumor, synovial sarcoma, RMS, non-Hodgkin’s lymphoma, small cell osteosarcoma, and neuroblastoma. IHC showed immunoreactivity to Myo-D1, myogenin, and desmin while were immunonegative to CD45, S-100, CD99, FLI-1, and NKX2.2, leading to the diagnosis of RMS (Fig. 3).

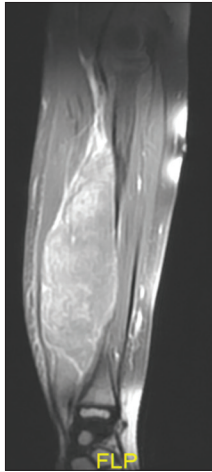
The patient underwent a positron emission tomography with 2-deoxy-2-(fluorine-18) fluoro-D- glucose integrated with CT (F-FDG PET/CT) scan as part of baseline evaluation, which corroborated the same findings and showed localized disease with no locoregional or distant metastasis.

The patient was started on neoadjuvant chemotherapy, after which a wide local excision of the left forearm tumor was

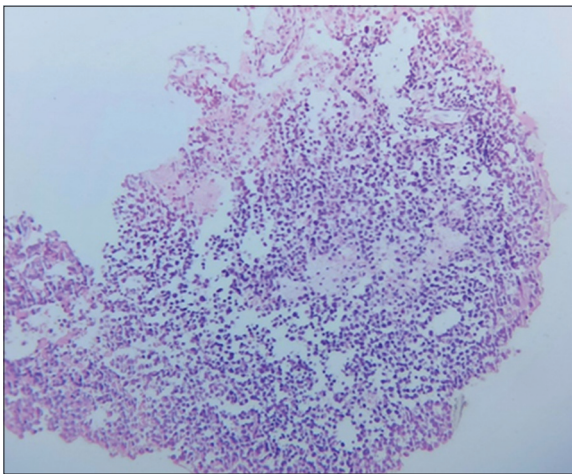
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**Correspondence to:** Dr. Yogita Devi, Department of Pathology, National Cancer Institute, Nagpur, Maharashtra, India. E-mail: yogitadevi.yd@gmail.com

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**Figure 1:** Magnetic resonance imaging shows a well-defined lobulated mass in the flexor compartment of the left forearm, measuring 13.4×4.1×3.5 cm, suggestive of soft-tissue neoplasm-sarcoma/nerve sheath tumor

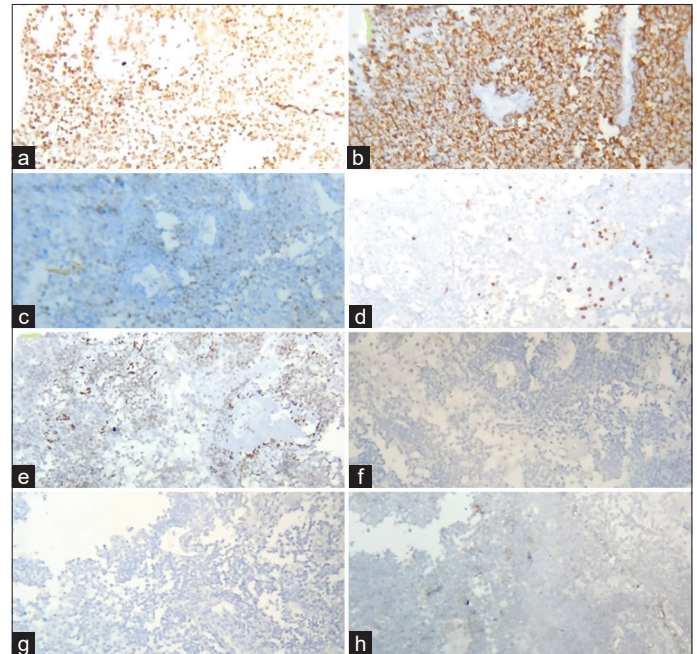


**Figure 2:** H and E stain at ×100 shows malignant round blue cells arranged in diffuse sheets

performed. Histopathology showed residual viable RMS with minimal treatment response. All surgical resection margins were free from tumors. She was further given adjuvant chemotherapy and radiotherapy and is doing well at 16 months of follow-up after primary diagnosis.

## DISCUSSION

Extremity swelling in a pediatric age group with malignant round cell morphology can pose diagnostic difficulties due to varied differential diagnoses. Hence, a detailed work-up including clinical examination, radiology, histopathology including IHC, and molecular studies is essential for an accurate diagnosis [6]. In our present case, light microscopy showed tumor cells in diffuse sheets; hence, differentiation between Embryonal RMS (ERMS) and the solid subtype of Alveolar RMS (ARMS) was not possible. However, by location, RMS of the deep soft-tissue of the extremity is more commonly ARMS and only rarely ERMS [7]. As per the molecular classification, the PAX3-FOXO1 and PAX7-FOXO1 gene fusions occur in 80% of cases



**Figure 3:** Immunohistochemistry stains demonstrate immunoreactivity to Myogenin (a), Desmin (b), Myo-D1 (c), while immunonegativity to CD45 (d), FLI-1 (e), NKX 2.2 (f), S 100 (g) and CD 99 (h)

with the alveolar subtype and are more predictive of poor event-free survival than histologic classification [8]. Unfortunately, due to pecuniary constraints, fusion status could not be evaluated for this patient.

Combined modality therapy, including surgery, chemotherapy, and radiotherapy, is the standard of care in pediatric RMS with localized disease [9,10]. Since our patient was diagnosed at an early stage with no nodal/distant metastasis and prompt institution of therapy was done, followed by complete surgical excision and adjuvant treatment; therefore, her prognosis is considered to be quite favorable.

## CONCLUSION

This case report emphasizes the evolving role of a multimodality approach to diagnosis as well as treatment for pediatric RMS.

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