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Rare Extra-Axial Sacrococcygeal Embryonal Tumor with Multilayered Rosettes (ETMR) in a Toddler: Imaging Features and Follow-Up (Case Report)

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Abstract **Case Report**

Embryonal tumors with multilayered rosettes (ETMR) are rare, highly aggressive WHO grade 4 embryonal tumors of the central nervous system, usually arising intracranially. Extra-axial and extracranial presentations are exceptional and may mimic more common pediatric sacrococcygeal tumors. We report the case of a 3-year-old girl presenting with a presacral and subcutaneous sacrococcygeal mass. MRI and CT demonstrated a large heterogeneous solid-cystic lesion with marked diffusion restriction, pelvic extension, and regional lymphadenopathy, initially suggesting a malignant sacrococcygeal teratoma. Histopathologic and immunohistochemical evaluation confirmed the diagnosis of ETMR. After neoadjuvant chemotherapy, follow-up MRI revealed a marked partial response with significant tumor shrinkage, decreased enhancement and vascularity, and regression of lymphadenopathy. Therapeutic response was assessed using both RECIST 1.1 and RANO criteria, demonstrating a significant partial response. This case emphasizes the diagnostic value of diffusion-weighted MRI in identifying hypercellular pediatric tumors in unusual extracranial locations and expands the differential diagnosis of aggressive sacrococcygeal masses in young children to include ETMR.

Keywords: Embryonal tumor with multilayered rosettes; ETMR; Sacrococcygeal mass; Diffusion-weighted imaging; Magnetic resonance imaging; Pediatric tumor; Presacral lesion; Hypercellular tumor.

INTRODUCTION

The 2021 fifth edition of the World Health Organization Classification of Tumors of the Central Nervous System unified several previously distinct embryonal entities, including Ependymoblastoma and embryonal tumor with abundant neuropil and true rosettes (ETANTR), into a single molecularly defined entity: Embryonal Tumor with Multilayered Rosettes [1].

ETMR is a highly aggressive WHO grade 4 pediatric embryonal tumors characterized by multilayered neuroblastic rosettes and recurrent amplification of the C19MC microRNA cluster located on chromosome 19q13.42, considered its molecular hallmark [2]. Although ETMRs predominantly arise as intra-axial supratentorial or posterior fossa tumors in infants and very young children, extra-axial and extracranial localizations remain exceedingly uncommon and may represent a major diagnostic challenge [3].

In atypical extracranial locations such as the sacrococcygeal region, radiologic evaluation plays a pivotal role in lesion detection, tissue characterization, locoregional staging, therapeutic planning, and post-treatment surveillance. The imaging differential diagnosis in this region is broad and mainly includes malignant sacrococcygeal teratoma, yolk sac tumor, neuroblastoma, rhabdomyosarcoma, and Ewing Sarcoma [4]. Advanced multimodal imaging—particularly magnetic resonance imaging (MRI) with diffusion-weighted imaging (DWI)—is essential for identifying features suggestive of highly cellular embryonal tumors, including marked diffusion restriction, heterogeneous enhancement, necrotic-cystic components, invasion of adjacent pelvic structures, and nodal involvement [5]. In addition, MRI provides crucial information for surgical planning and enables reliable longitudinal assessment of therapeutic response after chemotherapy [6].

We report a rare case of primary extra-axial extracranial sacrococcygeal ETMR in a 3-year-old child, with emphasis on the contribution of multimodality imaging—including CT, conventional MRI sequences, and DWI—in narrowing the differential diagnosis, evaluating tumor extension, guiding management, and monitoring response to treatment.

CASE REPORT

A 3-year-old girl was referred for evaluation of a progressively enlarging left sacrococcygeal swelling. Physical examination revealed a firm, mildly painful sacrococcygeal mass associated with cutaneous fistulization and discrete inflammatory skin changes. Neurological examination was unremarkable, with preserved motor function and no sphincter dysfunction.

Pelvic magnetic resonance imaging (MRI) (Figure 1) demonstrated a large lobulated extra-axial soft-tissue mass centered within the left sacrococcygeal region, exhibiting both presacral and subcutaneous components, with a maximal craniocaudal diameter of approximately 9 cm. Significant locoregional mass effect was observed, including anterior displacement of the rectum and superior deviation of the uterus.

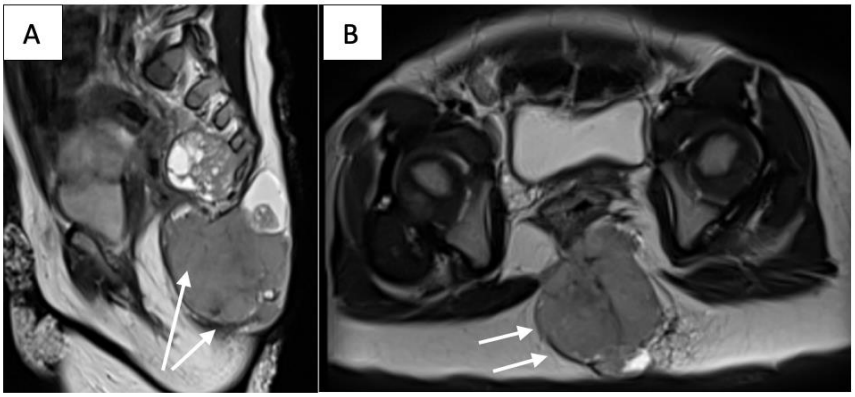

Figure 1: Initial MRI: Sagittal (A) and Axial (B) T2-weighted images showing a large heterogeneous mass (arrows) extending from the subcutaneous sacrococcygeal region through a bony defect into the presacral space, displacing the rectum and uterus

The lesion displayed marked internal heterogeneity, composed of intermixed solid and cystic-necrotic components. On MRI, the solid portions were predominantly isointense relative to skeletal muscle on T1-weighted imaging (T1WI) and heterogeneously hyperintense on T2-weighted imaging (T2WI), whereas the cystic areas exhibited fluid-equivalent signal intensity. Following gadolinium administration, the solid components demonstrated moderate heterogeneous enhancement.

Diffusion-weighted imaging (DWI) (Figure 2) revealed marked hyperintensity of the solid tumoral portions on high b-value sequences (b = 1000 s/mm²), associated with markedly reduced apparent diffusion coefficient (ADC) values, reflecting pronounced restricted diffusion secondary to high tumoral cellularity and elevated nuclear-to-cytoplasmic ratio (Figure 2).

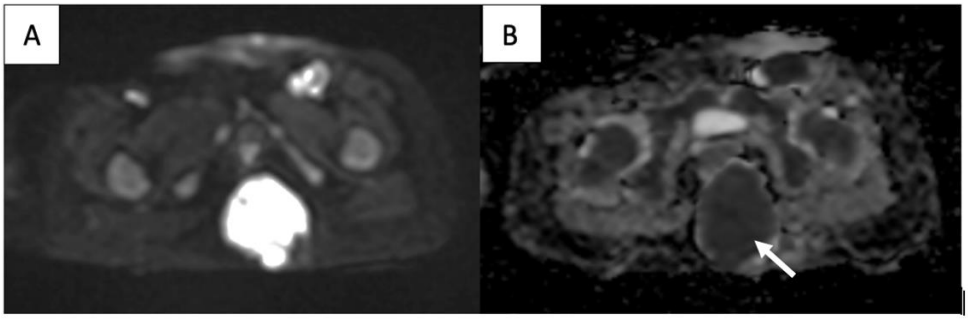

Figure 2: Initial MRI: Axial diffusion-weighted image (A) and corresponding ADC map (B) demonstrating marked restricted diffusion (low ADC, arrow) within the solid components, indicating high cellularity.

Regional staging demonstrated multiple enlarged inguinal and external iliac lymph nodes with diffusion restriction suggestive of metastatic involvement. A right iliac nodal conglomerate exerted ureterohydronephrosis without significant renal parenchymal alteration. No evidence of intraspinal extension, intradural invasion, vertebral marrow infiltration, or leptomeningeal dissemination was detected.

Thoraco-abdomino-pelvic computed tomography (CT) (Figure 3) was performed for metastatic staging and demonstrated no cortical bone destruction, pulmonary metastases, hepatic involvement, or other distant metastatic localizations.

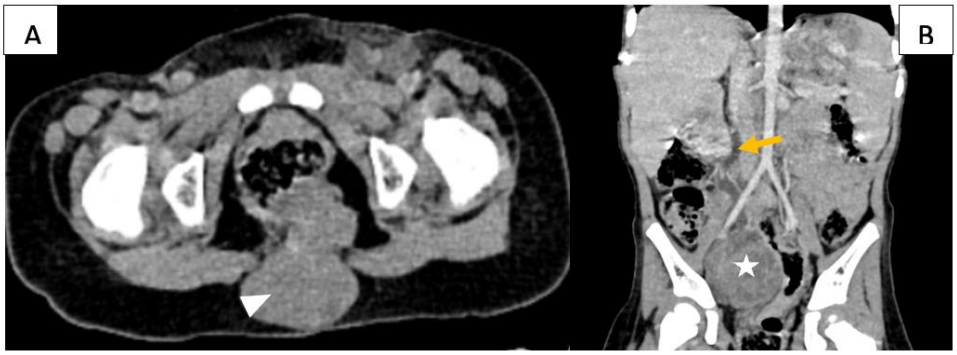

Figure 3: Initial CT – Extent and Complications. Axial (A) and Coronal(B) contrast-enhanced CT image demonstrating the presacral mass (arrowhead**) and the regional lymphadenopathy (asterisk) causing compression of the right iliac ureter with resultant moderate hydronephrosis (arrow)**

Based on the radiologic findings, the leading diagnostic consideration was a malignant sacrococcygeal germ-cell tumor, particularly Altman type III malignant teratoma with nodal dissemination. Differential diagnoses also included primitive neuroectodermal tumor (PNET), rhabdomyosarcoma.

Core needle biopsy was performed and the histopathologic examination revealed a highly cellular embryonal small round blue cell tumor with multilayered ependymoblastic rosettes and high mitotic activity. Immunohistochemistry showed focal CD99, NSE, and EMA expression, absence of GFAP staining, and a high Ki-67 proliferation index (~80%). FISH analysis confirmed C19MC amplification, establishing the diagnosis of Embryonal Tumor with Multilayered Rosettes, WHO grade 4.

Following four cycles of neoadjuvant chemotherapy, follow-up MRI (Figure 4) demonstrated a marked treatment response, with the largest dimension reduced from 9 cm to 5 cm [6]. The mass remained heterogeneous but showed a significant decrease in solid enhancing components, with an increase in cystic/necrotic areas. Residual solid tissue showed interval reduction in diffusion restriction consistent with therapy-induced decreased cellularity.

Regional lymphadenopathy had substantially regressed, though a right common iliac nodal conglomerate persisted, causing residual compression of the pelvic ureter with ongoing moderate hydronephrosis. Importantly, there was no evidence of spinal canal involvement, leptomeningeal spread, or osseous metastases.

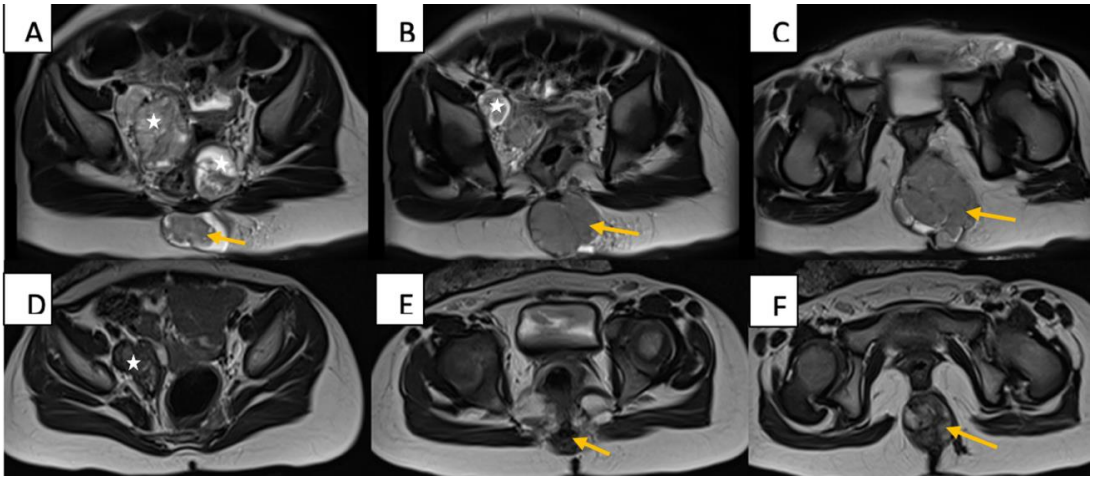

Figure 4: Baseline MRI (A–C) and post-chemotherapy follow-up MRI (D–F), axial T-weighted images, demonstrate a reduction in the size of the primary mass(arrow) and regional lymphadenopathy (asterisk), consistent with a partial treatment response

DISCUSSION

Embryonal Tumor with Multilayered Rosettes (ETMR) is an exceptionally rare and highly aggressive

pediatric embryonal neoplasm of the central nervous system, predominantly affecting children younger than 4 years of age [1]. Since the integration of molecular profiling into the WHO classification, ETMR has been defined by amplification of the C19MC locus at chromosome 19q13.42, frequently associated with LIN28A overexpression [1,2]. Most reported cases arise intracranially, particularly within the supratentorial compartment, whereas extra-axial sacrococcygeal localization remains exceedingly uncommon [3]. The present case therefore expands the limited spectrum of reported extracranial and spinal ETMR presentations.

Although imaging findings of ETMR have been mainly described in intracranial locations, radiology plays a particularly critical role in the management and longitudinal surveillance of these tumors rather than solely in their initial diagnosis. Due to the aggressive biological behavior of ETMR, imaging is essential for evaluating local tumor extension, treatment response, early recurrence, metastatic dissemination, and post-therapeutic complications [4]. In rare extra-axial sacrococcygeal presentations such as ours, MRI represents the cornerstone modality for follow-up because of its superior soft tissue contrast and multiplanar assessment of pelvic structures.

In previously reported ETMR cases, MRI commonly demonstrates large heterogeneous masses with mixed solid and cystic components, diffusion restriction related to hypercellularity, variable contrast enhancement, and occasional hemorrhagic or necrotic changes [2,5]. While these findings are not pathognomonic, serial MRI examinations provide valuable information regarding tumor evolution under therapy. Diffusion-weighted imaging (DWI) appears particularly useful during surveillance, as changes in apparent diffusion coefficient (ADC) values may reflect early cellular response before volumetric tumor reduction becomes evident [5]. In our case, MRI follow-up enabled accurate assessment of loco-regional progression and the relationship between the tumor and adjacent pelvic structures, information that was essential for therapeutic planning.

Radiology also plays a major role in detecting leptomeningeal dissemination, which is frequently associated with ETMR and significantly worsens prognosis [6]. Complete neuraxis MRI surveillance is therefore recommended during follow-up, even in patients with apparently localized disease at presentation. Given the rarity of extra-axial sacrococcygeal ETMR, no standardized imaging surveillance protocol currently exists; however, close interval MRI evaluation appears justified considering the high recurrence rate and rapid tumor progression reported in the literature [4,6].

Another important contribution of imaging in these rare tumors is the evaluation of treatment-related morbidity. Children undergoing multimodal therapy, including surgery and intensive chemotherapy with or without radiotherapy, are at risk for complications such as pelvic fibrosis, neurogenic dysfunction, growth disturbances, and therapy-induced neurotoxicity [7]. Longitudinal radiologic assessment therefore contributes not only to oncologic surveillance but also to the monitoring of functional and developmental outcomes in pediatric patients.

The prognosis of ETMR remains poor despite aggressive multimodal treatment strategies, with reported median survival frequently remaining below 12 months in several series [4,6]. Early identification of residual disease and recurrence through imaging surveillance may nevertheless help optimize therapeutic adaptation and improve supportive management. In this context, radiologists should be aware that ETMR may exceptionally present in extra-axial sacrococcygeal locations and should recognize the importance of structured MRI follow-up in the overall management of these patients.

Conclusion

Extra-axial sacrococcygeal ETMR is an exceptionally rare and highly aggressive pediatric tumor with a poor prognosis [1,4]. While molecular and histopathological analyses remain mandatory for definitive diagnosis, radiology—particularly MRI—plays a pivotal role in disease surveillance, assessment of therapeutic response, and early detection of recurrence or neuraxial dissemination [2,5,6]. In atypical presentations such as the present case, prolonged and structured imaging follow-up is essential because of the rapid progression and high recurrence rate associated with ETMR [4,6]. Awareness of these uncommon extra-cranial manifestations may improve multidisciplinary management and optimize follow-up strategies in pediatric patients [7].

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