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Embryonal Rhabdomyosarcoma in the Lumbar Region of a 2-Year-Old: When MRI Misleads

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Abstract **Case Report**

We report the case of a 2-year-old girl who presented with a subcutaneous mass in the right lumbar fossa. MRI revealed a well-circumscribed lesion with heterogeneous intermediate T2 signal intensity, peripheral enhancement after gadolinium injection, and diffusion restriction initially suggestive of an epidermoid cyst. Histological analysis confirmed an embryonal rhabdomyosarcoma. This case highlights the diagnostic challenge of soft tissue masses in children and the importance of correlating clinical, radiologic, and histologic data, especially when imaging findings appear deceptively benign.

Keywords: Lumbar mass, Pediatric soft tissue tumor, Embryonal rhabdomyosarcoma, MRI, Diagnostic pitfall, Epidermoid cyst mimic.

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INTRODUCTION

Soft tissue masses in children present a diagnostic challenge due to the broad range of possible etiologies, from benign lesions to aggressive malignant tumors. Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in pediatric patients, accounting for approximately 60–70% of all pediatric soft tissue sarcomas and about 5% of all pediatric solid tumors. Roughly one hundred new cases are diagnosed annually in France.

Involvement of the lumbar region is rare and may be misdiagnosed as benign entities such as epidermoid or dermoid cysts. MRI is the cornerstone of preoperative assessment; however, imaging interpretation can be misleading. We present the case of a 2-year-old girl with an embryonal RMS, initially interpreted on MRI as a benign epidermoid cyst.

CLINICAL PRESENTATION

A 2-year-old girl with no significant medical history was referred for evaluation of a painless, progressively enlarging subcutaneous mass in the right lumbar fossa over several weeks.

Lumbar spine MRI was performed using the following sequences: axial T2, axial T2 fat- saturated, coronal and sagittal T2 fat-saturated, axial STIR, axial T1, diffusion-weighted imaging (DWI), TRICKS, and post-gadolinium sequences in axial and sagittal planes (Figure 1).

Imaging revealed a well-defined, oval soft tissue mass in the subcutaneous tissues of the right lumbar fossa, measuring 28 x 16 x 37 mm. The imaging features were as follows:

- Isointense on T1-weighted images
- Heterogeneous intermediate signal on T2-weighted images
- No signal suppression on fat-saturated sequences
- Diffusion restriction
- Peripheral enhancement post-gadolinium injection Topographically:
- The mass displaced the right erector spinae muscle
- It was in contact with the right iliac wing
- It remained distant from the dural sac

Lumbar lordosis and vertebral alignment were preserved, with no abnormalities of vertebral height or signal.

An initial diagnosis of a benign epidermoid cyst was considered.

Surgical excision of the mass was performed. Macroscopically, the lesion appeared encapsulated.

Histopathological examination (figure 2) confirmed an embryonal rhabdomyosarcoma, with positive immunohistochemical staining for desmin and myogenin [3].

Given the localized and well-circumscribed nature of the lesion, and following multidisciplinary tumor board discussion, adjuvant chemotherapy was not initiated. Instead, the patient was placed under close radiologic surveillance.

Follow-up CT scans performed at regular intervals showed no evidence of local recurrence or distant metastasis at 6 months post-surgery. The postoperative course has been favorable to date.

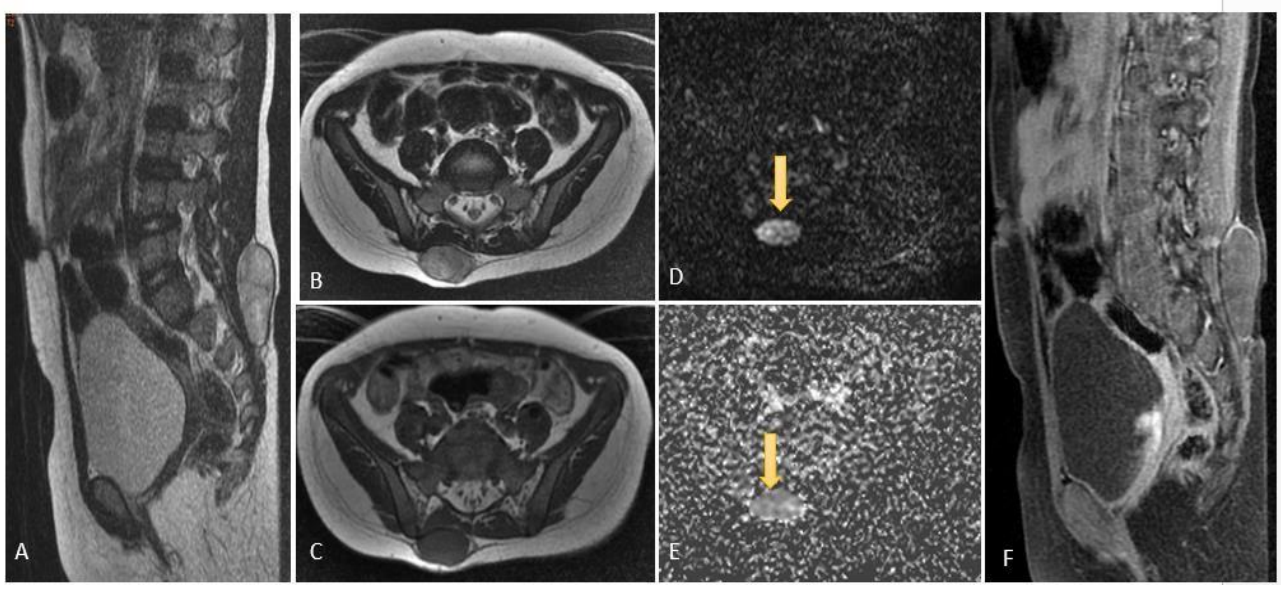

Figure 1: IRM lombaire : Sagittal T2 (A), Axiale T2 (B), Axiale T1 (C), Diffusion (D), ADC (E), Sagital T1FS injecté Oval, well-defined subcutaneous soft tissue mass in the right lumbar fossa (orange arrow), with regular margins. The lesion appears isointense on T1- weighted imaging (C), shows heterogeneous intermediate signal on T2-weighted imaging (B), with no fat suppression on fat-saturated sequences, diffusion restriction (D, E), and peripheral enhancement after gadolinium administration (F)

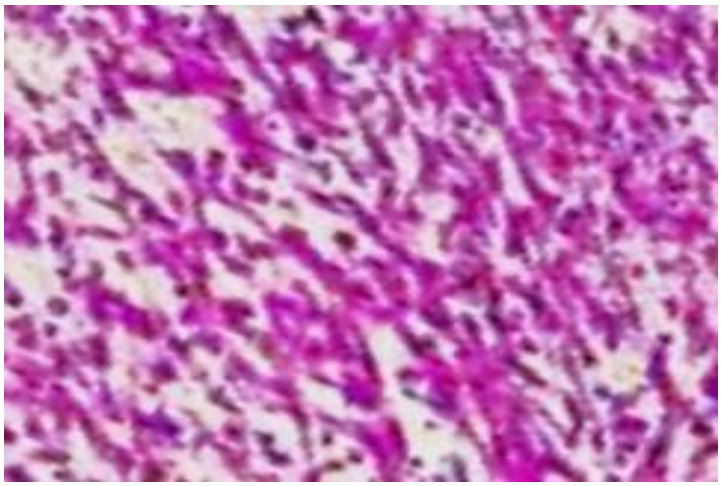

Figure 2: Histology shows a highly cellular tumor composed of small round to spindle cells in a myxoid stroma, with occasional rhabdomyoblasts. Immunohistochemistry is positive for desmin and myogenin, consistent with embryonal rhabdomyosarcoma

DISCUSSION

Rhabdomyosarcoma is a malignant mesenchymal tumor with striated muscle differentiation. The embryonal subtype is the most common in pediatric populations, with peak incidence in children under 10 years of age [1]. Common locations include the head and neck, genitourinary tract, and extremities. Lumbar or paravertebral involvement remains uncommon [2].

On MRI, RMS typically presents as a soft tissue mass that is iso- to hypointense on T1, hyperintense on

T2, and shows heterogeneous post-contrast enhancement with possible infiltration of adjacent structures. However, some cases may present with deceptively benign imaging features, including smooth margins, homogeneous peripheral enhancement, and lack of infiltration [2].

In our case, diffusion restriction often associated with high tumor cellularity was a key finding favoring a malignant process [4].

The accuracy of MRI in distinguishing benign from malignant soft tissue lesions in children is limited. Studies report that up to 30% of pediatric RMS cases may be initially misdiagnosed as benign lesions [2]. This case underscores the importance of maintaining a high index of suspicion and pursuing biopsy or excision when clinical or imaging features are atypical or incongruent.

CONCLUSION

In pediatric patients, soft tissue sarcomas such as rhabdomyosarcoma may mimic benign lesions, both clinically and radiologically. This case illustrates the need for close correlation between clinical, imaging, and pathological findings to establish an accurate diagnosis.

Even after complete surgical resection, close imaging follow-up is essential due to the risk of recurrence, especially in the presence of microscopic margin involvement. The favorable evolution in our case, under surveillance without immediate adjuvant therapy, also highlights the value of individualized, multidisciplinary management.

Informed Consent

The patient provided written consent for the publication of this case.

Conflicts of Interest: No conflicts of interest were declared by the authors.

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