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Extra Renal Rhabdoid Tumor: A Rare Cause of Congenital Soft Tissue Tumor

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Abstract

Rhabdoid tumors (RTs) are a well-defined entity in the kidney or central nervous system of infants or children. However, soft-tissue involvement is uncommon. It's an exceptional neonatal tumor of soft tissue. The imaging characteristics of this tumor are not specific. Biopsy allows diagnosis; the histomorphological characteristics of rhabdoid tumors, their immunoreactivity to epithelial markers and vimentin, and the INI-1 loss are important tools for diagnosis. RT tumors are aggressive and have a rapidly fatal clinical course in most cases. Despite multidisciplinary therapy, the survival rate is very low. We report a rare case occurring in a male neonate who presents at birth with a voluminous right axillary mass. A CT scan showed a well-limited tumor mass with lobulated contours. An ultrasound-guided biopsy was performed on day 8, showing the morphology and immunoprofile of RT. The mass showed rapid growth. The child was admitted for respiratory distress at 3 weeks. A thoraco-abdominal CT showed an increase in the size of the mass with the appearance of multiple lymph nodes and pleural, hepatic, and renal metastases. The child died two days later.

Keywords

Congenital, Rhabdoid Tumor, Soft Tissue, Diagnosis, Immunohistochemistry, INI-1

1. Introduction

Soft tissue tumors account for approximately 25% of neonatal tumors and are most often benign. Malignant tumors are rare, and the rhabdoid tumor (RT) is exceptional [1]. These tumors are a well-defined entity in the kidney or central

nervous system of infants or children. However, soft-tissue involvement is rare [2]. Few cases were reported in newborns [3]. Physicians confronted with RTs face two main challenges: reliability of diagnosis and treatment strategies [4]. In fact, the imaging characteristics of this tumor are not well determined [5]. The diagnosis is based on the biopsy and, therefore, on the morphological and immunohistochemical analyses [6]. RT tumors are aggressive and have a rapidly fatal clinical course in most cases. Despite multidisciplinary therapy, the survival rate is very low. We report a rare case of a neonatal RT soft tissue tumor.

2. Case Report

A new-born male child was admitted for the management of a right neonatal axillary mass. The pregnancy was not monitored, and no obstetric ultrasound was performed. The delivery was vaginal. Clinical examination found a soft mass of the axillary region measuring around 10 cm and inducing the upper limb into abduction. There were no foci of coagulative and hemorrhagic necrosis in the mass, and there was no palpable thrill (**Figure 1**). The CT scan showed a right axillary tumor mass that was well limited, with lobulated contours, heterogeneous in spontaneous contrast, enhanced after contrast injection, and containing linear calcifications (**Figure 2**). An ultrasound-guidedpercutaneous core needle biopsywas performed. The histological analysis has been performed on formalin-fixed and paraffin-embedded tissue sections, with hematoxylin-eosin-saffron staining. It revealed a round cell proliferation arranged in sheets with abundant eosinophilic cytoplasm and hypertrophic nuclei with atypical mitoses. Immunohistochemical analysis was performed on a 4 µm tissue section from formalin-fixed and paraffin-embedded blocks, using primary antibodies according to the manufacturer's guidelines, with immunohistochemical stainers (Ventana Ventana BenchMark ULTRA). The analysis showed a loss of INI-1 staining by tumor cells, which led to the diagnosis of a rhabdoid tumor. The mass showed rapid growth. The child was admitted for respiratory distress at 3 weeks. A thoraco-abdominal CT showed an increase in the size of the mass with the appearance of multiple lymph nodes, pleural, hepatic, and renal metastases (**Figure 3**, **Figure 4**). The child was intubated due to worsening of respiratory distress and died two days later.

3. Discussion

Soft tissue tumors account for approximately 25% of neonatal tumors and are most often benign. Malignant tumors are rare, and the rhabdoid tumor (RT) is exceptional [1]. Only a few cases of RT of soft tissue were reported in newborns [3]. RTs are a well-defined entity in the kidney or central nervous system of infants or children. However, soft-tissue involvement is rare [2].

RT was first described in 1978 by Beckwith and Palmer in the kidney as a sarcomatous variant of Wilms tumor [7]. In 1981, Hass *et al.* classified it as a distinct histopathologic entity of kidney tumors. It was originally called "rhabdoid"

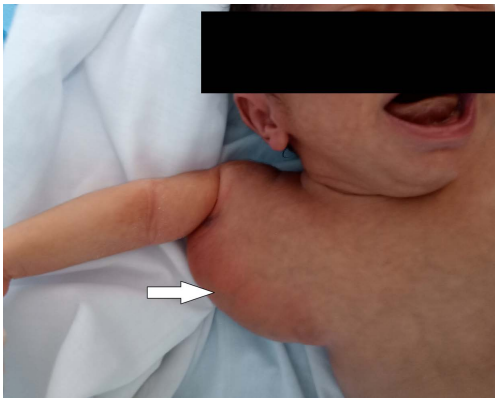

Figure 1. Clinical aspect of the mass at birth.

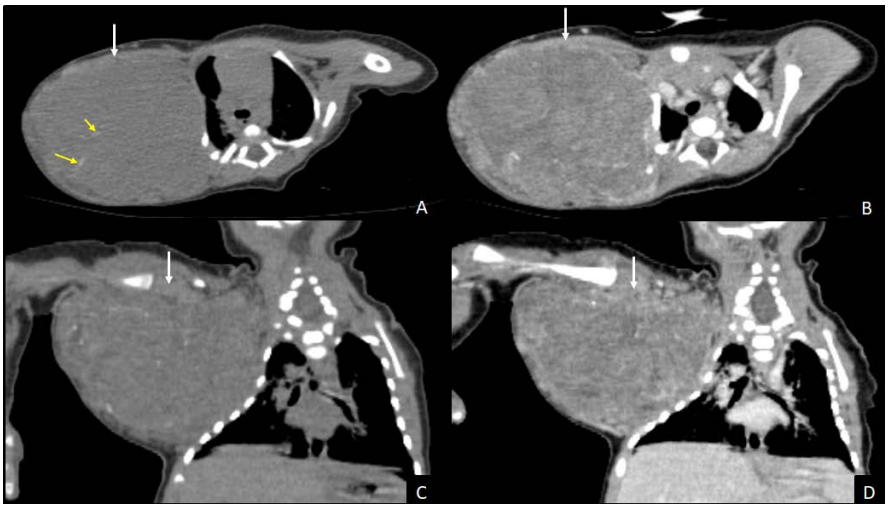

Figure 2. Admission CT scan: axial (A - B) and coronal (C - D) section: showing a mass of the right axillary region (white arrows), well limited with lobulated contours, heterogeneous in spontaneous contrast (A - C), and enhanced after contrast injection (B - D) containing linear calcification (yellow arrows).

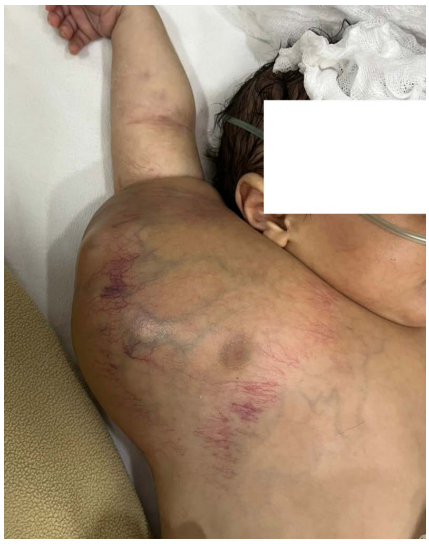

Figure 3. Clinical aspect at 3 weeks showing an increasing of the tumor size.

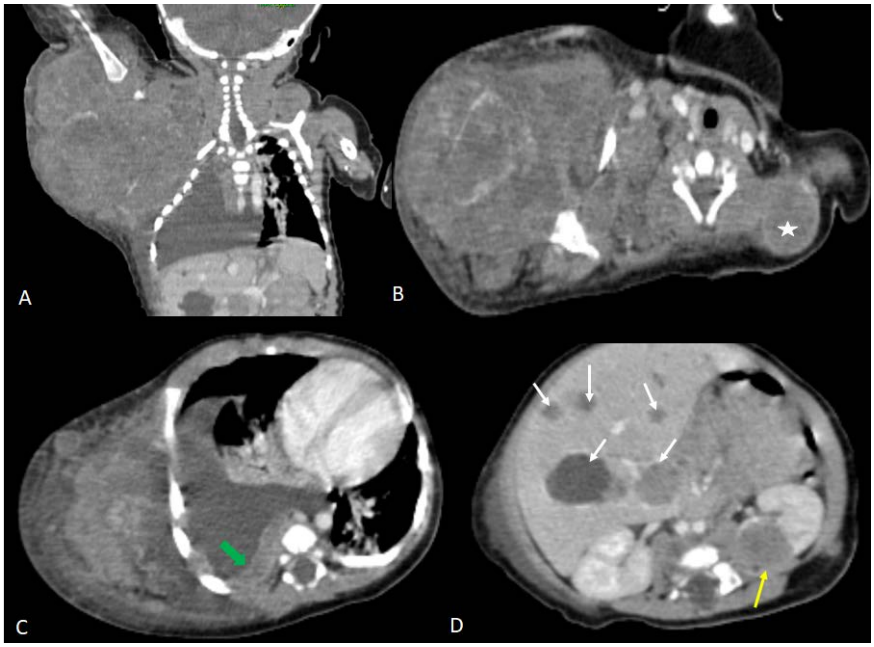

Figure 4. CT scan with contrast at 3 weeks: coronal (A) and axial (B - C - D) section showing an increase in size of the mass with appearance of multiple lymph node (star), pleural (green arrow), hepatic (white arrows) and left kidney (yellow arrow).

because of the resemblance of this tumor to rhabdomyosarcoma on light microscopic examination, and the lack of immunohistochemical and ultrastructural rhabdomyoblastic features led to it being referred to as a "rhabdoid" tumor [8]. Gonzalez Crussi *et al.* [9] reported in 1982 cases of extrarenal RT in infants involving the liver and the chest wall. Actually, the WHO book defines, morphologically, a rhabdoid tumor as a tumor containing a population of "rhabdoid" cells that are large with abundant cytoplasm, perinuclear spherical inclusions, and eccentric vesicular nuclei with large inclusion-like nucleoli, and genetically, by a bi-allelic inactivation of the tumor suppressor gene SMARCB1 (previously named hSNF5, BAF47, and INI-1) located on chromosome 22q11.2, secondary to inactivating mutations, deletions, or duplications of exons, resulting in a stop codon. This is revealed immunohistochemically by a complete loss of nuclear staining of INI-1 [10].

The incidence of soft-tissue RT has been reported to be 0.26% [2]. Only a few cases were reported in the literature [6] [11]. The tumor predominately involves a deep axial location, such as the neck or the paravertebral region. Its size ranged from 1.5 cm to 25 cm, with a medium size of 7.5 cm [2] [6]. The imaging aspects are not specific. The authors report a tendency for large tumors and hypodenseness in CT scans and heterogeneous hyper-intensity on T2-weapons in MRI [5]. This aspect suggests that this tumor should be considered in the routine differential diagnosis of soft-tissue tumors in infants and new-borns. The diagnosis is based on the biopsy. The histomorphological characteristics of rhabdoid tumors, their immunoreactivity to epithelial markers and vimentin, and the INI-1 loss are important tools for diagnosis [6].

Physicians confronted with RTs face two main challenges: reliability of diagnosis and treatment strategies [4]. Therapeutic management remains multidisciplinary in the absence of any pre-established consensus. However, complete surgical excision is the treatment of choice when it's possible. Neoadjuvant, or adjuvant, chemotherapy is associated with radiation. Targeted therapy is under investigationbased on various epigenetic pathways including DNA and histone methylation, histone deacetylation, cell cycle arrest and antimitotic mechanisms [12]. This tumor remains aggressive with a poor prognosis. It demonstrates a rapidly increasing volume, with metastasis occurring in a few weeks or months. Series reported dissemination to the lungs, lymph nodes, and liver at the time of diagnosis, and a death in an average period of 6 months occurs in 90% of cases [2] [13].

4. Conclusion

We reported a rare case of congenital RT located in soft tissue. This tumor is uncommon and aggressive. It should be included in the differential diagnosis of congenital tumors. In morphologic and immunohistochemical analysis, the loss of the nuclear staining of INI-1 in rhabdoid morphology represents the key to a positive diagnosis. This tumor is aggressive and highlights difficulties in therapeutic management.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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