Case Report

Malignant Rhabdoid Tumor of the Kidney in an 11-Month-Old Child: Case Report and Literature Review

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Abstract: Malignant renal tumors account for 6% of childhood tumors, with rhabdoid tumors (RT) being rare and representing 2% of pediatric renal tumors. RTs have a survival rate of only 22% at 5 years and are commonly diagnosed with metastasis. They are associated with mutations in the SMARCB1 gene, but recent literature has pointed to the presence of epigenetic characteristics. The recommended treatment includes radiotherapy, chemotherapy, and surgery. Despite the rapid advancement in knowledge regarding tumors in recent years, RT still lacks sufficient studies on its treatment and behavior, resulting in a reserved prognosis. This case aims to describe an atypical instance of tumor invasion and expansion of a rare childhood renal tumor that affected the patient's venous drainage, a situation never before reported in the literature.

Keywords: Rhabdoid Tumor; Surgical Oncology; Malignant Neoplasm.

1. Introduction

Pediatric tumors are rare but account for the second leading cause of mortality in children. According to estimates produced by the Instituto Nacional de Câncer José Alencar Gomes da Silva (INCA), from 2023 to 2025, there will be approximately 7,930 new cases of childhood cancer per year [1]. Childhood cancer differs from adult cancer as it is predominantly of embryonic origin with uncontrolled proliferation of abnormal cells [1], tending to have a shorter latency period, rapid growth, and high invasiveness, but responding better to chemotherapy [2].

The International Agency for Research on Cancer (IARC) has proposed a classification for tumors, in which renal tumors comprise group VI. Renal tumors are subdivided into: VIa (nephroblastoma and other non-epithelial renal tumors), which is further divided into nephroblastoma (Wilms' tumor), renal rhabdoid tumor, clear cell sarcoma, and peripheral primitive neuroectodermal tumors (PNET) of the kidney; VIb (renal carcinoma); and VIc (unspecified malignant renal tumors). Other renal tumors, such as sarcomas (ectomesenchymoma) and teratomas, are classified in groups IX and X [3].

Malignant renal tumors represent 6% of childhood tumors, with 90% being Wilms' tumors (WT) [2]. Rhabdoid tumors (RT) are rarer, accounting for only 2% of pediatric renal tumors. Most cases occur in children under 1 year of age [4]. RT is classified within subgroup VIa as it was initially considered a variant of WT. However, it is now recognized

as a separate entity due to its poorer prognosis and classification as high risk in therapeutic protocols [2].

Rhabdoid tumors have a neuroectodermal origin, with a 5-year event-free survival rate of only 22% [5], and are often diagnosed with lymphatic and hematogenous metastases [6]. Histologically, they exhibit diffuse infiltrative growth, rounded cells with large eosinophilic cytoplasm, and nuclei with prominent nucleoli [6]. Immunohistochemically, the most important finding is the loss of INI1 expression, but there can also be positivity for desmin, S-100, CD99, and other antigens. Coexpression of vimentin with cytokeratins is generally observed [6]. RTs are associated with other primary cancers in the body due to genetic predisposition [7]. This predisposition is related to mutations and deletions in the SMARCB1/INI1 gene on chromosome 22q [8] and also to biallelic inactivating disruptions of SMARCA4, although the latter is rarer. These genes are subunits of the SWI/SNF complex that regulates gene transcription and DNA repair [9]. It is believed that this genetic alteration contributes to tumorigenesis, but the specific genes involved are still unknown [10]. The loss of SMARCB1 expression is highly sensitive but not specific, necessitating correlation with histological, immunophenotypic changes, patient age, and tumor location for clinical diagnostic evaluation [7].

The treatment of renal rhabdoid tumors has seen little progress, but currently, for all rhabdoid tumors, radiotherapy, chemotherapy (with vincristine, dactinomycin, cyclophosphamide, doxorubicin, ifosfamide, carboplatin, and etoposide), and surgery are recommended [7]. Surgery, which is the part of the treatment described in this case, is complex. Our objective was to technically describe the complexity of tumor resection in this case and to report how venous return through vascular recanalization can be effectively used in situations of tumor invasion.

2. Case Report

Infant, 11 months old, previously healthy, when the mother noticed an abdominal mass during a bath, was referred on September 30, 2023, to a reference hospital for investigation. The patient presented with subocclusive abdomen, hematuria, arterial hypertension, microcytic and hypochromic anemia. A CT scan revealed a mass in the right renal fossa measuring 67mmx84mmx146mm (APxLLxCC) with invasion of the renal capsule, right renal hilum, and infrarenal vena cava, extending to the right iliac fossa with suspected retroperitoneal lymph node metastasis (largest retrocaval node 16x25mm) and pulmonary metastasis (multiple solid bilateral nodules, the largest being 0.6 cm in the left lower lobe) (Figure 1). The patient had refractory hypertension that was difficult to control with medication. Suspected of having a thrombus in the vena cava caused by the tumor, a color Doppler ultrasound was performed, which showed a normal caliber retro-hepatic inferior vena cava, but the remaining segments of the inferior vena cava presented reduced calibers and no thrombi, confirming extrinsic compression.

Initially suspected of Wilms' Tumor, chemotherapy (CT) was started with Vincristine and Actinomycin, and a total nephrectomy was performed via laparotomy on November 13, 2023. During the laparotomy, after mobilization of the right colon using the Cattell Maneuver and accessing the right renal fossa, a right renal tumor extending to the right iliac fossa with invasion of the left infrarenal vena cava was observed, causing recanalization of the left gonadal vein. After careful mobilization of the tumor, which was in close contact with the right diaphragm, dissection of the hilum and ligation of the right renal artery and ureter were performed. The inferior vena cava at the bifurcation level was clamped without causing hemodynamic repercussions, along with clamping of the left infrarenal vena cava. After clamping, the tumor was resected *en bloc* and sent for pathological examination along with retroperitoneal lymph nodes. Vascular reconstruction was not performed, as venous return was already compensated through the left gonadal vein and left renal vein.

Figura 1. Cross-sectional CT scan with contrast showing a heterogeneous solid-cystic tumor without calcifications, with some areas suggestive of central necrosis in the right renal fossa measuring 67 mm x 84 mm x 146 mm with contrast enhancement in the early arterial phase and fading in the portal phase, indicating high tumor vascularization. It can be seen that the tumor mass completely invades the infrarenal inferior vena cava, sparing the abdominal aorta.
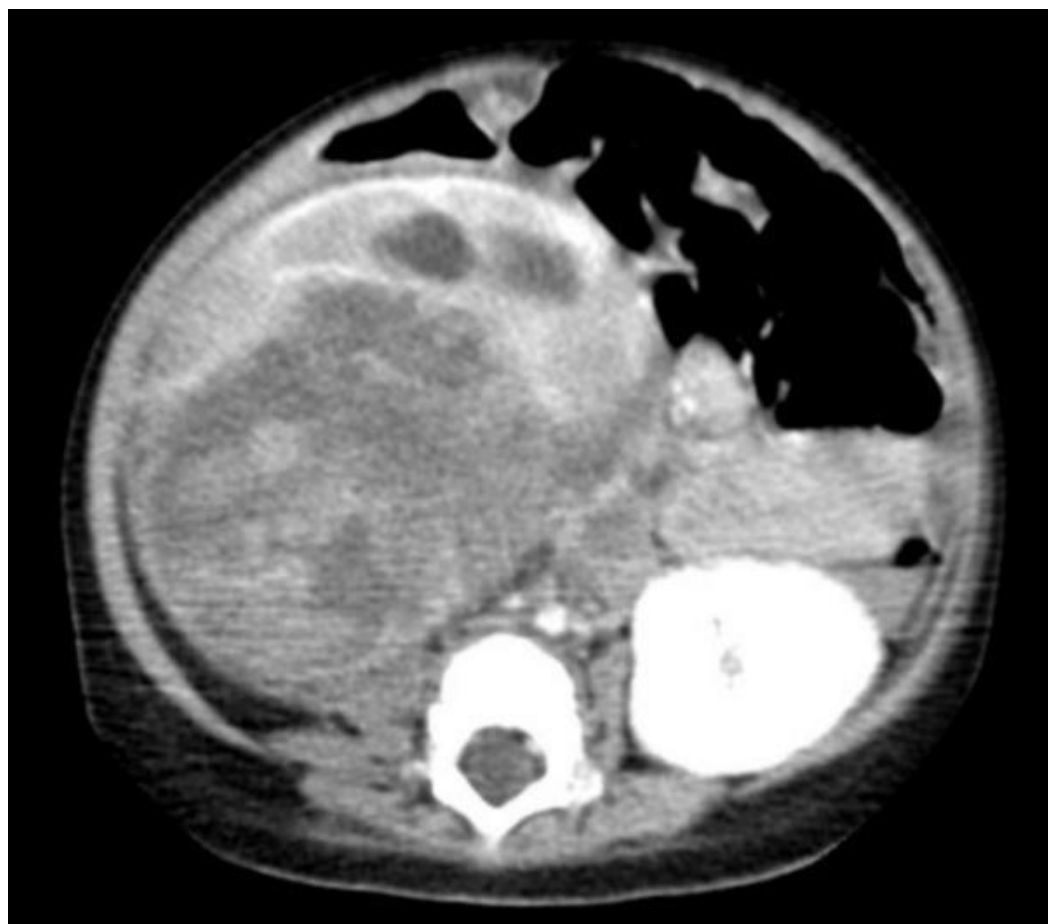

The pathological examination of the right kidney, measuring 13.2x10.2x7.9 cm, revealed an irregular solid multinodular lesion of 12.1x7.8x7.5 cm with necrotic areas comprising 30% of the tumor area. The tumor histology showed 66% blastema and no nephrogenic remnants. The tumor did not exhibit rupture, extension beyond the renal capsule, invasion of the right renal vein, ureter, adrenal gland, or adjacent organs. Thus, the diagnosis of malignant rhabdoid tumor of the kidney was confirmed with stage IV due to distant metastases.

Despite the successful surgery, pulmonary, peritoneal, and hepatic metastases did not respond to adjuvant chemotherapy with Doxorubicin, Vincristine, and Actinomycin, nor to radiotherapy. The patient, under pediatric palliative care, passed away on December 23, 2023, due to infectious complications.

3. Discussion

This is a clinical account of an uncommon instance of secondary syphilis, purportedly acquired through nonsexual means, in a 6-year-old male patient. Although the literature suggests that syphilis contracted through nonsexual routes is atypical [3-7, 11], the case discussed here plausibly represents such a mode of transmission.

Figura 2. In this image, we can see the complete anatomical architecture of the invasive rhabdoid tumor. It shows complete collapse of the infrarenal vena cava and full invasion of the proximal segment by the tumor, indicated by 1, 4, and 5. This vascular segment was likely completely non-functional, and venous return was primarily occurring through the left gonadal vein and left renal vein (3), given the caliber of these vessels and their direct drainage into the suprarenal vena cava, indicated by 2.
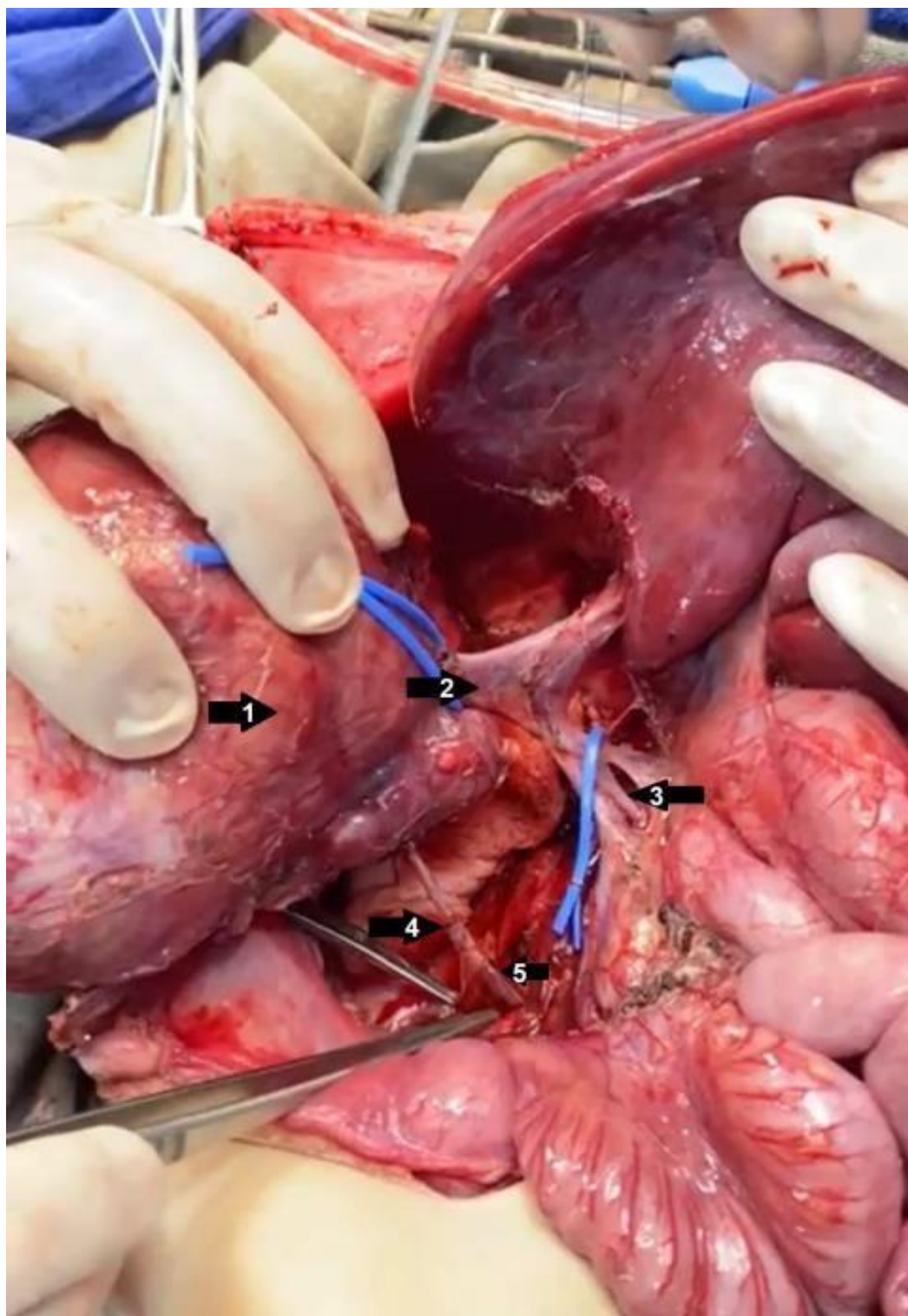
Rhabdoid tumors (RT) are rare, accounting for only 2% of pediatric renal tumors [4]. When renal, they can present with abdominal pain or agitation, macroscopic hematuria,

fever, hypercalcemia, and symptoms related to metastases [11]. Our patient presented with intestinal subocclusion due to tumor mass compression and macroscopic hematuria. At diagnosis, the patient was 11 months old, had metastases in abdominal lymph nodes and bilateral lungs, and presented with a tumor mass measuring 12.1x7.8x7.5 cm invading the renal sinus, pelvicalyceal system, and renal pelvis by contiguity. This is consistent with studies [12, 13] that show a grim prognosis when the age is <24 months and there are distant metastases. The tumor also invaded the inferior vena cava up to the left border of the vessel, which reduced venous flow through the common iliac veins and the inferior vena cava. Doppler ultrasound ruled out the presence of tumor thrombi, confirming that the obstruction to flow was extrinsic.

In the case report by Sharma et al. 2013 [11], an important similarity observed in our case was the initial hematuria. A significant portion of rhabdoid tumors that progress to the need for total nephrectomy have uncontrolled hematuria as the primary driver. Although it was not the patient's hematuria that prompted the surgery, we recognize it as the most morbid symptom associated with the tumor in this case, given the need for repeated transfusions.

During the laparotomy, it was observed that due to the tumor volume and its invasion into the infrarenal vena cava, there was an alteration in the patient's venous flow, with infrarenal venous return occurring primarily through the enlarged left gonadal vein, while the common iliac veins and the vena cava below the tumor mass were collapsed. There are no reports in the literature of altered venous drainage by renal rhabdoid tumors like the one presented. Unlike our case, Sharma et al. 2013 [11] reported intravascular tumor invasion by a tumor thrombus, favorably controlled by preoperative chemotherapy. The favorable tumor response to neoadjuvant therapy allowed a better surgical cleavage plane after 8 weeks. In our case, imaging studies showed no intravascular tumor invasion; only extrinsic compression without intravascular invasion was evident.

As observed with Shamberger et al. 2001 [14], preoperative chemotherapy can reduce postoperative complications, but it had no clinical significance when compared to primary surgical therapy. In the group that did not receive preoperative chemotherapy, 43% already had vascular wall involvement, and of the patients who received preoperative chemotherapy, 63% had tumor vascular invasion. In the present case, preoperative chemotherapy was administered over 4 weeks but did not result in any preoperative benefit. It is unclear if this result was due to the initial regimen directed at Wilms' variants or the ineffectiveness of preoperative therapy for these tumors.

Perhaps this adopted approach opens precedents for new discussions on the treatment of invasive malignant rhabdoid tumors that are not exclusively based on chemotherapy. The proposal of tumor cleavage with the possibility of vascular reconstruction could potentially expand treatment approaches for this type of tumor. Clearly, more observations are needed to establish more precise conclusions with the present case, but during the studied period, no organic or hemodynamic repercussions were noted in the patient that could be justified by the circulatory diversion performed after surgical resection.

As for major and long-term complications, potential renal overload could develop or perhaps pelvic and lower limb vascular pathologies due to impaired adequate venous return, as observed in the main postoperative complications listed by Shamberger et al. 2001 [14]. There is a scarcity in the literature of reports that bring discussions specifically about this intravascular involvement and extrinsic compressions in malignant renal rhabdoid tumors. Although the treatment of renal rhabdoid tumors is based on radical surgery associated with chemotherapy and radiotherapy, this traditional treatment has a poor prognosis [14]. Current literature suggests that RT should present epigenetic characteristics and heterogeneous gene expressions, requiring targeted therapies [12] for better survival.

Research [15, 16] demonstrates that SMARCB1 functions as a classical tumor suppressor due to its role in epigenetic regulation, cell cycle progression, and crosstalk sig-

naling, being the main factor responsible for the malignant pathophysiology of RT. Studies in mice have shown that homozygous inactivation of SMARCB1 is embryonically lethal, but heterozygotes are normal at birth, leading to cancer development 11 weeks after biallelic inactivation by a second hit at the SMARCB1 locus. Therefore, recurrent genetic deletions or lack of amplifications are necessary for cancer development, presenting an epigenetic character in pathogenesis.

More specific genetic studies were not performed on the patient in question due to the unavailability of epigenetic analysis and the still developing nature of these new treatment avenues.

5. Conclusions

Although knowledge regarding childhood tumors has rapidly advanced in recent years with the possibility of cure in 80% of cases [1], malignant renal rhabdoid tumors still lack studies on their treatment and behavior, resulting in a more reserved prognosis. Despite the peculiarity of the reported case, we recognize that primary surgery as a therapeutic arsenal, despite the tumor's complexity, can be an alternative despite an initially inadequate therapeutic response to chemotherapy. Although still modest and experimental, studies on targeted therapy and epigenetics could change the grim outlook of this specific tumor.

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