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Case Report

Wilms Tumor: Case Report

Tumor de Wilms: Relato de caso

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ABSTRACT

Wilms tumor is the most frequently occurring pediatric kidney tumor and it one of the most treatment-responsive tumors. The present study aims to report a case of Wilms tumor in a pediatric patient. A 4-year-old female child presented to the consultation with constipation and an abdominal mass on the right, which did not exceed the midline of the abdomen. The diagnosis was confirmed through ultrasound and computed tomography. Treatment includes surgery and chemotherapy, with the tumor being extremely chemosensitive. This report reiterates the importance of accurate and early diagnosis of Wilms Tumor, thus ensuring even better survival, given the advances already achieved in recent decades.

RESUMO

O Tumor de Wilms é o tumor renal pediátrico mais frequente e um dos mais sensíveis ao tratamento. Este estudo tem como objetivo relatar um caso de Tumor de Wilms em um paciente pediátrico. Trata-se de uma criança de 4 anos, do sexo feminino, que compareceu à consulta apresentando constipação intestinal e uma massa abdominal à direita, sem ultrapassar a linha média do abdome. O diagnóstico foi confirmado por meio de ultrassonografia e tomografia computadorizada. O tratamento inclui cirurgia e quimioterapia, sendo o tumor altamente quimiossensível. Portanto, este relato reforça a relevância de um diagnóstico precoce e preciso, essencial para otimizar os resultados terapêuticos e melhorar a qualidade de vida dos pacientes.

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INTRODUCTION

Wilms Tumor (WT), or nephroblastoma, represents the most common malignant pediatric renal tumor, accounting for 80% of all pediatric renal tumors within the SERVA group[1-3]. Regarding age distribution, 95% of cases occur in children younger than 10 years, predominantly under 5 years of age[1]. It presents either unilaterally or bilaterally in up to 13% of patients[4].

Although generally considered a sporadic tumor, Wilms Tumor has a genetic predisposition component in its pathophysiology, accounting for approximately 15% of cases. Various genetic mutations are implicated, including

alterations in the *WT1*, *p53*, *FWT1*, and *FWT2* genes, as well as mutations in the chromosomal region 11p15.5. Recent studies have also identified changes in the *CTR9*, *REST*, and *TRIM28* genes[1-3].

The survival rate is high and has significantly improved over recent decades, currently reaching approximately 90% globally. However, about 25% of cases involve subgroups of children with factors that may negatively influence this outcome, including anaplastic histology, certain molecular markers, bilateral involvement, and recurrence. Moreover, these subgroups are more susceptible to treatment-related complications such as renal insufficiency, infertility, cardiac toxicity, restrictive pulmonary disease, and secondary malignancies. Given this scenario, it is essential to minimize therapy-associated toxicity. Tumor recurrence occurs in 10% to 20% of cases, being most frequent within the first two years after diagnosis. Distant metastases are present at initial diagnosis in approximately 20% of cases, predominantly involving pulmonary metastases[1,4].

In most cases, patients present with a palpable abdominal mass, typically asymptomatic and large, identified either during routine clinical examinations or by caregivers themselves. Usually, the mass does not cross the midline and is located in the flank or lumbar region. Other symptoms may be present, including fever, urinary tract infection, hematuria, arterial hypertension, abdominal pain, nausea, vomiting, loss of appetite, shortness of breath, and constipation[5].

Imaging plays a crucial role in the diagnosis of WT, as well as in staging and surveillance. Diagnostic evaluation begins with ultrasonography (US), which confirms the presence and origin of the mass. The echogenicity of WT shows a wide spectrum and is highly variable, depending on tissue necrosis and intraluminal hemorrhage. After confirming the renal mass, Doppler ultrasonography is necessary to exclude vascular extension of the tumor, present in approximately 10% of cases[1].

Nevertheless, computed tomography (CT) or magnetic resonance imaging (MRI) of the abdomen with contrast are essential for locoregional staging. Additionally, chest CT, preferably with contrast, is recommended to evaluate for secondary implants due to the importance of assessing the pulmonary hilum and detecting possible thrombi[1].

Histology of Wilms Tumor (WT) is crucial for prognosis. According to the histological classification system of the Children Oncology Group (COG), there are three distinct histological groups based on the tumor's degree of anaplasia: favorable histology, focal anaplasia, and diffuse anaplasia. Favorable histology is the most common type, accounting for 75% of tumors, characterized by the absence of anaplasia and composed of blastemal, stromal, and epithelial cells[1]. Treatment of WT involves two different approaches, according to the COG and the European Société Internationale D'Oncologie Pédiatrique (SIOP), both demonstrating similar overall survival rates. Surgery and chemotherapy form the cornerstone of treatment, as WT is highly chemosensitive[1,6].

The COG approach is based on initial nephrectomy followed by chemotherapy and/or radiotherapy (as required), whereas the SIOP approach utilizes preoperative chemotherapy to reduce tumor size and facilitate surgical resection[1]. Although the SIOP and COG strategies differ regarding their initial treatment approach, both demonstrate comparable results in terms of overall survival (OS) for Wilms tumors, currently approaching 90%[6].

The aim of this paper is to describe a case of Wilms Tumor, presenting clinical aspects, diagnosis, surgical treatment, and postoperative outcomes. This case report was approved by the ethics committee of the Faculdade de Medicina de Campos under CAAE number 58700222.6.0000.5244. The approval report number is 5.566.247.

CASE REPORT

A female patient, aged 4 years, residing in Campos dos Goytacazes, presented for consultation with complaints of intestinal constipation and an abdominal mass. On physical examination, she exhibited good general condition; regular two-beat heart rhythm, normophonic heart sounds, no murmurs, heart rate of 120 bpm; normal cardiopulmonary auscultation; arterial blood pressure of 120/80 mmHg; respiratory rate of 30 breaths per minute; body weight of 13.5 kg; palpable abdominal mass on the right side, extending from the diaphragmatic dome to the midline of the abdomen. Abdominal ultrasonography revealed a nodular image with lobulated contours and cystic areas, measuring approximately 14 x 8 cm. Abdominal computed tomography (**Figures 1 and 2**) identified a large right renal tumor compressing adjacent structures, extending up to the diaphragmatic dome. Additionally, a secondary implant was observed at the base of the left lung, along with a small gallbladder calculus. Laboratory tests showed mild anemia, leukocytosis with monocytosis, elevated erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP), as well as ketones detected in urinalysis.

The child was referred to an oncology institution to complete the investigation and initiate treatment. Neoadjuvant chemotherapy (cyclophosphamide and doxorubicin) was prescribed according to the SIOP protocol. Subsequently, the patient underwent surgery (adrenal-nephro-ureterectomy, hepatic segmentectomy of segment 6, resection of thrombus from the inferior vena cava, retroperitoneal lymphadenectomy, resection of a diaphragmatic implant, as well as tumor resection and right-sided pulmonary metasta-

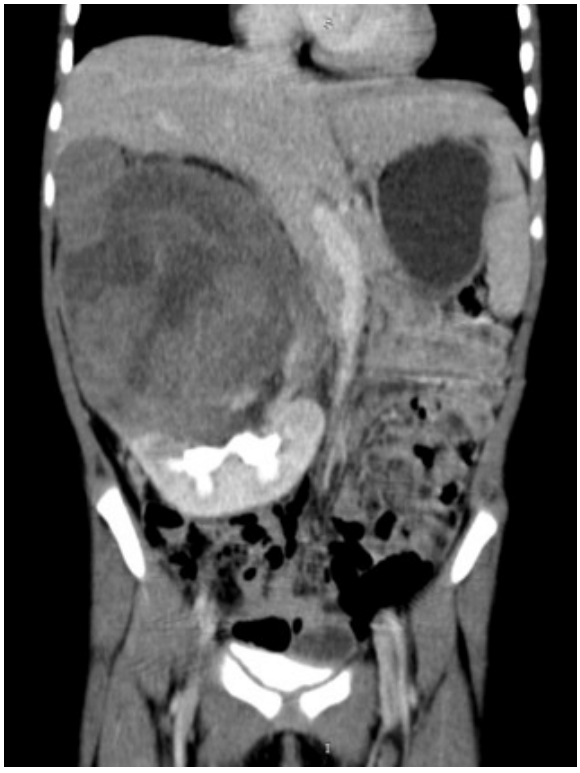

Figure 1. Coronal CT scan following intravenous contrast administration, demonstrating a large tumor originating from the upper third of the right kidney, with heterogeneous contrast enhancement and extensive central necrotic areas, extending superiorly up to the diaphragmatic dome and slightly crossing the midline by approximately 0.8 cm. The lesion measures 18.1 cm in the sagittal plane.

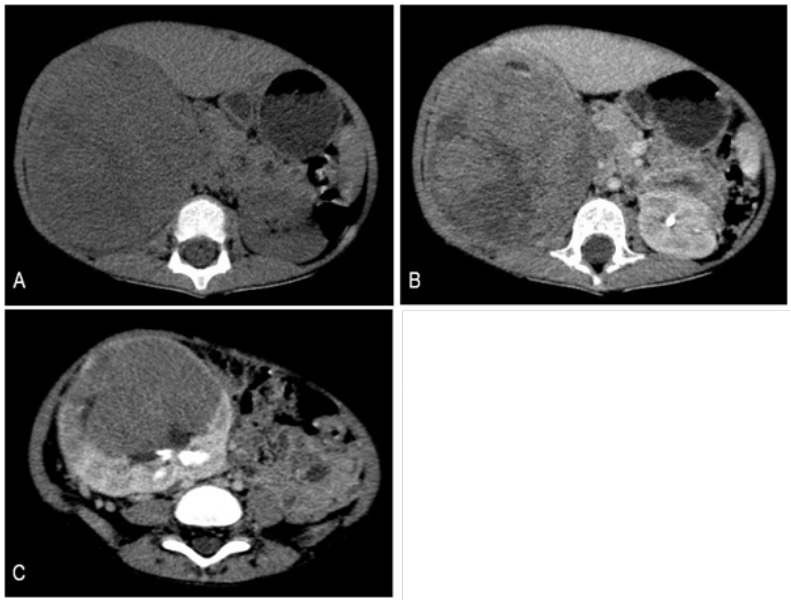

Figure 2. Axial CT scan without contrast (image A) and after intravenous contrast administration (images B and C), demonstrating central necrotic areas measuring approximately 9.5 x 11.0 cm, circumferentially involving the right renal vein and artery and displacing the inferior vena cava medially, without signs of thrombosis. Additionally, there is compression and displacement of the second portion of the duodenum and pancreatic head towards the left, as well as anomalous and enlarged vessels in the posterior right perirenal region.

sectomy), combined with adjuvant radiotherapy. Histopathological analysis demonstrated a predominantly stromal component (75%), epithelial component (13%), and blastemal component (2%). The tumor infiltrated the renal capsule, peri-renal adipose tissue, and involved the right adrenal gland. Multiple foci of perilobar nephrogenic rests were observed within the adjacent renal parenchyma, as well as chronic pyelonephritis with stromal fibrosis and glomerular sclerosis. Post-treatment histological classification indicated intermediate risk (stromal subtype), with a pathological staging of stage III. The patient showed clinical improvement during the postoperative period and continues outpatient follow-up at the institution.

DISCUSSION

Wilms Tumor (WT) is the most common abdominal neoplasm in the pediatric population, accounting for over 80% of cancers within this age group, and the overall survival rate exceeds 90%[1]. The 4-year-old patient falls within the age range with the highest incidence, which extends from 2 to 5 years. The clinical presentation was consistent with commonly reported findings: an asymptomatic abdominal mass, accompanied by constipation and elevated blood pressure for the patient's age.

The initial investigation of a suspicious renal mass in children should always start with ultrasonography (US). After confirming the renal mass, additional evaluation with contrast-enhanced CT or MRI is essential for staging purposes. The echogenicity of Wilms Tumor may vary widely, depending on the degree of tissue necrosis or intratumoral hemorrhage[1]. In this reported case, the CT imaging revealed a large tumor with heterogeneous enhancement and extensive central necrotic areas.

It is imperative that the radiologist carefully assesses the renal vein and inferior vena cava, as vascular extension of the tumor occurs

in approximately 10% of cases[1]. On the axial CT scan (**Figure 2**), extensive central necrotic areas are observed, circumferentially involving the right renal vein and artery and displacing the inferior vena cava medially, with no signs of thrombosis. Additionally, anomalous and enlarged vessels are noted in the posterior right perirenal region.

Additionally, metastases are present in 20% of WT cases at initial evaluation, with the pulmonary parenchyma being the most common site[1]. Chest CT assessment may be performed either with or without intravenous contrast, though contrast-enhanced imaging is recommended to evaluate the pulmonary hilum and potential intravascular thrombus. Ipsilateral pleural effusions can also be observed, frequently resulting from tumor rupture. Therefore, it is crucial to evaluate the lungs prior to nephrectomy, as basal atelectasis and pleural effusions are common in the postoperative setting and may compromise adequate assessment[1]. The liver and pelvic cul-de-sac should also be evaluated for signs of hepatic metastasis and peritoneal effusion, respectively[1]. In the patient described, a secondary implant at the left lung base was identified at diagnosis, with no evidence of hepatic metastases.

The main differential diagnosis must be made with neuroblastoma, a tumor originating from nerve cells and typically affecting patients younger than 10 years. Similar to Wilms Tumor, neuroblastoma may present as an abdominal mass, frequently occupying the renal fossa and extending towards the hypochondrium and flank, potentially crossing the abdominal midline[5].

Regarding radiological differences, neuroblastoma commonly demonstrates characteristic findings, such as calcifications in up to 90% of cases, as well as encasement of vascular structures without typically invading the vascular lumen. Neuroblastomas often cross the midline, extend into the spinal canal, and involve retroperitoneal lymph nodes. Bilateral masses are also more frequently seen with neuroblastoma, and bone metastases are common. It rarely arises from the kidney itself, whereas WT primarily originates from renal tissue[7]. Thus, a classical radiological sign associated with WT is the "claw sign," visible in image C of Figure 2, indicating the renal origin of the tumor, characterized by renal parenchyma surrounding the tumor mass. Conversely, neuroblastomas, arising from adrenal tissue, typically displace rather than encircle the kidney[7].

In contrast, calcifications are observed in only about 15% of cases of Wilms Tumor (WT). More frequently, vascular invasion occurs, alongside a higher prevalence of findings such as hemorrhage and necrosis, as illustrated by the presented case[7]. Additionally, unilateral involvement, renal origin of the tumor mass, and the presence of pulmonary metastases represent the most common presentations of WT, all of which were identified in this study.

Lastly, it is important to highlight that this case report not only provides updated information regarding Wilms Tumor and its imaging features but also emphasizes the necessity of early suspicion and diagnosis to ensure successful therapeutic outcomes.

AUTHOR CONTRIBUTIONS:

ECBM and EBOS conducted the study conception and design, data analysis, and manuscript writing. EBS, MJSMS, and AGMJ performed data collection, statistical analysis, and critical review of the manuscript. RCSCF performed the final text revision. All authors read and approved the final manuscript version and agree to take responsibility for its content.

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CONFLICT OF INTEREST:

We wish to confirm that there are no known conflicts of interest associated with this publication and that no significant financial support has influenced its results.

DECLARATION REGARDING THE USE OF GENERATIVE AI:

The authors declare that generative artificial intelligence tools (such as ChatGPT, Grammarly, Deepseek, etc.) were not used in the preparation of the manuscript. However, the editorial board made the decision to utilize ChatGPT, an AI language model developed by OpenAI, for the translation of this manuscript from the original language, Portuguese, to English.

REFERENCES

1. Servaes SE, Hoffer FA, Smith EA, Khanna G. Imaging of Wilms tumor: an update. Pediatr Radiol. outubro de 2019;49(11):1441–52.
2. Pater L, Melchior P, Rübe C, Cooper BT, McAleer MF, Kalapurakal JA, et al. Wilms tumor. Pediatr Blood Cancer. maio de 2021;68(S2):e28257.
3. Maciaszek JL, Oak N, Nichols KE. Recent advances in Wilms' tumor predisposition. Hum Mol Genet. 20 de outubro de 2020;29(R2):R138–49.
4. Aldrink JH, Heaton TE, Dasgupta R, Lautz TB, Malek MM, Abdessalam SF, et al. Update on Wilms tumor. J Pediatr Surg. março de 2019;54(3):390–7.
5. INCA. Tumor de Wilms: Câncer renal raro que afeta crianças [Internet]. 2022. Disponível em: https://www.gov.br/inca/pt-br/assuntos/cancer/tipos/infantojuvenil/especificos/tumor-de-wilms
6. Prasad M, Vora T, Agarwala S, Laskar S, Arora B, Bansal D, et al. Management of Wilms Tumor: ICMR Consensus Document. Indian J Pediatr. junho de 2017;84(6):437–45.
7. Chung EM, Graeber AR, Conran RM. Renal Tumors of Childhood: Radiologic-Pathologic Correlation Part 1. The 1st Decade: From the Radiologic Pathology Archives. RadioGraphics. março de 2016;36(2):499–522.