# Wilms tumor in a 3.5-year-old boy – case report

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## Abstract

Wilms tumor is the most common primary renal malignancy in children, and 95% of Wilms tumors occur in patients under 10 years of age. Initial imaging of a renal tumor includes abdominal ultrasound, followed by CT and MRI to assess its stage. We present the case of a 3.5-year-old boy complaining of abdominal pain with visible abdominal asymmetry on physical examination. After imaging studies, a diagnosis of Wilms' tumor was made and treatment was initiated, which reduced the tumor volume by 91%, followed by nephrectomy. The boy experienced two tumor recurrences, which were treated, but a third recurrence occurred. Long-term survival for patients with nephroblastoma has steadily improved over recent decades and currently exceeds 85%. Neoadjuvant chemotherapy for Wilms' tumor reduces tumor size and preserves renal function. Imaging plays a crucial role in the initial diagnosis, staging, and monitoring of Wilms' tumor.

**Keywords**: Wilms' tumor, renal tumor, pediatric cancer

## Introduction

Wilms tumor (WT) is the most common primary malignant renal tumor in children, with an estimated incidence of 80% of all renal tumors and 7% of all childhood cancers [1, 2]. 95% of Wilms tumors occur in children under 10 years of age, with the vast majority presenting within the first 5 years of life. Most abnormalities and syndromes associated with Wilms tumor involve the genitourinary system [3]. Wilms tumor is typically a sporadic disease, although 10% of cases are associated with a genetic predisposition such as Beckwith-Wiedemann syndrome, Denys-Drash syndrome, or WAGR (Wilms tumor, aniridia, genitourinary anomalies, and mental retardation) [2, 4, 5]. Nephroblastomatosis is considered a precursor lesion for Wilms tumor and occurs in as many as 93–99% of bilateral cases [5]. Synchronous bilateral disease is detected in approximately 5% of patients with WT [6]. Wilms tumor is associated with mutations in multiple genes: WT1, p53, FWT1, and FWT2, as well as mutations at the 11p15.5 loci. The most common symptoms associated with Wilms tumor include abdominal distension, dull abdominal pain, and progressive weight loss with malnutrition [1]. Initial imaging of a renal tumor includes abdominal ultrasound to identify the primary site of tumor origin, followed by cross-sectional imaging of the chest, abdomen, and pelvis using computed tomography or magnetic resonance imaging to further assess the primary site and identify possible metastases. Distant metastases occur in 10–20% of cases, with the lungs being the predominant site of spread [6, 2, 7]. There are two main treatment strategies for Wilms tumor, according to the International Society of Paediatric Oncology (SIOP) and the Children's Oncology Group (COG). COG guidelines recommend surgery as the initial treatment before chemotherapy [8]. The treatment strategy for SIOP involves neoadjuvant chemotherapy to reduce tumor volume and thus the risk of perioperative tumor rupture [9, 8]. In both COG and SIOP protocols, diagnostic tumor biopsy is not standard of care, but may be performed in cases of atypical renal tumor presentation or equivocal biological or imaging findings [4]. Treatment of Wilms tumor typically involves multimodality therapy with surgery, chemotherapy, and radiotherapy. The chemotherapy regimen depends on the histology (favorable or unfavorable), stage, and the presence of unfavorable molecular alterations [4, 10].

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## Case description

A 3.5-year-old boy was admitted to the Provincial Clinical Hospital No. 2 in Rzeszów in April 2018 due to abdominal pain and asymmetry noted on physical examination. The boy also had hemorrhagic ecchymosis in the area of the left costal margin, a small, pale, non-itchy, small, macular rash on the chest near the sternum, and small, single, bilateral cervical lymph nodes up to 1 cm in size were found on the neck. On the left side, a palpable, firm, and painless mass bordered the midline in the epigastrium and mid-abdomen up to the umbilicus. Laboratory tests revealed elevated C-reactive protein and D-dimer levels. An ultrasound examination confirmed the presence of a large tumor in the left kidney, which fills the left upper abdomen from the aorta to the spleen, displacing the stomach and spleen upward. A heterogeneous, predominantly hypoechoic lesion measuring 119x98x114 mm was relatively well-defined, with visible vascularization and small fluid spaces. Abdominal computed tomography (CT) revealed a large, heterogeneous, low-density mass with a volume of 445 ml, arising from the left kidney. Additionally, a chest X-ray was performed, which ruled out pulmonary metastases. Following imaging studies, the disease was classified as stage III due to the concomitant involvement of the periaortic lymph nodes. Due to the large size of the tumor, it was decided to immediately begin preoperative chemotherapy according to the SIOP 2001 protocol. After prior implantation of a vascu-port, the boy was administered chemotherapy based on vincristine and actinomycin D. One month later, a follow-up CT scan was performed, which showed a 91% reduction (the initial volume was 445 ml, currently approximately 40 ml) of the tumor compared to its initial volume, regression in size and a change in the morphology of the left renal tumor as a result of a positive response to treatment, and a reduction in the para-aortic lymph nodes. Subsequently, a left nephrectomy was performed, and the patient was qualified for postoperative chemotherapy according to the SIOPEN 2001 protocol for stage II, HR, but without radiotherapy (blastemic form with moderate anaplasia). A follow-up ultrasound examination revealed an abnormal, polycyclic tumor mass within the surgical site, with small cystic spaces measuring 92x67x58 mm and abnormal vascularization. A CT scan confirmed a recurrence of Wilms tumor in the surgical site (a solid cystic tumor measuring 80x70x76 mm), with metastatic disease in the tail of the pancreas (40x27x32 mm) and involvement of the mesenteric lymph nodes. A 5-6 mm lymph node was visible in the mediastinum, with ambiguous characteristics and strong contrast enhancement. The boy was scheduled for second-line chemotherapy (ifosfamide, carboplatin, etoposide, topotecan). One year after completing second-line chemotherapy, follow-up examinations revealed lesions suggestive of a second recurrence in the site following the removal of the left kidney. Third-line chemotherapy was administered, including vincristine and irinotecan. The patient was disqualified from undergoing allo-HSCT due to failed stem cell separation. Radiotherapy was also administered concurrently with third-line chemotherapy. The patient recently underwent a follow-up ultrasound, which revealed a new, heterogeneous mass in the renal bed following nephrectomy. MRI revealed a solid tumor measuring 33x29x38mm in the renal bed, with heterogeneous, predominantly marginal signal enhancement after contrast enhancement. The tumor adheres to the psoas muscle, anteriorly to the pancreatic tail and gastric fundus, superiorly and posteriorly to the spleen, and inferiorly to the intestinal loops.

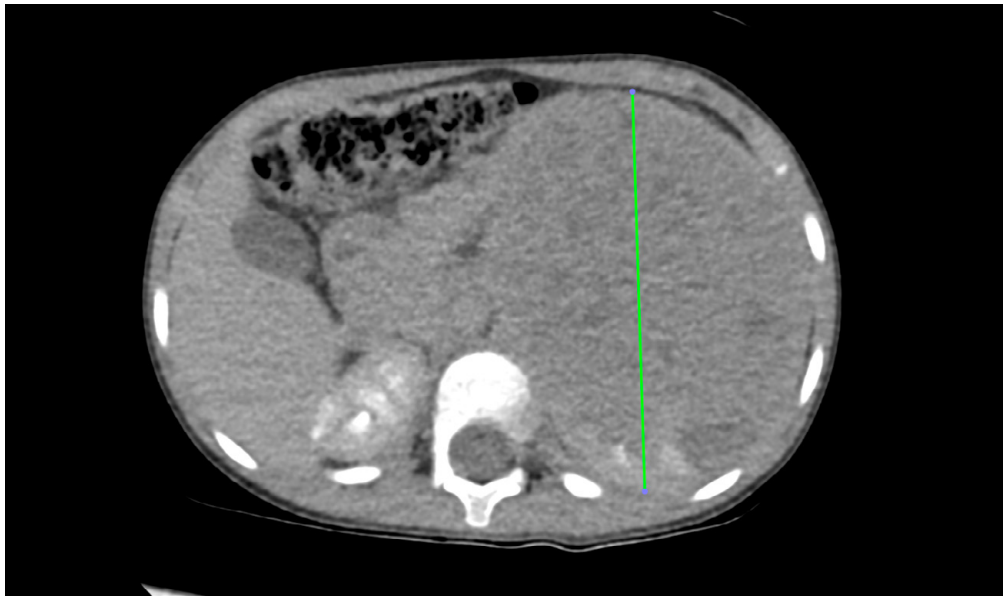

***Fig. 1*** *Diagnosis: CT scan with intravenous contrast agent administration. A large tumor mass was noted in the upper and left mid-abdominal regions Source: Own.*

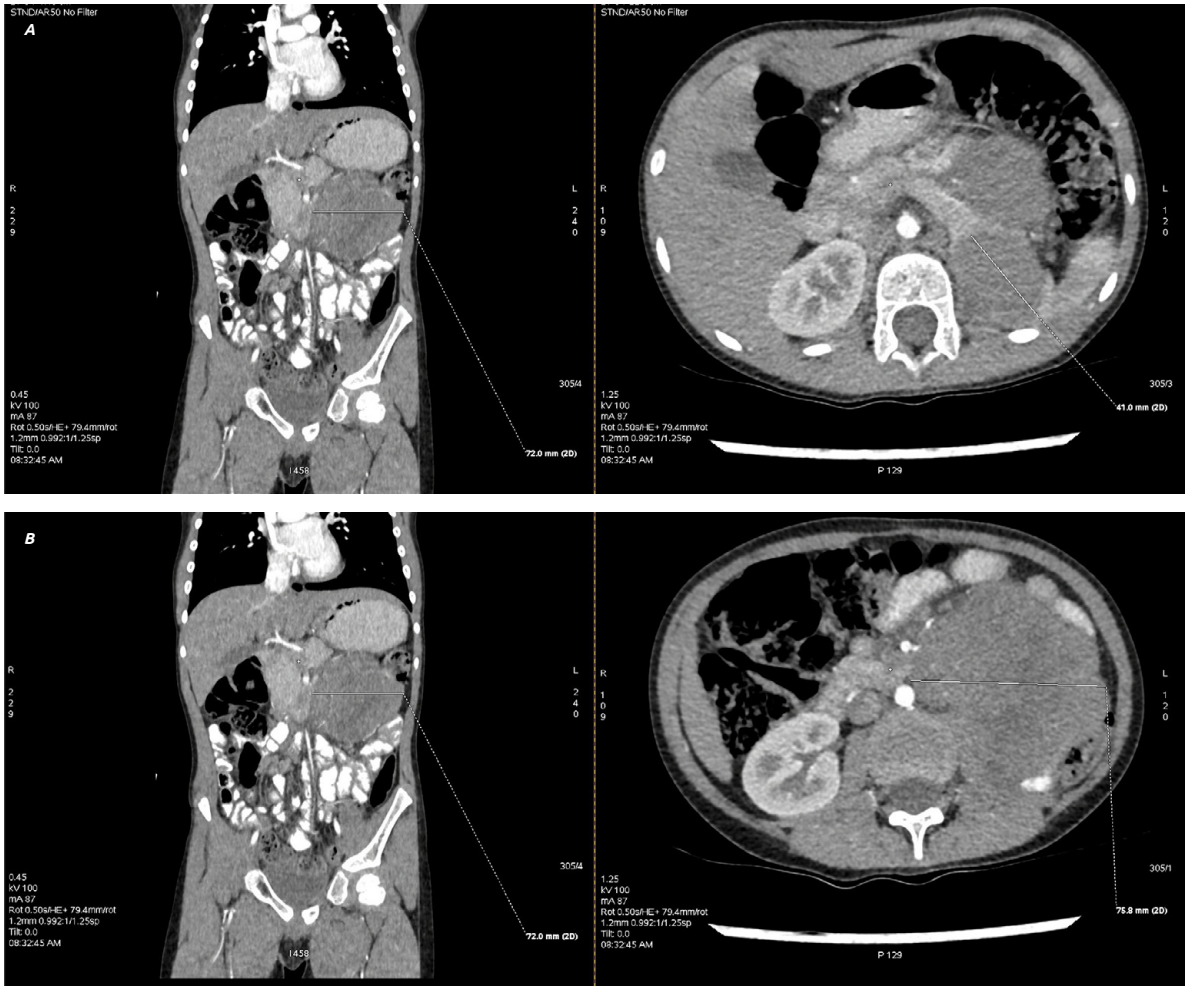


***Fig. 2 A, B*** *Recurrence 1: Coronal (A) and transverse (B) CT scans confirmed a recurrence of Wilms' tumor in the postoperative site (solid cystic tumor)*
*Source: Own.*

***Fig. 3*** *CT scan confirmed a metastatic lesion of Wilms tumor in the tail of the pancreas (solid tumor)*
*Source: Own.*

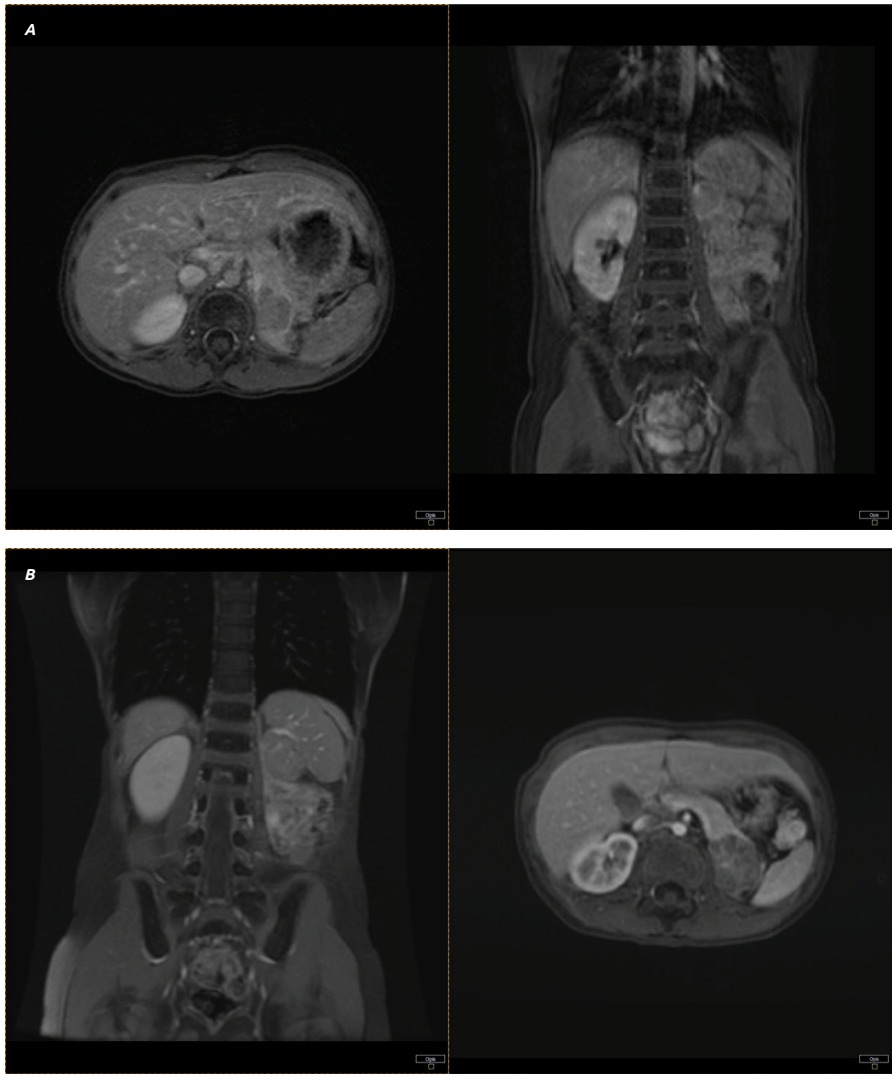


***Fig. 4 A, B*** *Recurrence 2: A fat-saturated SE(T1) MRI scan in the transverse (A) and coronal (B) planes from June 2021 confirmed the presence of a local recurrence in the nephrectomy site and the pancreatic tail*
*Source: Own.*

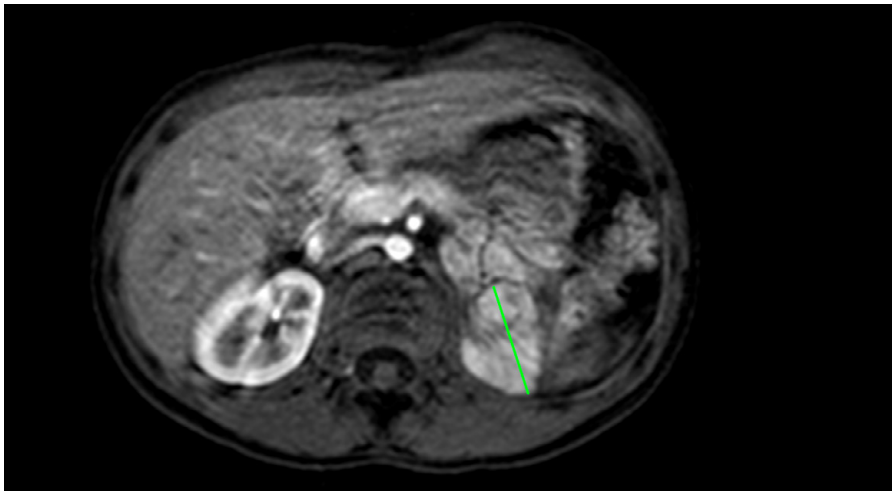

***Fig. 5*** *A fat-saturated SE(T1) MRI scan after contrast administration revealed a tissue structure consistent with tumor recurrence in the upper part of the left nephrectomy site adjacent to the pancreatic tail*
*Source: Own.*

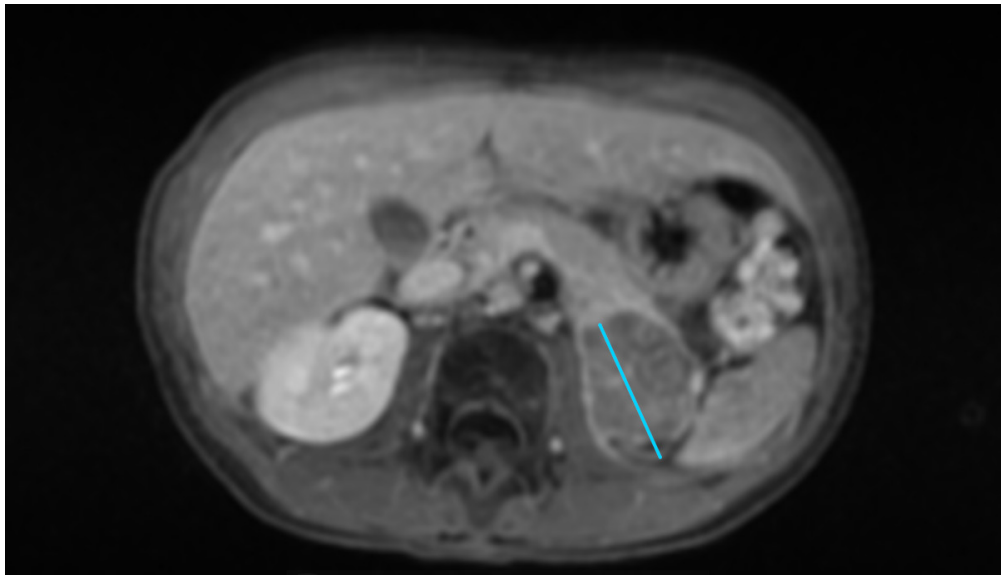

***Fig. 6*** *Recurrence 3: A fat-saturated SE(T1) MRI scan after contrast administration revealed a recurrence of Wilms' tumor (solid tumor) in the nephrectomy site, demonstrating heterogeneous, predominantly marginal MR signal enhancement*
*Source: Own.*

## Discussion

This case describes a 3.5-year-old boy with a stage II Wilms tumor, a blastemic form with moderate anaplasia. Wilms tumors most often have favorable histology, occurring in 75% of cases, while the anaplastic subtype of Wilms tumor, with a poor prognosis, accounts for approximately 5% of all pediatric renal tumors. It has been shown that in children undergoing preoperative chemotherapy (SIOP approach), a predominance of blastema after therapy is a poor prognostic factor [2]. In our patient, one of the unfavorable factors was tumor spread beyond the kidney, with involvement of the para-aortic lymph nodes.

The patient initially underwent an abdominal ultrasound for diagnostic purposes. Subsequently, abdominal CT and MRI were performed to further enhance the diagnosis. Both CT and MRI are important diagnostic imaging techniques because they allow for a precise assessment of the lesion, including its dimensions, size, and invasion of adjacent structures. Additionally, diffusion-weighted DWI MRI has been shown to aid in identifying high-risk blastemal WT and can be incorporated into the preoperative assessment of response to chemotherapy in bilateral WT [6].

The Polish Pediatric Solid Tumor Group examined the recurrence rate in children and found that 11.8% of children experienced recurrences, which were unrelated to the patient's age, histopathology, gender, or time of recurrence [11]. The prognosis for patients with metastatic recurrence was similar to that of those with local invasion. Overall survival after the first recurrence was 77%, but after the second recurrence, it dropped dramatically to 13% [10]. Long-term survival of patients with Wilms' tumor has improved over the past several decades and now exceeds 85%. The long-term effects of cancer therapy alone result in one-quarter of WT survivors suffering from kidney failure, infertility, myocardial damage, restrictive lung disease, and the development of subsequent malignancies [7].

## Summary

The symptoms of Wilms' tumor are delayed and nonspecific, delaying diagnosis. Imaging tests (ultrasound, CT and/or MRI) are the primary diagnostic tool for Wilms' tumor, as biopsy is not routinely performed due to the risk of spread or rupture. Correct diagnosis and prompt treatment are crucial for achieving good outcomes, which in developed countries reach 85%.

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