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Extrarenal Wilms tumor in the retroperitoneum of a 6-year-old girl: a case report and review of the literature

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Abstract

Introduction Extrarenal Wilms tumor is an extremely rare condition, typically documented only in isolated case reports. Unlike classical intrarenal Wilms tumors, extrarenal Wilms tumors arise outside the kidney, often presenting a diagnostic challenge owing to its unusual location and overlapping features with other retroperitoneal tumors. Its rarity necessitates further documentation to improve recognition and management. This report presents a case of extrarenal Wilms tumor located in the retroperitoneal space of a 6-year-old girl.

Case presentation A 6-year-old white Iranian girl presented with abdominal pain and swelling in the upper left abdomen. Physical examination revealed a firm, nontender, immobile mass. Ultrasound imaging identified a well-defined mass with significant necrotic and cystic areas. Abdominopelvic computed tomography scan showed a large mass on the left side of the abdomen, exerting pressure on the adjacent pancreas, spleen, and left kidney. The patient underwent laparotomy and received 19 weeks of chemotherapy, including actinomycin-D and vincristine. Post-treatment, she fully recovered and underwent monthly sonography follow-ups for 6 months after completing chemotherapy and has shown no signs of recurrence to date.

Conclusions Extrarenal Wilms tumor should be considered in the differential diagnosis of abdominal pain, especially in young children, owing to its rarity and the potential for misdiagnosis as other retroperitoneal tumors. A definitive diagnosis is made through surgical intervention followed by histopathological examination.

Keywords Extrarenal Wilms tumor, Nephroblastoma, Case report, Pediatric oncology, Abdominal mass, Retroperitoneal mass

Introduction

Nephroblastoma, commonly known as Wilms tumor, is the second most common abdominal tumor in children, following neuroblastoma [1]. Worldwide, about 1 in every 10,000 children is affected by this condition before the age of 15 years, with a median age of onset of 38 months in the USA, occurring slightly later in girls than in boys [2]. Wilms tumor makes up around 6% of all cancers in children and is responsible for over 95% of kidney tumors in this age group [3]. Additionally, around 10% of patients present with pulmonary metastases at diagnosis [4].

When this tumor originates outside the kidney, it is referred to as an extrarenal Wilms tumor (ERWT), a rare

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condition that has primarily been documented through case reports. Moyson *et al.* first described this condition in 1961 [5]. ERWT accounts for approximately 0.5–1% of all Wilms tumor cases, with just over 100 cases reported in the literature to date [6].

ERWT is characterized by both kidneys being normal, and the tumor is not connected to the kidneys, although it has a morphological similarity to the primary intrarenal Wilms tumor [7]. Its rarity and the uncommon locations where it can occur—such as the retroperitoneum, inguinal region, or spinal canal—make ERWT particularly challenging to diagnose and treat [1].

Most cases of ERWT are found in children with an average age of approximately 4 years and a slight predominance in females [8]. This report presents a case of ERWT located in the retroperitoneal space of a 6-year-old girl. By documenting this case, we aim to contribute to the limited body of knowledge on ERWT and provide insights that could aid in future diagnosis and management of similar cases.

Case presentation

A 6-year-old white Iranian girl presented with acute abdominal pain and swelling in the left upper abdomen that had been noticed for the past 2 days. Prior to this, she was asymptomatic. There was no reported history of nausea, vomiting, constipation, oliguria, hematuria, peripheral edema, or weight loss. Her medical history was unremarkable, with no prior medical interventions. There was no family history of cancer.

The child was born prematurely via cesarean section, with a birth weight of 2500 g. She was born to non-consanguineous parents, has achieved normal developmental milestones, and is up to date on her immunizations.

The child's vital signs were normal during the physical examination. Abdominal examination revealed a distended, firm, nontender, and immobile mass in the upper left abdomen. The mass was well defined and oval-shaped, and had smooth borders. No overlying skin changes, visible pulsations, or palpable thrill were noted. Routine laboratory tests, including a complete blood count, kidney and liver function tests, serum electrolytes, and urinalysis, were all within reference ranges.

Ultrasonography revealed a well-defined heterogeneous mass in the left upper abdomen, containing significant necrotic and cystic areas. The mass exhibited internal vascularity and measured $150 \times 112 \times 110$ mm^3, with no evidence of calcification.

An abdominopelvic computed tomography (CT) scan with intravenous contrast revealed a well-defined, large, heterogeneous enhancing mass on the left side of the abdomen. This mass exerted a mass effect on the adjacent pancreas (body and tail), spleen, and left kidney but was distinct from these organs (Fig. 1).

On the basis of imaging findings, the initial differential diagnoses included Wilms tumor, primitive neuroectodermal tumor (PNET), and sarcomatoid tumors. The well-defined, encapsulated nature of the mass, along with its separation from adjacent organs, was highly suggestive of Wilms tumor. However, histopathological confirmation was required to establish the final diagnosis.

There were no obstacles in accessing diagnostic investigations, and no financial or cultural factors influenced the diagnostic process in this case.

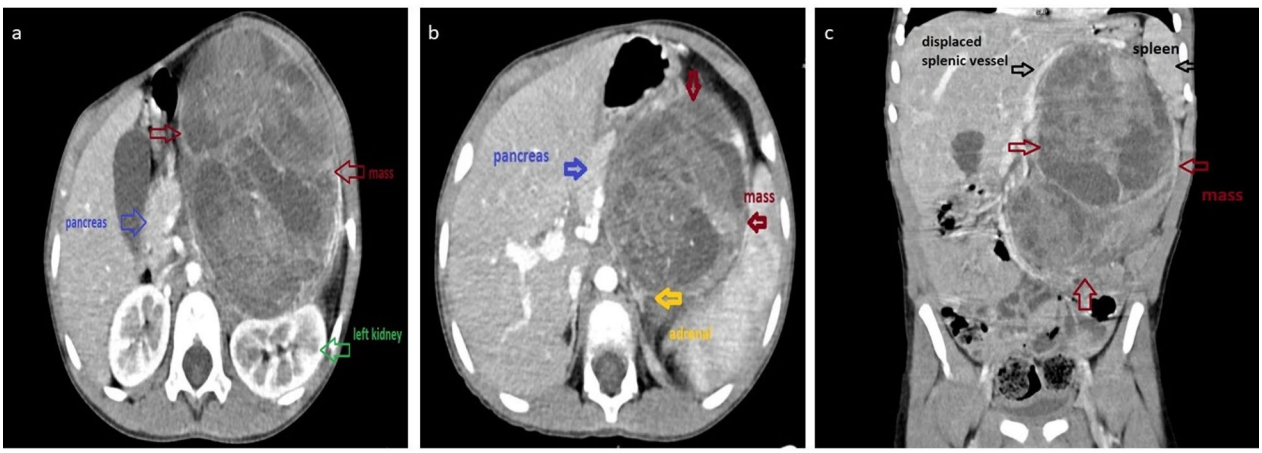

Fig. 1 In axial (**a** and **b**) and coronal (**c**) views of abdominal CT scan with IV contrast, a large heterogeneous enhancing mass with prominent cystic/necrotic components is identified in the left upper quadrant. The mass is distinct from surrounding structures, including the pancreas, left kidney, left adrenal gland, and spleen. It causes anterior displacement of the stomach and the body and tail of the pancreas. No evidence of direct invasion into adjacent organs is noted

The child underwent laparotomy, during which an encapsulated mass was identified on the left retroperitoneal side, distinct from the left kidney. The mass was removed and sent to the pathology department in a formalin container.

Macroscopic pathological examination revealed a round to oval, encapsulated, well-defined grey–brown rubbery mass covered by fibrofatty tissue. The mass weighed 800 g and measured $14.5 \times 9 \times 3.5$ cm^3. Cut sections of the mass displayed a solid cystic surface with variegated fibrotic bands. The cystic areas contained turbid yellowish fluid, while the solid parts exhibited a heterogeneous tan appearance with multiple necrotic and hemorrhagic foci.

Microscopic histopathologic examination revealed features consistent with Wilms tumor, including nests, cords, and tubules of epithelial cells surrounded by stromal spindle cells. No evidence of anaplasia or extension into the surrounding fibrofatty tissue was observed. An immunohistochemical (IHC) study for inhibin and calretinin yielded negative results. However, positive nuclear staining for *WT1*, combined with the absence of residual kidney parenchyma, led to the diagnosis of extrarenal Wilms tumor with favorable histology (Fig. 2).

According to the National Wilms Tumor Study (NWTS) guidelines, this case has been classified as stage II. Following surgery, the patient underwent 19 weeks of chemotherapy based on the EE4A regimen, which included actinomycin-D and vincristine. During the first 11 weeks, vincristine was administered intravenously (IV) at a dose of 1.5 mg/m^2 once weekly. Actinomycin-D was given on weeks 1, 4, 7, 10, 13, 16, and 19 at a dose of 0.045 mg/kg, also administered IV. From week 13 onward, the vincristine dose was increased to 2 mg/m^2 and was given IV once weekly during weeks 13, 16, and 19.

In our institution, the NWTS protocol is followed, prioritizing initial tumor resection before risk-adapted chemotherapy. In contrast, the SIOP protocol—developed by the Société Internationale d'Oncologie Pédiatrique (International Society of Paediatric Oncology)—prioritizes preoperative chemotherapy to shrink the tumor before surgical intervention. For our patient with an extrarenal Wilms tumor, we adapted the NWTS approach, focusing on tumor resection rather than surgical resection, tailoring it to the clinical presentation and aligning with our established institutional practice.

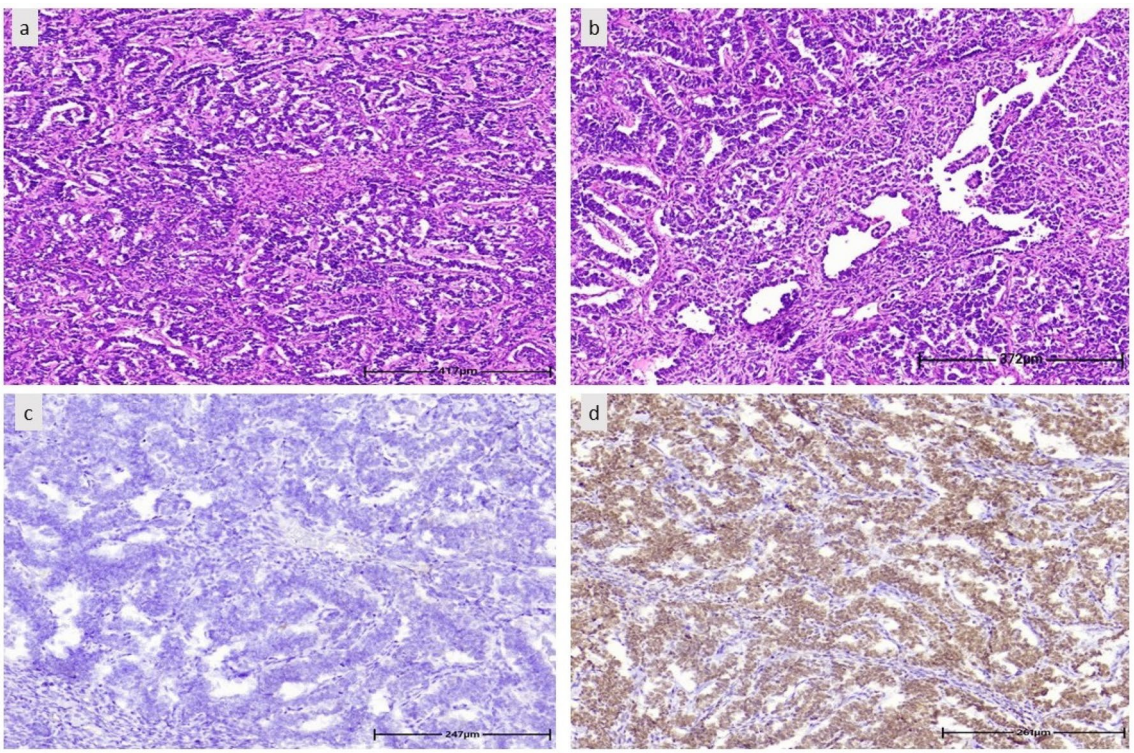

Fig. 2 **a**, **b** Epithelial and stromal elements are depicted, showing nests and cords of epithelial cells with intervening stromal spindle cells (hematoxylin and eosin staining). **c** Inhibin: negative staining observed (immunohistochemistry). **d** *WT1*: nuclear positivity in tumor cells (immunohistochemistry)

After completing the treatment, the patient fully recovered. End-of-treatment examinations revealed no abnormalities, and there were no adverse or unexpected events during the treatment and follow-up period. Her estimated glomerular filtration rate (eGFR) and renal function test results were within the reference range. Follow-up sonography revealed no abnormal findings, with standard kidney size, shape, and cortical echogenicity. There was no evidence of hydronephrosis, stones, or structural abnormalities. Monthly sonography follow-ups were conducted until 6 months after chemotherapy was completed. To date, she has shown no signs of recurrence.

Discussion

Wilms tumor, or nephroblastoma, is the most common renal tumor in childhood. In rare cases, it can originate from an extrarenal location, known as an extrarenal Wilms tumor (ERWT). ERWT can occur at any age but is more prevalent in children under 4 years old and is slightly more common in females [6, 9]. It can be found in various locations, including the retroperitoneum [10], inguinal canal [11], ovary [12], uterus [13], testis [14], pelvis [15], colon [11], bladder [16], and lumbosacral region [17].

Clinical presentation varies on the basis of the tumor's location and may range from an asymptomatic mass to symptoms such as abdominal pain, swelling, weight loss, and urogenital symptoms [10].

There are various hypotheses about the origins of ERWT, including ectopic metanephric blastema cells, Connheim's cell rest theory, and primitive mesodermal tissue [15]. The presence of the *WT1* gene in 25% of ERWT suggests that oncogenic mutations in nephrogenic rests play a role in the emergence of ERWT [9, 18].

Differential diagnoses for ERWT include primary intrarenal Wilms tumor with extrarenal metastasis, teratoma, neuroblastoma, pelvic rhabdomyosarcoma, and extragonadal germ cell tumors [14, 17]. ERWT arises outside the kidney, unlike intrarenal Wilms tumor, which presents as a palpable flank mass. Imaging studies such as CT or magnetic resonance imaging (MRI) are essential to confirm ERWT's extrarenal location and exclude renal origins or metastasis. Distinction from neuroblastoma is possible through elevated urinary catecholamines (homovanillic acid [HVA] and vanillylmandelic acid [VMA]), found in 90–95% of cases. Additionally, calcifications on imaging are rare in ERWT. In contrast to neuroblastoma, calcifications are present in a small percentage (about 5–10%) of Wilms tumors. Teratomas and extragonadal germ cell tumors may exhibit mixed solid-cystic components and elevated markers such as α-fetoprotein or β-human chorionic gonadotropin (β-hCG), which are typically not seen in ERWT. Pelvic rhabdomyosarcoma, a distinct soft tissue sarcoma, reveals unique imaging characteristics. A thorough approach combining history, examination, imaging, and targeted laboratory tests is essential for accurately differentiating ERWT from these tumors [19].

Imaging characteristics of ERWT and intrarenal Wilms tumors are generally heterogeneous and nonspecific. While imaging can suggest the presence of ERWT, a definitive diagnosis relies on pathological evaluation [20]. In this case, contrast-enhanced CT was selected owing to its quick availability, even though MRI is the preferred imaging method for Wilms tumor because of its superior soft tissue contrast and absence of ionizing radiation [4]. While MRI is especially helpful for identifying small bilateral tumors and nephroblastomatosis, as well as for monitoring after treatment, CT was prioritized in this instance for a timely initial evaluation.

A biopsy or surgical specimen is essential for confirming the diagnosis [21]. In addition to primary ERWT, nephroblastoma can also appear as metastases or teratoid Wilms tumors outside the kidneys. If ERWT is suspected, it is important to assess the kidney for the primary nephroblastoma tumor and determine whether teratoid elements are present in the specimen postoperatively [9, 22]. Histologically, ERWT is classified similarly to intrarenal Wilms tumors, typically showing a triphasic pattern with stromal, epithelial, and blastemal components [10, 11]. A teratoid Wilms tumor is characterized by containing a significant majority of teratoid elements, making up more than half of the tumor [22].

There was no specific treatment or staging for the extrarenal Wilms tumor. Instead, intrarenal staging and treatment were utilized. In most previous cases, including ours, the National Wilms Tumor Study (NWTS) staging was utilized. However, in some cases, the International Society of Pediatric Oncology (SIOP) and TNM staging were used [6]. According to NWTS guidelines, ERWT cannot be classified as stage I owing to its extrarenal location [9].

There is currently no standard treatment for patients with ERWT. The recommended course of action is complete tumor removal, followed by adjuvant therapy with a specific protocol. The chemotherapy regimen is determined on the basis of the tumor's histology and stage. It typically involves a combination of vincristine and actinomycin-D. Radiotherapy should be considered for patients with unresectable tumors, gross residual disease, or metastatic cancers [9, 10, 12]. The prognosis for ERWT is generally similar to that of intrarenal Wilms tumor [15]. Owing to the rarity of ERWT, large-scale studies on optimal treatment protocols are limited.

ERWT presents unique clinical decision-making challenges owing to its rarity and the lack of standardized management guidelines. Preoperative diagnosis remains difficult as ERWT shares imaging characteristics with other retroperitoneal tumors, such as neuroblastoma and germ cell tumors. This can lead to delays in appropriate management. Additionally, because ERWT is not as well studied as intrarenal Wilms tumor, clinicians often rely on existing Wilms tumor treatment protocols, which may not be optimal for all cases. There is a need for international registry-based studies to better define the prognosis, treatment responses, and long-term outcomes of ERWT. Such registries could help refine therapeutic strategies and determine whether ERWT requires a distinct staging and treatment approach.

Conclusion

Even though ERWT is rare and challenging to diagnose, it should be considered in the differential diagnosis of abdominal pain, particularly in young children, as it can be mistaken for other retroperitoneal tumors. Preoperative diagnosis remains challenging owing to its radiologic similarity to these tumors, requiring histopathological confirmation for a definitive diagnosis, which is typically made post-surgery. Treatment, staging, and prognosis for ERWT are similar to intrarenal Wilms tumor. However, owing to its rarity and the unique clinical challenges it presents, further studies could help refine treatment strategies and improve outcomes, as current evidence may not fully capture the specific characteristics of ERWT.

Abbreviations

β-hCG	Beta-human chorionic gonadotropin
CT	Computed tomography
eGFR	Estimated glomerular filtration rate
ERWT	Extrarenal Wilms tumor
HVA	Homovanillic acid
IHC	Immunohistochemical
IV	Intravenously
MRI	Magnetic resonance imaging
NWTS	National Wilms Tumor Study
PNET	Primitive neuroectodermal tumor
SIOP	International Society of Pediatric Oncology (Société Internationale d'Oncologie Pédiatrique)
VMA	Vanillylmandelic acid

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Author contributions

NA: patient management, follow-up, and supervision of the case report. AM: manuscript preparation and writing. EZ: radiological evaluation and writing of the radiology section. NM: pathological assessment and writing of the pathology section. All authors read and approved the final manuscript.

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Availability of data and materials

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Declarations

Ethics approval and consent to participate

Informed consent for participation was obtained from the patient parents.

Consent for publication

Written informed consent was obtained from the patient's legal guardian for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests

The authors declare that they have no competing interests.

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References

1. Itoshima R, Kobayashi R, Sano H, Hori D, Kishimoto K, Suzuki D, *et al*. Extrarenal nephroblastoma of the retroperitoneal space in children: a case report and review of the literature. J Pediatr Hematol Oncol. 2017;39(4):296–8.
2. Breslow N, Olshan A, Beckwith JB, Green DM. Epidemiology of Wilms tumor. Med Pediatr Oncol. 1993;21(3):172–81.
3. Davidoff AM. Wilms tumor. Adv Pediatr. 2012;59(1):247–67.
4. Dumba M, Jawad N, McHugh K. Neuroblastoma and nephroblastoma: a radiological review. Cancer Imaging. 2015;15:1–14.
5. Moyson F, Maurus-Desmarez R. Gompel C [Mediastinal Wilms' tumor?]. Acta Chir Belg. 1961;2:118–28.
6. Karim A, Shaikhyzada K, Abulkhanova N, Altyn A, Ibraimov B, Nurgaliyev D, *et al*. Pediatric extra-renal nephroblastoma (Wilms' tumor): a systematic case-based review. Cancers. 2023;15(9):2563.
7. Wabada S, Abubakar A, Adamu A, Kabir A, Gana L. A retroperitoneal extrarenal Wilms' tumour: a case report. Niger J Clin Pract. 2017;20(3):388–91.
8. Willis KR, Sathe AA, Xing C, Koduru P, Artunduaga M, Butler EB, *et al*. Extrarenal anaplastic Wilms tumor: a case report with genomic analysis and tumor models. J Pediatr Hematol Oncol. 2022;44(4):147–54.
9. Shojaeian R, Hiradfar M, Sharifabad PS, Zabolinejad N. Extrarenal Wilms' tumor: challenges in diagnosis, embryology, treatment and prognosis. Brisbane: Exon Publications; 2016. p. 77–93.
10. Park J. Extrarenal retroperitoneal Wilms' tumor with subsequent pulmonary and peritoneal metastasis in a 4 year-old girl: a case report and review of literature. J Pediatr Surg Case Rep. 2016;8:19–21.
11. Liang H, He Y, Fu L, Tian J, Sun N, Yu T, *et al*. Extrarenal Wilms tumor in children: a retrospective observational case series. J Pediatr Urol. 2020;16(5):664.
12. Albiroty KA, Al Sabahi A, Al Shabibi S, Al Ajmi ZI, Al Hinai K, Al-Mashaikhi N. Extrarenal Wilms' tumour of the ovary: a case report. Sultan Qaboos Univ Med J. 2022;22(4):566.
13. Leblebici C, Behzatoğlu K, Yıldız P, Koçyıldız Z, Bozkurt S. Extrarenal Wilms' tumor of the uterus with ovarian dermoid cyst. Eur J Obstet Gynecol Reprod Biol. 2009;144(1):94–5.
14. Morandi A, Fagnani A, Runza L, Farris G, Zanini A, Parolini F, *et al*. Extrarenal testicular Wilms' tumor in a 3-year-old child. Pediatr Surg Int. 2013;29:961–4.
15. Andrews PE, Kelalis PP, Haase GM. Extrarenal Wilms' tumor: results of the national Wilms' tumor study. J Pediatr Surg. 1992;27(9):1181–4.
16. Parkhi M, Peyam S, Peters NJ, Sodhi KS, Trehan A, Bal A. Primary Wilms tumor of the urinary bladder. Autops Case Rep. 2022;12:e2021390.
17. Armanda V, Čulić S, Pogorelić Z, Kuljiš D, Budimir D, Kuzmić-Prusac I. Rare localization of extrarenal nephroblastoma in 1-month-old female infant. J Pediatr Urol. 2012;8(4):e43–5.
18. Roberts D, Haber D, Sklar J, Crum C. Extrarenal Wilms' tumors. A study of their relationship with classical renal Wilms' tumor using expression

of WT1 as a molecular marker. Lab Investig J Tech Methods Pathol. 1993;68(5):528–36.
19. Golden CB, Feusner JH. Malignant abdominal masses in children: quick guide to evaluation and diagnosis. Pediatr Clin. 2002;49(6):1369–92.
20. Goel V, Verma AK, Batra V, Puri SK. 'Primary extrarenal Wilms' tumour': rare presentation of a common paediatric tumour. Case Rep. 2014;2014:bcr2013202172.
21. Rojas Y, Slater BJ, Braverman RM, Eldin KW, Thompson PA, Wesson DE, *et al*. Extrarenal Wilms tumor: a case report and review of the literature. J Pediatr Surg. 2013;48(6):e33–5.
22. Song JS, Kim IK, Kim YM, Khang SK, Kim KR, Lee Y. Extrarenal teratoid Wilms' tumor: two cases in unusual locations, one associated with elevated serum AFP. Pathol Int. 2010;60(1):35–41.

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