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Case Report

Adult-onset WT-1 negative Wilms tumor with predominant blastemal morphology: a rare case report

Priti Soin, Aqiba Bokhari, Kenneth A Iczkowski

Department of Pathology and Laboratory Medicine, University of California-Davis Health, Sacramento, CA, USA

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Abstract: Wilms tumor or nephroblastoma is the most common pediatric cancer of the kidney cancer. Its occurrence in adults is rare, accounting for <1% of all adult kidney malignancies. Histologically, Wilms tumor exhibits a triphasic morphology comprising of blastemal, epithelial, and stromal components. Morphologically, adult and pediatric Wilms tumors are similar and there are no histological or radiological differences between adult and a pediatric Wilms tumor. Wilms tumor-1 protein (WT-1) is the most important immunostain to help in the diagnosis of the entity and is positive in the blastemal component. This staining may be absent in the epithelial and stromal components. Rare WT-1 negative Wilms tumors exist, but most of them lack a blastemal component. Here we present a diagnostically challenging case of an adult- onset Wilms tumor presenting in a 19-year-old female that was blastemal-predominant and yet WT-1 negative, making it extremely rare. Additional Immunohistochemical stains revealed the tumor was positive for cytokeratin AE1/3 and paired box 8 (PAX-8) and negative for synaptophysin, chromogranin, CD45, and B-Rapidly Accelerated Fibrosarcoma (BRAF-1), thus ruling out other potential differential diagnoses.

Keywords: Wilms tumor, adult onset, WT-1 negative

Introduction

Wilms tumor or nephroblastoma, is the most common pediatric cancer of the kidney. It is usually seen in the first 5 years of life with a peak incidence between 3 to 4 years of age [1]. Wilms tumor in adults is rare (incidence of about 0.2 per million per year and represents less than 1% of all diagnosed renal tumors [2]. Adult Wilms tumor usually presents with abdominal pain and hematuria; whereas in children, the tumor is rapidly enlarging palpable abdominal mass that is frequently painless [3].

There are no significant histological or radiologic differences between adult and childhood Wilms tumor [1]. The classical triphasic histopathological features such as blastemal, epithelial and stromal elements are strikingly similar in both age groups [3].

The well-known criteria described by Kilton et al. are followed for diagnosing adult Wilms tumor. They include the following: 1. The tumor under consideration should be a primary renal neoplasm; 2. Presence of primitive blastemal spindle or round cell component; 3. Formation of abortive or embryonal tubules or glomerular structures; 4. No area of tumor diagnostic of renal cell carcinoma; 5. Pictorial confirmation of histology and 6. Patient's age should be >15 years [1]. Diagnosis of Wilms tumor includes basic comprehensive blood panel including renal function tests, urinalysis followed by imaging modalities which include renal ultrasonography, CT scan and abdominal MRI for localization of mass. Cytogenetic study for deletion of 1p and 16q may aid in diagnosis. Histopathologic confirmation is the gold standard where identification of primitive morphology (blastemal, epithelial and stromal) directs towards a sequence of immunostain, WT-1 being the most important positive marker. Several other immunostains are used which help to rule out other potential differentials. The clinical outcome of the adult tumors is usually poor compared to pediatric cases, more so due to lack of specific regimen or protocol for adult tumors. Adjusted pediatric protocols have been formulated by The Children's Oncology group and the International Society of Pediatric

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Table 1. Summarizes existing cases of adult type Wilms tumor with WT-1 negative expression

Case no.	Year	Country	Age/sex	Clinical presentation	Site/ laterality	Size (cm)	Predominance	WT-1 expression	Reference
1	2025	USA	19/female	Flank pain × 1 week	Left kidney	9.7	Blastemal	Negative	Current case
2	2025	Czechia	21/male	Abdominal pain and internal bleeding	Left kidney	8.5	Blastemal	Negative	4
3	2022	Austria	40/male	Persistent high blood pressure	Right kidney	25.5	Epithelial	Negative	5
4	2022	China	25/male	incidental	Left kidney	5.2	Epithelial	Negative	6
5	2020	Iran	39/female	Right flank pain	Right kidney	4	Epithelial	Negative	7

Oncology. The recommended guideline for therapy either includes surgical abscission or radical nephrectomy of the tumor followed by chemotherapy and radiotherapy 4 weeks later. Preoperative chemotherapy may be given in some cases to reduce the tumor size [1].

Wilms tumors are often positive for keratin, vimentin, desmin, actin, and Wilms Tumor-1 (WT-1), which distinguishes this type of tumor from other morphologic diagnostic mimics. WT-1 positivity is usually present in the blastemal component and may be completely absent in the epithelial and stromal parts. To date, only three negative WT-1 adult-type Wilms tumors have been reported in the literature, although one of them had non-specific expression of WT-1 (**Table 1**) [4-7].

We present a diagnostically challenging case of a young adult female with large kidney mass with characteristic morphology of Wilms tumor (blastemal and epithelial) which was WT-1 negative.

Case presentation

A 19-year-old female with no significant past medical history presenting with one week of left-sided flank pain was admitted for expedited malignancy workup following a CT finding of a large heterogeneous mass in the left kidney. A kidney biopsy was performed, followed by a radical nephrectomy after three days.

Gross findings

Sectioning revealed a 9.7 × 7.2 × 6.3 cm unifocal, tan, white to yellow-white, heterogeneous firm mass located in the mid to lower pole of the kidney. The mass extended into the hilum, into the renal vein and was present at the perinephric fat/vein margin (**Figure 1A**).

Microscopic findings

Biopsy revealed a primitive small round blue cell tumor with epithelial and blastemal components along with abundant mitotic figures (**Figure 1B**, **1C**). There were no features of anaplasia identified. Immunohistochemical stains revealed the tumor was positive for cytokeratin AE1/3 and paired box 8 (PAX-8) (image not shown), but negativity for WT-1 was the striking feature (**Figure 1D**). Additional stains showed tumor was negative for synaptophysin and chromogranin (Figure S1A, S1B), and there was patchy positivity for CD99 while INI-1 was retained (Figure S1C, S1D), CD45, and B-Rapidly Accelerated Fibrosarcoma (BRAF-1).

Radical nephrectomy was performed which showed similar morphology with 60% blastemal and 40% epithelial components with 20% necrosis. Invasion of the renal sinus, presence of lymphovascular invasion, and presence of tumor thrombus in renal vein (renal vein margin uninvolved) were identified. There were no features of anaplasia, and multiple lymph nodes which were sampled were negative for tumor.

WT-1 stain was repeated and was found to be consistently negative (**Figure 1E**). Tumor genetic evaluation was performed which revealed copy number gains of chromosomes 1q, 8, 10, 12 and 13q; and copy number loss of chromosomes 1p and 15q. No focal amplifications or deep deletions were identified.

Subsequently patient started abdominal radiation and systemic chemotherapy with vincristine, actinomycin and doxorubicin. Following the completion of the treatment regimen, her most recent evaluation revealed no progression of disease or evidence of metastatic disease.

Discussion

Wilms tumor, the most common pediatric malignancy, is an embryonal neoplasm that originates from nephrogenic blastemal cells and mimics the histology of the kidney with various patterns of differentiation. It is assumed to progress from persistent nephrogenic rests or

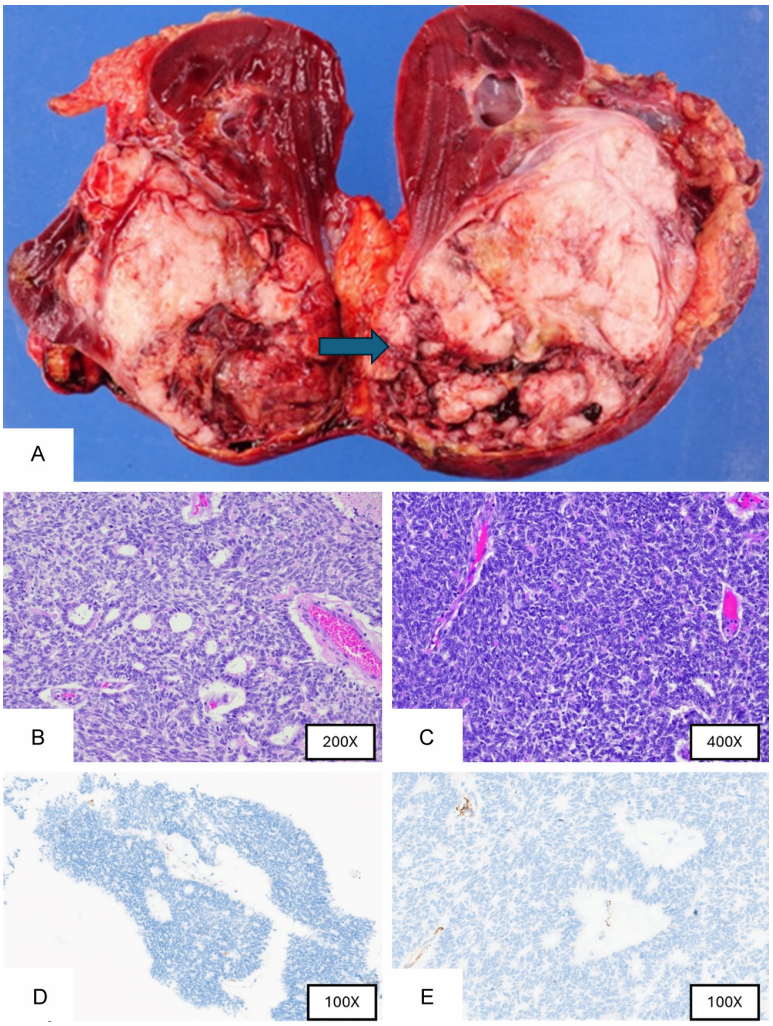

Figure 1. Gross and microscopic findings of specimens. (A) Gross nephrectomy shows a 9.7 unifocal, tan, white to yellow, heterogeneous firm mass in the mid to lower pole of the kidney (see blue arrow). (B) In the biopsy specimen, the epithelial component is noted in glandular and rosette forms, 200×, and (C) the undifferentiated or blastemal component is noted elsewhere, 400×. (D) Demonstration of WT-1 negativity in biopsy, 100×, and (E) in the resection specimen, 100×.

metanephric tissue. The tumor, showing no specific gender predisposition, has the average age of onset of 35 months for males and 45 months for females. Bilaterality is seen in about 5% of the cases. 10% of the tumors show mutation of WT-1 gene (11p13) along with other mutations which include 7p, 16q and 1p chromosomes.

Adult tumor is very rare. Adult Wilms tumor is diagnosed based on well-known criteria described by Kilton et al. [1]. The characteristic triphasic histology of Wilms tumor is composed of blastemal, epithelial and stromal elements in varying percentages. A blastemal predominant Wilms tumor is associated with poor outcome despite therapy and thus represents high risk tumors whereas the epithelial and stromal components present intermediate risk tumors. Tumors that are stromal-predominant are low-risk tumors. Anaplasia in Wilms tumor, which is seen in about 10% of total cases, is characterized by multipolar mitotic figures, enlarged nuclei at least three times the normal size, and hyperchromasia [1].

The use of immunohistochemistry is critical for definitive diagnosis. It mostly serves to distinguish distinct units with similar morphology and microscopic appearance. The main differential diagnosis to be considered with similar morphology includes metanephric adenoma (WT-1 variable and CD57 and BRAF positive), Papillary renal cell carcinoma (CK7 positive, WT-1 negative), Neuroblastoma (synaptophysin, chromogranin and CD-56 positive), Ewing's sarcoma (CD99 positive), rhabdoid tumor (INI-1 loss) and lymphoma (CD45 positive) [2, 3, 8, 9].

Multiple genes are involved in the pathogenesis of Wilms' tumors, including the *WT1* gene at 11p13, one or more genes at the Beckwith-Wiedemann syndrome locus at 11p15, at least two familial Wilms' tumor genes at 17q and 19q, plus other loci defined by allele loss and/or rare translocation breakpoints. *WT1* is mutated in ~10% of sporadic Wilms' tumors. Extensive allele loss studies have suggested that loss of heterozygosity (LOH) for 1p, 16q, and possibly 22q are adverse prognostic features in Wilms' tumors [10].

In our case the histopathology of the 9.7 cm tumor revealed a blastemal and epithelial dom-

inant tumor which was WT-1 negative thus imposing a diagnostic challenge. The case did not have any anaplasia, thus categorizing the tumor as favorable histology. Extensive workup was performed with the aim of ruling out all potential differentials which included lymphoma, neuroblastoma, metanephric adenoma, papillary renal cell carcinoma, Ewings sarcoma and rhabdoid tumor. With the characteristic morphology in the setting of WT-1 being negative, the diagnosis of WT-1 negative Wilms tumor was reached.

Conclusion

The case report has conveyed the diagnosis of adult-onset WT-1 negative Wilms tumor, which is a very rare entity. This highlights the importance of morphology in the appropriate setting even when one immunostain does not support the diagnosis. The immunostains should be judiciously interpreted, and other entities should be considered and ruled out with immunostains.

Disclosure of conflict of interest

None.

Address correspondence to: Dr. Kenneth A Iczkowski, Department of Pathology and Laboratory Medicine, University of California-Davis Health, 4400 V Street, Sacramento, CA 95817, USA. Tel: 916-734-7177; E-mail: kaiczkowski@health.ucdavis.edu

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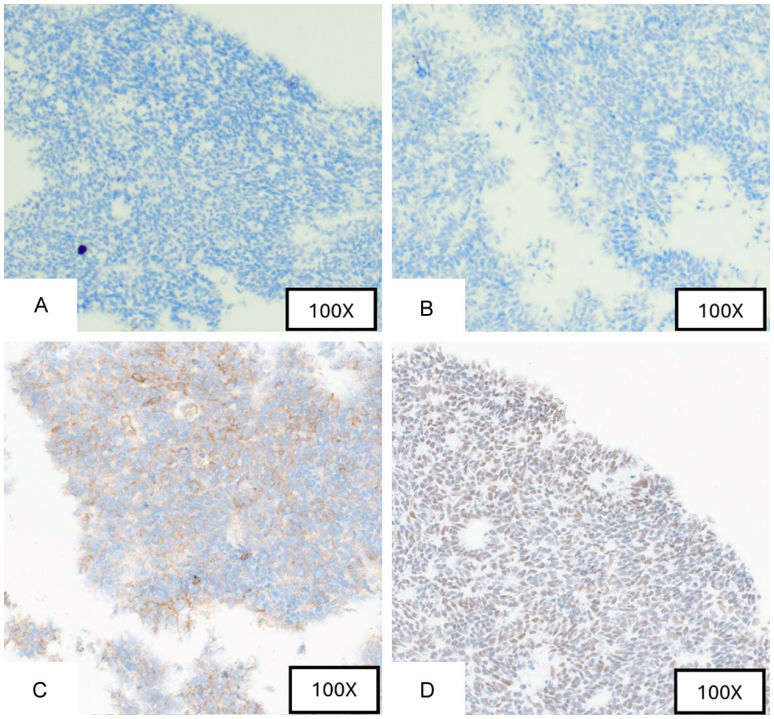

Figure S1. Additional immunostains in the biopsy. (A) Negative synaptophysin, 100× and (B) negative chromogranin, 100×; (C) Non-specific CD99, and (D) Retained INI-1.