Rhabdoid Tumor of Kidney Associated With Brain Mass; Case Report and Review of Literature

Mehrazma M[1], Hooman N[2], Saber Afsharian M[3]

Abstract

Background: Rahbdoid tumor of kidney (RTK) is a childhood rare neoplasm, previously thought as a sarcoma variant of Wilms' tumor, but now is recognized as a distinct pathologic entity.

Method: We report a pathologically proved case of RTK, associated with brain mass in a 4.5-month-old girl that has referred for gross hematuria.

Shortly after initiating chemotherapy, because of retractile vomiting, convulsion, and cerebral salt wasting syndrome.

Result: A tumor mass has found in mid-brain and pons in brain CT scan.

Conclusion: Because of association of RTK with same or other pathologic brain masses, CT or MR of brain is recommended for all pathologically proven rhabdoid tumors or for patients with neurologic symptoms.

Keywords: Rhabdoid tumor; Kidney; Brain

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1. Oncopathology Research Center and Ali-Asghar Children Hospital, Iran University of Medical Science, Tehran, Iran
2. Pediatric Department of Ali-Asghar Children Hospital, Iran University of Medical Science, Tehran, Iran
3. Pathology Department of Ali-Asghar Children Hospital, Iran University of Medical Science, Tehran, Iran

Corresponding Author:
Malihe Saberafsharian
Resident of Pathology,
Iran University of Medical Science
Tel: (+98) 21-88 27 90 43
Email:saberafsharian@yahoo.com

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Introduction

Renal tumors comprise 7-8% of all tumors under15 year's children [1]. Rahbdoid tumor of kidney (RTK) is a rare neoplasm of childhood once thought to be a sarcoma variant of Wilms' tumor [2]. It is now recognized as a distinct pathologic entity. RTK is a rare tumor, comprising about 2% of renal tumors of childhood [2-6]. The tumor has a distinctive predilection for infants, with a median age of 11months and over 80% of patients are under 2 years [7]. A highly distinctive feature of this tumor is its association with early brain metastases and primary intracranial masses such as primitive neuroectodermal tumor (PNET), ependymoma and cerebella and brainstem astrocytomas. Without an association with a brain neoplasm, RTK is a hardly distinguishable tumor from Wilms' tumor, and surgical excision and pathologic evaluation could make the definite diagnosis [8].

Because of rare incidence of RTK and its different aspects of clinical manifestations, we describe the clinical manifestations, lab tests and imaging of a pathologically proven case of RTK associated with brain mass in a 4.5-months-old girl.

Case Presentation

A 4.5- month- old girl has presented with gross hematuria, passage of clot, nausea, vomiting, and fever since three weeks ago. On physical examination she was pale, the abdomen was distended and a large firm mass with 15x 8 cm dimensions has felt on the left side of the abdomen. Wt = 6 Kg, HC= 40.5 cm, Lt=61 cm, BP:75/55 mmHg, PR:110 /min, RR:38/min, T:38.2C.

The results of lab tests were normal except for high level of LDH (4285 IU), ALT (87 IU) and marked anemia (Hct=24%).

Abdominal sonography revealed a heterogeneous lobulated intraparenchymal mass with low vascularity extended to the pelvic on the left kidney. Abdominal X-ray (Figure 1) has shown a large opacity in the left side of the abdomen displacement of bowel gas. Abdominal CT-scan (Figure 2) with contrast has revealed a heterogeneous lobulated intraparenchymal mass with low vascularity, detected on the left kidney, that its lower boarder extended to the pelvic.

On open surgery, a very huge fragile retroperitoneal mass has detected which has

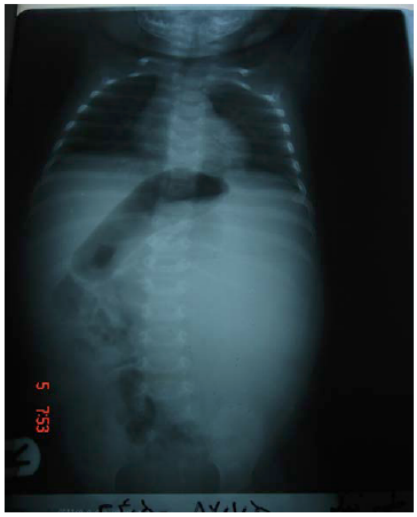

Figure 1. Large haziness in left side of abdomen with displaceme nt of bowel gases

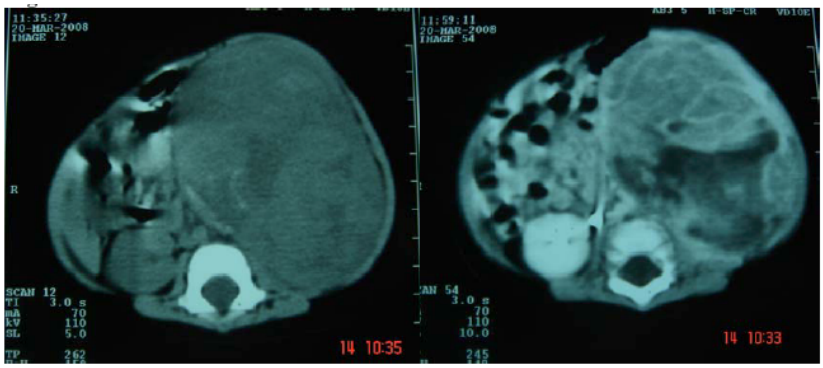

Figure 2. A heterogeneous lobulated intraparenchymal mass with low Vascularity on the left kidney. Its lower border extended to the pelvic

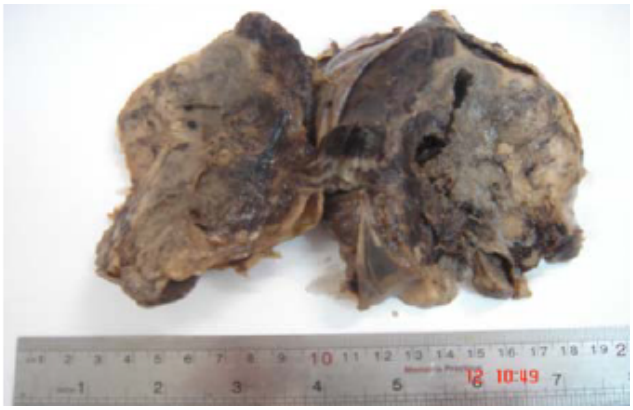

Figure 3. Gross appearance: a necrotic and Hemorrhagic mass that almost totally replaced the kidney

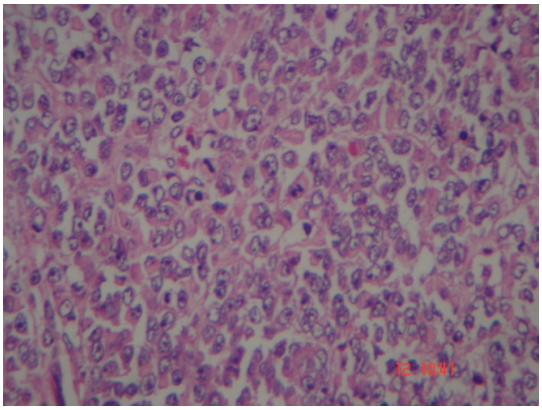

Figure 4. Neoplastic sheets of large tumor cells with rhabdoid features, vesicular nuclei, prominent nucleoli and eosinophilic cytoplasm. Few mitoses are also seen (H&E, X 200)

extended across the midline from diaphragm to pelvic with adhesion to liver and spleen. The patient underwent left radical nephrectomy.

On gross examination the left kidney has extensively replaced by a necrotic and hemorrhagic tumor. The dimensions were 12x11x7cm, and the weight was 380gr. (Fig. 3).

Microscopic examination revealed neoplastic tissue characterized by sheets of large cell with rhabdoid features, vesicular nuclei, prominent nucleoli and eosinophilic cytoplasm. Some of them contained cytoplasm inclusions. Also high mitotic activity and area of necrosis have noted (Fig. 4).

Immunohistochemical staining of tumor cells have shown that the tumor cells were only positive for vimentin. Tumor cells were negative for epithelial membrane antigen, neuron specific enolase, cytokeratin, myogenin, and desmin.

Standard protocol of chemotherapy with dactinomycin and vincristin; has started immediately after operation. Then after 4-5 days, she has experienced several episodes of mild to moderate dehydration while the urine output was 2.5 cc/kg/h and appropriate fluid therapy has prescribed.

In the 10th day after operation, she has developed lateral gaze, upper extremities twitching, mottling, tachypnea (60/min), tachycardia (188/min), and

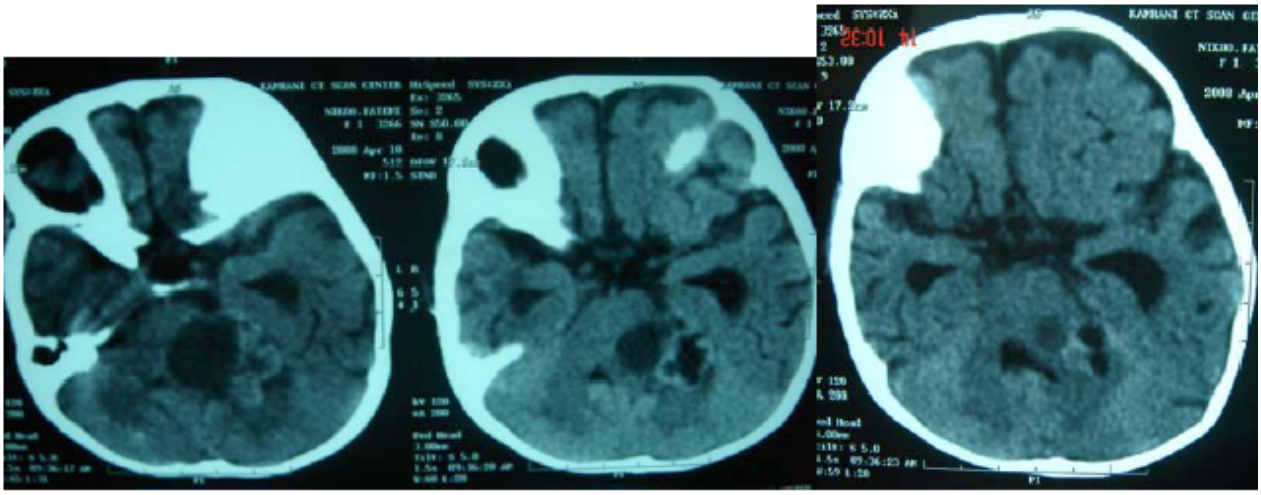

Figure 5. Brain CT scan: a large tumor mass in pons and left CP-angle

diffuse rales on both lungs. Blood pressure was *75/55* mmHg.

The lab test results has shown severe hyponatremia (Serum Na=110meq/L), diluted urine (SG= 1.008), and natriuresis (Urine Na= 120 meq/l), that has associated with high urine output (14ml/kg/h) suggesting cerebral salt wasting syndrome.

A brain CT scan obtained the revealed mass of midbrain and cerebellum. (Figure 5). It has large cystic component with some solid parts. It obliterates the 4th ventricle and caused obstructive hydrocephaly.

Discussion

According to all clinical, laboratory and histological findings; the diagnosis is Rhabdoid tumor, that immunohistochemical staining has confirmed it. Kidney Rhabdoid tumor is the most aggressive malignant kidney neoplasm in the children [1-3].

It is a very rare tumor, only 2.5% among all pediatric renal tumors, but too dangerous; 80% of RTK patients would die within the first year [9].

The tumor has initially identified in 1978 by the the national wilms' tumor study pathologists [1-3].

Histologically, it bears a superficial resemblance to rhabdomyosarcoma with abundant eosinophilic cytoplasm containing filamentous inclusions, which consists of tangles of vimentin filaments but not for actin or myosin that are present in tumors of myogenic origin [4].

Because of this resemblance to myogenic tumors, pathologists has named it "rhabdomyosarcomatoid tumor" and this has later shortened to "rhabdoid tumor" [3]. It occurs exclusively in children, affecting males more than females (1.5:1), approximately has diagnosed within 18 months age [8].

RTK most commonly presents as a palpable abdominal mass. Hematuria, fever, and hypertension are other common presenting symptoms [1].

Early brain metastasis is a highly distinctive feature as well as association with primitive neuroepitheilal tumors of the posterior fossa and middle cranial fossa. These tumors include PNETs, ependymoma, and cerebella and brainstem astrocytomas. Neurologic symptoms might occur as squeal of these intracranial masses [10-13].

RTK is similar in gross pathologic appearance to Wilms' tumor and, therefore, usually hardly distinguishable by imaging.

Like wilms', it involves intrarenal and therefore renal hilum, and there might be invasion of the renal vein and inferior vena cava. RTK does not typically encase or envelope aorta, that is a feature of the neuroblastoma[2,9].

In ultrasound, RTK appears as a large lobulated mass with heterogeneous echogenicity due to hemorrhage, fat, necrosis, or calcification [3].

Some authors have reported RTK CT scan, in comparison to Wilms' tumor, more likely to demonstrate a central location within the kidney, and also with presence of a peripheral crescent-shaped sub capsular fluid collection, tumor lobules outlined by calcifications and low-density separated areas hemorrhage and necrosis [10].

Also this tumor might involve extra renal sites, particularly the central nervous system.

Unfortunately, among 15% of these patients CNS lesions develop as resembling medulloblastomas or primitive neuroectodermal tumors;

The vast majority of these tumors upon careful genetic analysis are classified as atypical teratoid/rhabdoid tumors.

It has been suggested that most patients with both renal and central nervous system rhabdoid tumors have germ line mutations involving one copy of the hSNF5/INI1 gene. RTK in infants and children has a dismal prognosis, independent from localization. The presence of metastasis at diagnosis seems to be the only prognostic factor of outcome.

Favored metastatic sites include bone, brain, and regional lymph nodes. It is important to remember that a wide range of neoplasm's (including nephroblastoma, renal cell carcinoma, and mesoblastic nephroma) could be associated with rhabdoid cytology. These need to be considered prior to making a diagnosis of rhabdoid tumor.

Immunohistochemistry has shown the cytoplasmic inclusions to stain positively for both vimentin and cytokeratin. Recently an immunohistochemical stain for the INI-1 protein product has been shown to be diagnostically useful [9, 12, 13].

As we present the case, CT scan or MR imaging of brain is recommended for all pathologically proven rhabdoid tumors or for patients with neurologic symptoms prior to resection. However, routine imaging of the brain looking for brain metastases in the absence of neurologic symptoms prior to surgery is not recommended, because most renal tumors will prove to be Wilms' on histological evaluation [4].

As a rule, this case underwent abdominal CT scan and assumed a Wilms' tumor, therefore underwent left kidney excision. The case had not any CNS symptom before surgery, and after that, the first differential diagnosis of dehydration and other symptoms assumed electrolyte imbalance.

After proving of RTK by pathologist and with appearance of extra pyramidal symptoms, brain CT scan has shown a midbrain-cerebella mass.

The differential diagnosis of solid renal tumors in childhood is as follows: Wilms' tumor (most common), Clear cell sarcoma, Rhabdoid tumor, Lymphoma, Renal medullary carcinoma, Renal cell carcinoma, Neuroblastoma encasement.

In summary in a child with abdominal mass and associated CNS symptoms, RTK is the top list of differential diagnoses.

After proving RTK by pathologic examination, a brain imaging should be done, because of 15 percent association of RTK with same or other pathologic brain masses.

Acknowledgments

None

Conflict of Interest

The authors declare that they have no conflict of interest in this article.

Authors' Contribution

All of the authors contributed in all parts of the article.

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