

Rhabdoid Tumor of the Kidney: A New Case of Prenatal Diagnosis with Metastases Immediately

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Abstract

Rhabdoid tumor of the kidney is a rare pediatric malignancy arising most of the time before the age of two years. The diagnosis is generally difficult. The treatment is not well codified and the forecast remains very dark in spite of the progress of the pediatric oncology. We report a new case of rhabdoid renal tumor diagnosed prenatally and we specify our diagnostic and therapeutic approach.

Keywords: Rhabdoid tumor; Kidney; Newborn; Prenatal Diagnosis; Metastases

Introduction

Rhabdoid tumor of the kidney is a rare neoplasm of enfant [1]. It is now recognized as a distinct pathologic entity typically arises in infants with a median reported age of 22.5 months [2]. These tumors are aggressive and invasive [3].

Case Report

A newborn child, 4 days old, was admitted in our service for a left renal tumor with prenatal diagnosis. Examination on admission to the hospital revealed a preserved general state with a left upper abdominal mass and a purplish cutaneous nodule beside the scapula. The biological check up was normal. The abdominal ultra sound showed superior polar mass of the left kidney measuring 5 cm with pelvic dilation. The abdominal CT ended in the presence of a superior polar mass of the left renal measuring 5.7×5.2×6 cm with pyelic dilation, associated with multiple hepatic metastases (Figures 1 and 2). The osseous scintigraphy showed the presence of hyperfixation zones towards osseous metastases.



Figure 1: Scannographic section showing the large left renal tumor.

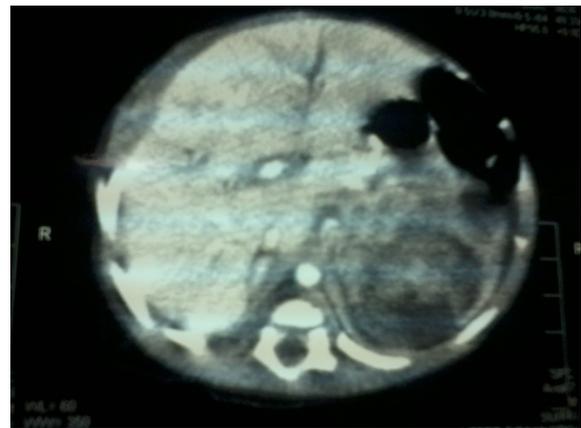


Figure 2: Large renal mass containing lobules separated by areas of hemorrhage.

The child was operated at the age of 20 days. We had found a voluminous retro peritoneal mass in the upper left kidney adhering to the left suprarenal gland and a lymph node in the renal hilum (Figure 3).



Figure 3: Per operative picture of the kidney replaced by rhabdoid tumor. The tumor contains areas of necrosis and hemorrhage.

We also noted the presence of a nodule at the level of the hepatic dome measuring 0.5 cm. The child had a left nephrectomy,

lymphadenectomy and a resection of the cutaneous nodule. The pathological examination found a widely necrosis rhabdoid tumor (Figure 4), infiltrating the residual parenchyma and the adrenal gland with an exceeded renal capsule and ganglion and skin metastases (Figure 5).

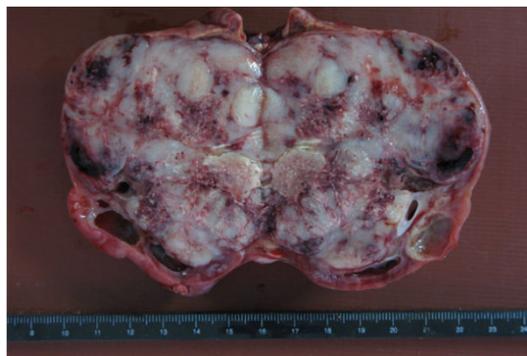


Figure 4: Macroscopically, the tumor is mole, pale, uniform, well-defined renal parenchyma but not encapsulated.

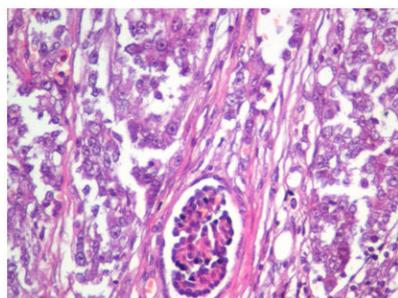


Figure 5: Renal parenchyma infiltrated by a monomorphic tumor cell proliferation made large, non-contiguous, with vesicular nuclei and nucleoli strongly.

The child was then transferred to the pediatric oncologic service, a chemotherapy protocol was started. He received 2 courses of vincristine half doses. After the first cure, he developed new cutaneous metastases of the scalp and died ten days after.

Discussion

Rhabdoid tumor of the kidney is the most aggressive malignant neoplasm of the kidney in children. It represents 1.5% of all the pediatric cancers of the kidney [4], and 8% of those arising before seven months [5]. The tumor was first identified in 1978 by the pathologists of the National Wilms' Tumor Study [3]. The tumor was given the name "rhabdoid" because microscopically it resembles to a rhabdomyosarcoma although it does not show skeletal muscle markers by electron microscopy, immunoperoxidase, or cytogenetic studies [6]. The tumor occurs in the perinatal period, during the first year of life and occasionally in older individuals. It affects males more than females (1.5:1), with the mean age at diagnosis approximately 18 months [7]. In addition to the kidney, the malignancy is found in the soft tissues, skin, CNS, and other extrarenal sites [8]. When RT occurs in utero, it is more likely to present at birth with multiple metastases

and a rapidly progressive, downhill clinical course ending in early death [8]. Rupture of the tumor in utero causing a severe fetal anemia has been described [9].

The prenatal diagnosis of renal rhabdoid tumor is rare. Only two cases were reported in the literature between 1970 and 2009 published in a systematic review by Hart Isaacs Jr [10].

Presentation of this tumor may vary from abdominal mass and haematuria in the new born to metastases that mainly affect regional lymph nodes, lungs, peritoneum, liver and central nervous system. Some patient presented with hypercalcemia, secondary to elevated parathormone levels, has also been reported [11,12].

These tumors include PNETs, ependymoma, and cerebellar, and brain stem astrocytomas Neurologic symptoms can occur as sequelae of these intracranial masses [13]. For our patient, the clinical examination showed the presence of a left abdominal mass. Metastases were subcutaneous but also in the liver revealed by radiological investigations.

Biological tests may reveal nonspecific abnormalities such as anemia or hypercalcemia (4-18% of cases).

No pathognomonic imaging feature can be used to diagnosed or differniate malignant rhabdoid tumor from the other renal tumors of childhood. In the abdominal ultrasonography, tumoral invasion of the renal vein and/or the inferior vena cava is sometimes seen with malignant rhabdoid tumor and is best diagnosed with Doppler ultrasonography or magnetic resonance angiography

In the abdominal CT, malignant rhabdoid tumor typically appears as a large, lobulated mass in the center or periphery of the kidney.

Others radiological investigations are indicated to search metastases. The chest CT is indicated because lungs remain the most common site of metastatic disease from non-CNS malignant rhabdoid tumor. The brain CT is indicated to exclude the possibility of a synchronous primary or metastatic brain tumor.

Macroscopically, the tumor is typically large, haemorrhagicand necrotic, with ill defined borders that reflect its highly invasive nature [14].

Histologically, this tumor is unencapsulated, and feature sheets of tumor cells that aggressively overrun native nephrons. Vascular invasion is usually extensive. Tumour cells characteristically display the cytologic triad of vesicular chromatin, prominent cherry-red nucleoli and hyaline pink cytoplasmic inclusions. A subset of tumors may be composed predominantly of primitive undifferentiated small round cells, but on closer inspection small foci of cells with diagnostic cytologic features can be identified [15].

Histologic diagnosis is supported by lack of immunohistochemical staining for SMARCB1/INI1, indicating loss of protein expression [16] typically due to exonic deletions or mutations resulting in biallelic inactivation of SMARCB1 (hSNF5/INI1) tumor suppressor gene at chromosomal locus 22q11.23 [2].

Treatment planning by a multidisciplinary team of cancer specialists (pediatric surgeon or pediatric urologist, pediatric radiation oncologist, and pediatric oncologist) with experience treating renal tumors is required to determine and implement optimum treatment.

No treatment has proved its effectiveness for these children. The National Wilms Tumor Study (NWTs) recommended combinations of etoposide and cisplatin; etoposide and ifosfamide; or ifosfamide,

carboplatin, and etoposide [17]. The International society of Pediatric Oncology (SIOP) has showed that preoperative chemotherapy does not improved prognosis. Delays in surgical treatment increase mortality [5].

Complications related to treatment are myelosuppression, tubular disorders with loss of protein, phosphorus, bicarbonate, electrolytes, a schwartz-Barter, heart problems and neurological disorders.

The prognosis is very dark with 80-90% of mortality despite intensive treatment. Overall survival in trials NWTs-1 to NWTs-4 is 23.6%. The prognostic factors were stage III-IV and age 1 year. This tumor is the most deadly in small infants, 13% of these children develop a brain tumor from a different histological type: medulloblastoma, primitive neuroectodermal tumor or glioma [18].

Conclusion

The rhabdoid tumor of the kidney is a rare and very aggressive tumor that mainly affects infants. It is characterized by rapid growth and early metastasis. The diagnostic confirmation is based on histology and immunostaining.

Its clinical features were dominated by abdominal mass and hematuria. Its prognosis is very poor despite advances in pediatric oncology. The 2-year survival after diagnosis rarely exceeds 15%.

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