ISSN No:-2456-2165

Rhabdoid Tumor of the Kidney in Children: A Rare Case Report

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Abstract:-
Introduction: Renal rhabdoid tumor is a rare and extremely aggressive tumor that results in multiple and early metastases. Through our case, we recall its diagnostic peculiarities and its therapeutic modalities.
Case presentation: We report the case of a 5-year-old girl with a rhabdoid tumor in the left kidney with multiple retroperitoneal lymphadenopathies.
Discussion: Kidney rhabdoid tumor was originally described as a "Rhabdomyosarcomatoid" variant of Wilms tumor due to the resemblance of cells to rhabdomyoblasts. Currently, this type of tumor is recognized as a distinct and unique malignant renal tumor and it usually affects children under 2 years of age. Sampling of tumor tissue is mandatory for diagnosis of Malignant rhabdoid tumor of the kidney (MRTK), either based on nephrectomy, baseline biopsy, or autopsy samples. There is no established therapeutic standard due to the rarity of cases. However, so far, surgery is considered to be the first choice of treatment whenever possible.
Conclusion: In order to be able to discuss possible treatment options, it is important to recognize this tumor entity and its specificities.

Keywords:- *Chromosome 22, GFA NEPHRO 2005, Radiotherapy, Rhabdoide.*

I. INTRODUCTION

Rhabdoid kidney tumor in children is a rare but very aggressive tumor. It represents 2 to 3% of pediatric renal tumors. Rhabdoid tumors were initially described in the kidney as variants of Wilms tumors, characterized by young age of onset, unusual aggressiveness, and a clearly distinct morphology from nephroblastoma [1]. We report the case of a rhabdoid kidney tumor in a 5-year-old girl.

Case presentation

We report the case of a 5-and-a-half-year-old child of a non-consanguineous marriage with no particular personal or family history who presented with abdominal pain and recent deterioration in general condition that had progressed over a month.

On clinical examination, there was an abdominal mass at the left hypochondrium and the epigastrium of firm consistency, fixed and painful, edge of the rib cage was not palpable. The umbilical perimeter measured at 57cm; the rest was unremarkable.

The thoraco-abdomino-pelvic scanner showed the presence of 2 large masses, one at the upper pole and the other at the lower pole of the left kidney, measuring 170×75×144mmand 60×54×70mm respectively, with irregular heterogeneous density, not containing calcifications and enhancing heterogeneously after contrast, delimiting large areas of necrosis (Figure1). The test has also shown the presence of retroperitoneal lymphadenopathy as well as 2 pulmonary parenchymal micronodules of suspicious appearance.

ISSN No:-2456-2165

The child was put on preoperative chemotherapy according to the GFA NEPHRO protocol. She had then undergone total nephrectomy. The surgical exploration had revealed a large mass at the left kidney occupying almost the entire abdominal cavity and pushing back the intestine, the liver, the spleen and extending towards the thorax through the esophageal hiatus. This led to a pyelo-ureteral junction syndrome.

The anatomopathological study had concluded the existence of a rhabdoid tumor. The sinuses were infiltrated, areas of necrosis were estimated at 70% of the total tumor volume, the limit of ureteral resection was healthy. The tumor was classified: ypT3aNx (AJCC 2009). Immunohistochemistry demonstrated focal expression of cytokeratin and vimentin by tumor cel20 days after surgery, the child received three-dimensional conformational radiotherapy at a total dose of 21Gy with 14 fractions at a dose of 1.5Gy per fraction. Post-operative chemotherapy was delivered according to the protocol GFA NEPHRO HR 2005. after 5 years regular follow-up care, there was a clear regression of the tumor (Figure2).

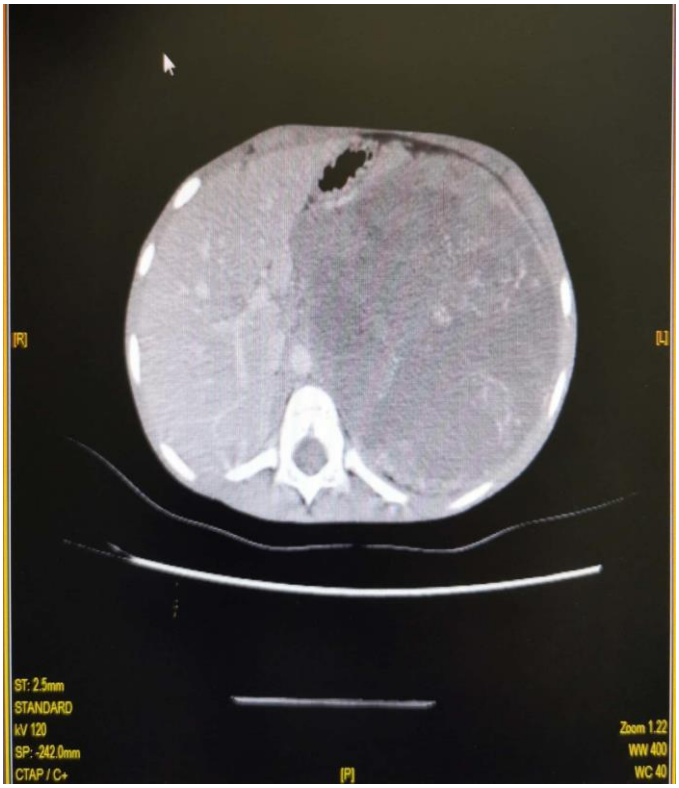

Figure1: CT scan shows the presence of a large masse on the left kidney

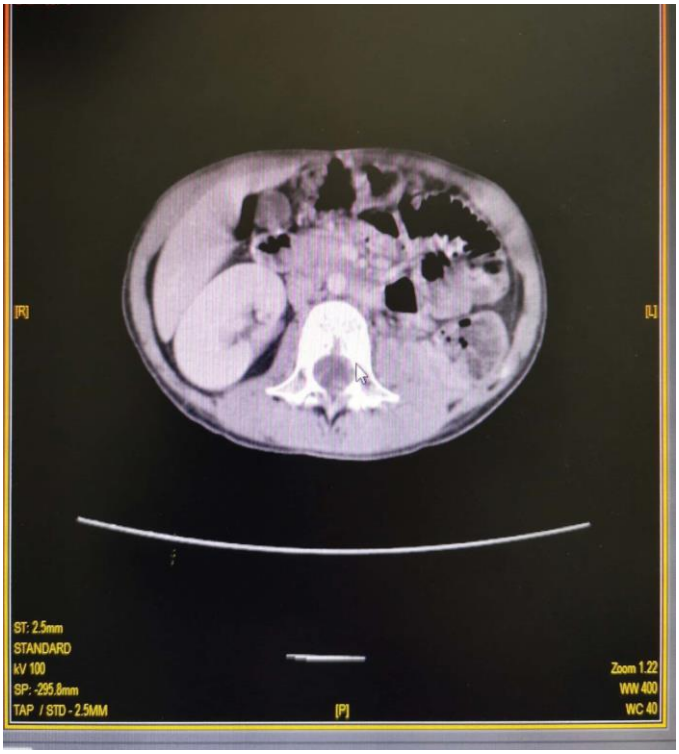

Figure 2: CT scan shows a clear regression of the tumor

II. DISCUSSION

Renal rhabdoid tumor is a rare and extremely aggressive entity. It represents 2 to 3% of kidney cancer in children. It was initially described in 1978 by Beckwith as a kidney tumor distinct from nephroblastoma, of high grade of malignancy [2]. The term rhabdoid was assigned to it because the tumor cells looked like rhabdomyoblast when examined under the microscope.

This tumor can occur in patients from one-day-old to 18 years old, the average age is 17 months with majority of cases under one year old [3]. However, the neonatal form is rare. It can be associated with a brain tumor in 10% to 15% of cases and it is often a primary neuroectodermal tumor or a medulloblastoma [4]. The symptoms are not specific for renal rhabdoid tumor and is dominated by abdominal mass (82% of cases) and hematuria (80% of cases). The later is only present in 24.4% of cases of nephroblastoma.

In more than 70% of cases, this tumor is discovered at a metastatic stage. Metastases mainly concern the locoregional lymph nodes, the peritoneal cavity, the liver, the central nervous system [5] and the lungs. Per our case, lungs metastases were present. Bilateral involvement is exceptional. Skin metastases are also exceptional and are mainly associated with neonatal forms [6]. Laboratory results may reveal non-specific abnormalities such as anemia or hypercalcemia (4 to 18% of cases) [5]. This hypercalcemia is not pathognomonic of renal rhabdoid tumor, but it can be a biological marker of tumor activity. Indeed, in the absence of any associated metastatic bone lesion, the secretion of parathyroid hormone by tumor cells would be the most plausible humoral mechanism. Likewise, the increase in prostaglandin E2 could explain, in some cases, these disorders. Furthermore, obtaining

ISSN No:-2456-2165

pathognomonic diagnostic elements via radiology is not conclusive for this tumor.

However, the lobular appearance of a renal mass, the presence of a subcapsular collection and large areas of linear calcifications would be characteristic signs of the renal rhabdoid tumor on the tomodensitometric examination which also makes it possible to establish a balance sheet for loco-regional extension [7]. The CT results in our case show 2 large masses of the left kidney as well as retro peritoneal lymphadenopathy.

On gross examination, the renal rhabdoid tumor is indistinguishable from nephroblastoma. The histological architecture typically consists of dense patches of large rhabdoid, ovoid or polygonal cells with acidophilic cytoplasm. The nucleus is eccentric and vesicular with a large owl's eye nucleolus. The discovery of specific chromosomal abnormalities on the long arm of chromosome 22 (22q11.2) is characteristic of rhabdoid tumor [8].

Histologically, the renal parenchyma in our case was the site of a rhabdoid-like proliferation which was arranged in diffuse sheets, with the presence of large non-cohesive cells with eccentric nuclei, the cytoplasm contained a large eosinophilic inclusion with large tumor cells. The cells were of big size and multinucleated with the presence of tumor necrosis represented 70% of the total tumor volume.

The differential diagnosis of renal rhabdoid tumor arises with congenital mesoblastic nephroma and metanephric stromal tumor in the neonatal period, and with nephroblastoma and clear cell sarcoma in infants. The therapeutic protocol for renal rhabdoid tumor is disputable. Indeed, for the National Wilms Tumor Study (NWTS) [9], renal rhabdoid tumors are treated by an enlarged nephrectomy, followed by chemotherapy (carboplatin, cisplatin and cyclophosphamide) for 24 weeks and by postoperative radiotherapy. However, for International Society of Pediatric Oncology (SIOP), treatment consists of first chemotherapy followed by surgery and postoperative chemotherapy (etopside, carboplatin, ifosfamide and epirubicin) for 34 weeks, and then postoperative radiotherapy [6]. Our patient received preoperative chemotherapy followed by nephrectomy and postoperative radiotherapy, and finally adjuvant chemotherapy.

Despite advances in pediatric oncology, the prognosis of patients with renal rhabdoid tumor remains particularly poor with 5.5 months relative survival rate in 80% to 90% of cases [5]. However, being female and the absence of lymph node metastases would be elements of a good prognosis [7]. In our case, after 5 years regular follow-up care, there was a clear regression of the tumor.

III. CONCLUSION

Renal rhabdoid tumor is a rare, extremely aggressive tumor that results in multiple and early metastases. Its clinical symptoms are predominately characterized by abdominal mass and hematuria. Certain elements may point to the diagnosis of renal rhabdoid tumor, namely the existence of a brain tumor apart from a metastasis, damage to chromosome 22, a lobular aspect of the mass, a subcapsular collection or large areas linear calcifications on CT examination. Its prognosis remains bleak despite advances in pediatric oncology. The 2-year survival after diagnosis hardly exceeds 15%. Therefore, our case demonstrates a rare exception with clear regression of tumor 5 years post treatment.

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