

## Malignant Renal Tumor Associated with Genetic Disorders

C Caridad Verdecia Cañizares<sup>1\*</sup>, Magda Alonso Pérez<sup>2</sup>, Alina GarcíaGarcía<sup>3</sup>, Yeidi Reyes Montero<sup>4</sup> and Odelay Gómez Marrero<sup>5</sup>

<sup>1</sup>Second Degree Specialist in Oncology and Pediatric Surgery, Doctor in Medical Sciences, Professor and Senior Researcher, Head of Onco-surgery Service, William Soler Pediatric Hospital, Havana, Cuba

<sup>2</sup>First-degree Specialist in MGI and Pediatrics, Master in Integral Attention to the Child, Oncopediatrics Training, William Soler Pediatric Hospital, Havana, Cuba

<sup>3</sup>First Degree Specialist in MGI and Genetics, William Soler Pediatric Hospital, Havana, Cuba

<sup>4</sup>First-degree Specialist in Pediatrics, William Soler Pediatric Hospital, Havana, Cuba

<sup>5</sup>First-degree Specialist in MGI and Pediatric Surgery, Hospital William Soler, Havana, Cuba

**\*Corresponding Author:** C Caridad Verdecia Cañizares, Second Degree Specialist in Oncology and Pediatric Surgery, Doctor in Medical Sciences, Professor and Senior Researcher, Head of Onco-surgery Service, William Soler Pediatric Hospital, Havana, Cuba.

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### Abstract

Rhabdomyosarcoma is a malignant embryonic tumor, located in soft tissues such as perineal, retroperitoneal region, the botryoid variant tends to be located in the gallbladder, bladder, middle ear, very rare in kidney. It can cause abdominal pain, nausea, vomiting and fever, palpable tumor mass, hematuria and arterial hypertension. Our goal is to raise awareness through a clinical case, the kidney rhabdomyosarcoma and in a skin biopsy performed before the nephrectomy, metaphases are detected from mosaic aneuploidy, by genetic studies. We described a child with rapidly progressive disease kidney. This case report aims to raise awareness of this rare disease so that individuals with Rhabdomyosarcoma kidney are identified and provided with appropriate care.

**Keywords:** Infrequent Renal Malignant Tumor; Genetic Anomaly

### Background

The pediatric kidney is a common site for tumors carrying specific chromosomal alterations. The most common of these is the nephroblastoma or Wilms tumor, which is associated with anomalies in two loci: 11p13 and 11p15, the latter also linked to Beckwith-Wiedemann syndrome. Two other genes that seem to be implicated are WT3 and WT4. In addition, 1q gains or 22 deletions have been shown to independently be associated with a worst prognosis in WTs [1]. Other neoplasms with chromosomal rearrangements, such as Renal Cell carcinomas are much less frequent in children (between 1.8 and 6.3% of all malignant renal tumors). Among these, the "translocation renal carcinomas" have been identified involving the locus Xp11 with two main types of translocations: t(X; 1), and t(X; 17) [1,2]. Congenital mesoblastic nephroma is a renal tumor affecting newborns and young infants, a specific translocation t(12;15) (p13;q25), which is also present in congenital fibrosarcomas outside of the kidney. These findings have led to conclude that these two tumors are genetically equivalent [1,3,4]. Rhabdoid tumors of the kidney are very rare and aggressive neoplasms that appear with a mean age of 11 months. At least 50% of these carry abnormalities in the 22q11.2. This gene is probably involved in transcriptional modulation of other genes such as the oncogene c-Myc [1].

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### Case Presentation

- Patient: Franco Bismark González Cots HC: 711030 CI: 12041364401.
- Age: 6 years, Sex: M, Race: B, Proceed: Cattlemen.
- APP: Light to moderate mental retardation.
- Delay growth and development.
- Language disorders.
- Chromosomopathy?
- APF: parent carrier of chromosomal involvement.
- HEA: a 6-year-old schoolchild, who was born 28 days after birth (4/5/2013), is diagnosed with perineal hemangiopericytoma and is surgically resected.

At 11 months of age (4/9/2014) the lesion in the surgical scar reappears, (See figure 1) it is re-intervened and the pathological anatomy revealed embryonic rhabdomyosarcoma of the perineum, with immunohistochemistry: positive vimentin, positive desmin, HHF35 positive, Ki67 positive, 50%, S 100 negative, LCA negative.



**Figure 1:** Perineal tumor lesion at 11 months of age, rhabdomyosarcoma.

A multidisciplinary study was carried out, staging studies were completed with no tumor lesion in another part of the organism, tumor markers were performed, with 555 U/L high dehydrogenase, alpha protein and negative chorionic gonadotrophin, normal hemoglobin and erythro sedimentation levels, and chemotherapy was initiated. postoperative with VAC scheme: vincristine, cyclophosphamide, and actinomycin D, whose treatment concluded on August 06, 2014 with very good response.

Followed by monthly follow-up oncological consultation, here the physical examination was detected on March 20, 2014 an accelerated growth at the level of perianal scar, high LDH figures that had been negativized, now at 550 UI, in ultrasound of the region described injury hypoechoic 25 mm at the edge of the surgical scar, it is surgically intervened (Figure 2) the result of pathological anatomy was reactive chronic inflammatory process, no residual tumor at the edges of section, Ki 67 negative, HHf 35 negative, positive LCA, M10 D negative.



**Figure 2:** Thickened surgical scar.

Follow-up is continued for 5 years, also by consultation of physiatrist and speech therapy, with the patient being able to improve language and walking markedly, entering a special circle achieving acceptable learning, and also being evaluated by clinical genetics from birth.

In August 2018 he began with abdominal pain, vomiting, and fever 38°C. He was evaluated twice in the gastroenterology clinic, discarded parasitism, and sent to our clinic for a palpable tumor mass in the left hypochondrium.

Abdominal ultrasound: left renal tumor that occupies upper two thirds of the organ, irregular surface, vessels of neoformation, extends to the midline of the abdomen, 11 long x 8.7 cm wide. Shifting intestinal loops rest the normal intrabdominal structures, not ganglia or ascites.

Rx thorax anteroposterior: Metastasis-like lesions are detected in both bases.

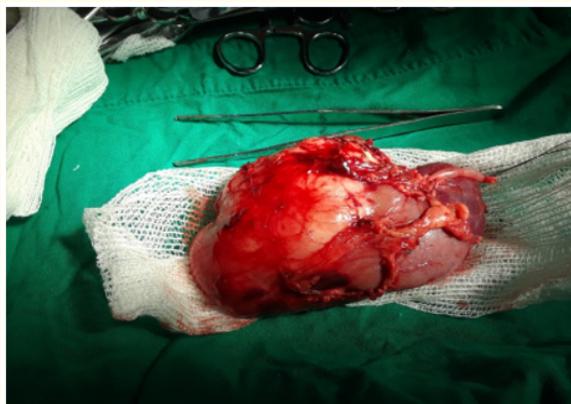
TAC contrasted tumor abdomen 119 mm x 102 mm x 95 heterogeneous lobed, with contrast uptake, without calcifications.

An echocardiogram is performed where light pulmonary stenosis is reported.

- Medulogram: Negative
- Bone marrow biopsy: Negative
- High milk dehydrogenase (LDH) figures: 560 U/l
- Accelerated erythro sedimentation: 130 mm/h.

It is agreed in the tumor committee to perform fine needle aspiration biopsy (FNAB) under ultrasound control and this was positive for malignant cells. Possible metastasis of a rhabdomyosarcoma.

It was decided in a given team the magnitude of the tumor to initiate a debulking chemotherapy: Ifosfamide, Vincristine and Actinomycin-D two preoperative cycles with a very good response and a 50% reduction in tumor volume, disappearance of lung metastases. Surgery was performed surgically on 1/11/2018 transverse to supraumbilical left laparotomy, completely resecting the left kidney with the upper tumoral two-thirds. The vessels are well connected, not infiltrating nearby structures (Figure 3).



**Figure 3:** Resected left kidney, upper two thirds of the tumor.

Result pathological anatomy embryonic rhabdomyosarcoma, without invasion of the capsule or vessels of the renal hilum, tumor-free resected ureter. Very good postoperative recovery, only remained in intensive therapy 24 hours.

Pathological anatomy result with immunohistochemistry.

Rhabdomyosarcoma with extensive maturation of rhabdomyoma.

#### **Immunohistochemistry:**

- CK: Negative
- Vimentina: Negative
- Desmin: Positive

Postoperative chemotherapy was given one week after surgery with the same treatment schedule for 3 postoperative cycles with 21 rest days between them.

In addition, a skin biopsy was obtained at the operative stage before starting the surgery for a cytogenetic study, which yielded 20 metaphases from mosaic aneuploidy.

#### **Discussion**

Primary retroperitoneal sarcomas are those solid or cystic tumors, malignant, that develop in the retroperitoneal space from tissues (lymphatic, nervous, vascular, muscular, connective and fibroareolar) independent of the organs and the great vessels contained in it as, the kidney, the suprarenal glands and the retroperitoneal parts of the pancreas, colon and duodenum [1,2]. Retroperitoneal rhabdomyosarcoma is a malignant primary tumor, which is one of the most frequent sarcomas in the pediatric age and less frequent in adults, constituting around 3.0% of solid tumors. It is usually diagnosed before the age of 30 in 70% of the cases, with the maximum ages of presentation between 18 and 30 years of age. Its location is genitourinary in 20% of cases and retroperitoneal and pelvic in approximately 5% of patients [1-3].

Rhabdomyosarcoma constitutes 50% of soft tissue sarcomas, which originates from skeletal muscle cells undergoing neoplastic transformation this tumor represents 10 - 15% of malignant solid tumors and 6% of all malignant neoplasms in children under 15 years of age [1]. It is more frequent in the male than in the female with a ratio of 1.5: 1, the tumor is twice as common in patients of Caucasian origin as in African-American patients and about 250 new cases are diagnosed each year in the United States [1,4,5]. It is suggested that

it can be associated with "identifiable" genetic risk factors: the most frequent of these genetic "syndromes" are Li-Fraumeni syndrome, Neurofibromatosis, Beckwith-Wiedemann syndrome and Costello syndrome [1]. The mutation of the p53 gene in the germline and anomalies in chromosome 11p15 is cited [1,5].

The diagnosis of primary retroperitoneal tumors is usually delayed, since the retroperitoneum is an adaptable space and the tumor remains asymptomatic for a long period of time, it can be clinically silent. It is not uncommon for the first symptom, although delayed, to be the appearance of a visible and palpable mass. On other occasions the symptoms derive from the compression or invasion of neighboring organs: frequent urination, sometimes with micturition or pain in different locations, unexplained fever and fatigue [6,7].

The diagnosis of retroperitoneal rhabdomyosarcomas is mainly carried out by imaging studies, without forgetting the clinical exploration and the analytical determinations. Currently, radiological exploration of the retroperitoneal space is based on the use of computed tomography (CT), especially with positron emission tomography with fluorodeoxyglucose and nuclear magnetic resonance (NMR), along with abdominal ultrasound [3,7,8].

Histologically there are four subtypes: embryonic, alveolar, botryoid and pleomorphic. The botryoid variant is located mainly in cavities such as bladder, vagina, bladder and medium urinary cavity [3]. It tends to metastasize other organs such as the lung, which is the most frequent organ, followed by the bone marrow, lymph nodes and bones [9]. This gene is probably involved in transcriptional modulation of other genes such as the oncogene c-Myc [1].

Surgery is the main pillar of the treatment, there are others such as chemotherapy and radiotherapy [10,11]. The resection route should be through laparotomy, with dissection of retroperitoneal blood vessels, retroperitoneal organs, such as the kidneys, spleen and pancreas, are often affected by the growth of the expansive tumor [12,13]. In our case the tumor was of renal origin, the rest of the surrounding structures were not affected. There are reports of the presence of a sentinel lymph node as a metastatic manifestation of the disease in advanced cases [14,15].

### Conclusion

There are few cases published with this variant of renal tumor in pediatrics, generally the most frequent renal tumors at this age are nephroblastoma, clear cell renal sarcoma and Rhabdoid tumor, also congenital mesoblastic nephroma, for what we consider important make it known to the medical community, even more because of its association with chromosomal abnormalities.

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