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CASE REPORT

RHABDOID TUMOR OF THE KIDNEY IN A FIVE MONTHS OLD FEMALE INFANT

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ABSTRACT

Rhabdoid tumor of the kidney is a very rare disease with poor overall survival. We report a five- month- old female infant who was diagnosed to have Stage IV Rhabdoid Tumor of the kidney at a tertiary institution in Ethiopia. She presented with hematuria of 1 week duration and on physical examination had a 6 x 8 cm right bimanually palpable abdominal mass. She had stage II hypertension. She had persistent hypercalcemia which resolved in the post-operative period. A left radical nephrectomy and para-aortic lymph node excision were done. The diagnosis was biopsy proven and supported by imaging. The infant was put on ifosfamide, carboplatin and etoposide but died 12 days after onset of the first cycle from sepsis. Rhabdoid tumor of the kidney is very rarely reported from low-income countries. Awareness of this disease may help in early detection and better prognosis.

***Keywords**: Rhabdoid Kidney. Hypercalcemia. Hypertension. Hematuria .Ethiopia*

INTRODUCTION

Rhabdoid tumors of the kidney (RTK) are one of the most lethal and aggressive diseases with a poor outcome. Rhabdoid tumor of the kidney was first reported as a distinct histopathologic entity in 1978 (1). It was described as a "rhabdomyosarcomatoid pattern".

This rare but aggressive malignancy accounts for 2% of paediatric renal tumours with 80% occurring in those under the age of 2 years and 60% in those under the age of 1 year, and has an overall male predominance (ratio 1·5:1) (2). RTK accounts for an estimated 0.9 -2 % of renal cancers (3, 4). The age ranges from 3 weeks to 94 months.

The median age was 7 months (3). Tumors of higher stage and younger infants were associated with an overall survival of 15.9% and 8.8%, respectively (5).
We report a five months old female infant who was diagnosed to have stage IV RTK and hypercalcemia at the largest tertiary hospital in Ethiopia, Tikur Ambessa Specialized Hospital (TASH).

Case presentation

A five months old female child from Adama City (99 kilometers outside Addis Ababa) presented with an initial complaint of recurrent reddish discoloration of urine of 1week duration.

The infant had no fever, vomiting or crying on micturition. She was diagnosed as having urinary tract infection at a local clinic and started on an unspecified antibiotic which she took for 4 days. She underwent an abdominal ultrasound scan since her symptoms had persisted. The study revealed a left renal mass and she was then referred to TASH.

The parents had not noticed any abdominal enlargement or palpable swelling earlier. The pregnancy was uneventful. Prenatal care ultrasound examinations done during antenatal care were non-revealing. The infant was born at term and had no events following delivery. The infant had no history suggestive of any bleeding disorders. Apart for the unspecified antibiotic, she had not been on any other drugs.

She was exclusively breast fed and vaccinated for her age according to the adopted national schedule. She was able to support her head at 3 months of age. She had started to sit up supported. She was the second child of two. Her parents and her elder sister were apparently healthy.

Her examination revealed a comfortable infant breast feeding quietly. The infant had no dysmorphic features. Apart for her BP, her other vital signs were within the normal range. Her blood pressure was in the range of 100 -118/ 85- 90 (Stage II hypertension).

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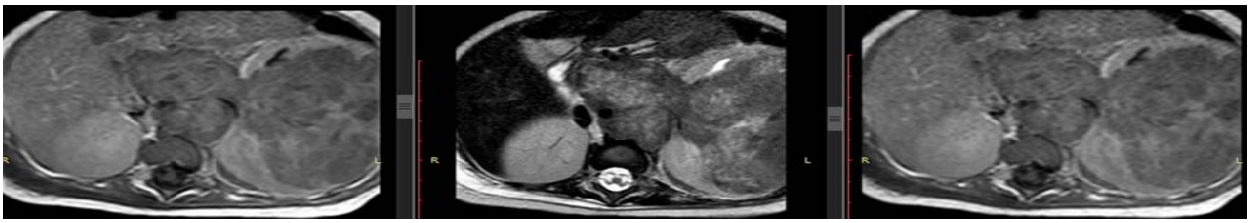
Her anthropometry was not affected. She had pink conjunctivas and non-icteric sclera. No palpable lymph nodes or glands. The pertinent physical finding was a 6 x 8 cm bimanually palpable non-tender firm lumbar right mass. In addition to these findings the infant had palmar pallor. The external female genitalia were normal. The infant was alert with a normal neurological examination. There were no abnormalities in other systems.

Prior to surgery, the laboratory studies revealed serum calcium in the range of 2.63 -5.88 mmol/L (normal range 1.1 – 1.25). Post- surgery serum calcium dropped to 1.23 mmol/L. The urine analysis revealed microscopic hematuria of 10 -12 RBC/mm3. CSF analysis was normal without any cells. The CSF cytology showed no malignant cells.

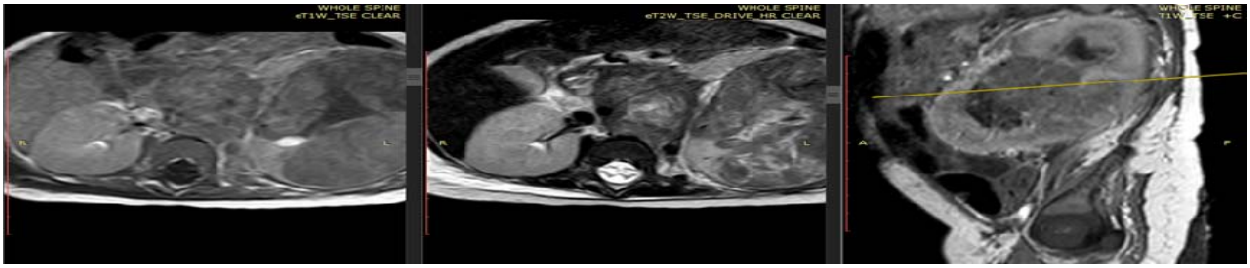
The infant was on anti-hypertensives (oral nifedipine, oral atenolol and repeated intravenous doses of hydralazine) medication for stage II hypertension. She showed no response to treatment. Following the surgery, the hypertension resolved and the medications were stopped.

The infant was started on ICE (Ifosfamide, carboplatin and etoposide) protocol. The infant developed febrile neutropenia post – chemotherapy. Her absolute neutrophil counts were below 500 / mm3 with temperature records of 38.5° C and above. Despite potent intravenous antibiotic coverage, she died within 72 hours of sepsis on day 12 post – ICE. Blood culture revealed no growth.

A Radiology report



A T1 axial T2 axial T1 + C sagittal



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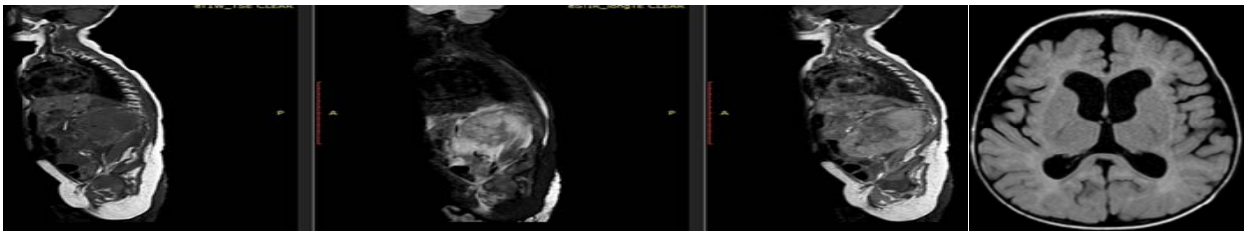
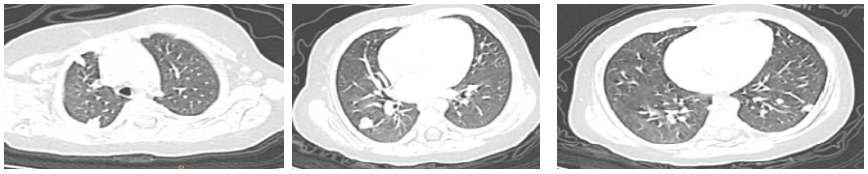


C T1 sagittal T2 sagittal T1 sagittal PC FLAIR



D : Chest CT scan

Figure 1: there is a huge heterogenous mass lesion with solid and cystic components in the left kidney with heterogenous enhancement forcing a moderate pelvi-calyceal dilatation. There are multiple sites of low signal intensities indicating tumor lobules. There are also associated left para-aortic huge lymph nodes elevating the aorta **(A&B)**. There are multiple lung nodules visible in the basal lung fields **D**. There was no brain or spinal lesions seen **C (PC: post contrast)**

Intra-operative findings

A left renal mass measuring approximately 8 x 7 cm in size was found. There was a huge para-aortic lymph node in the hilar area extending anteriorly which was closely adherent to the aorta.

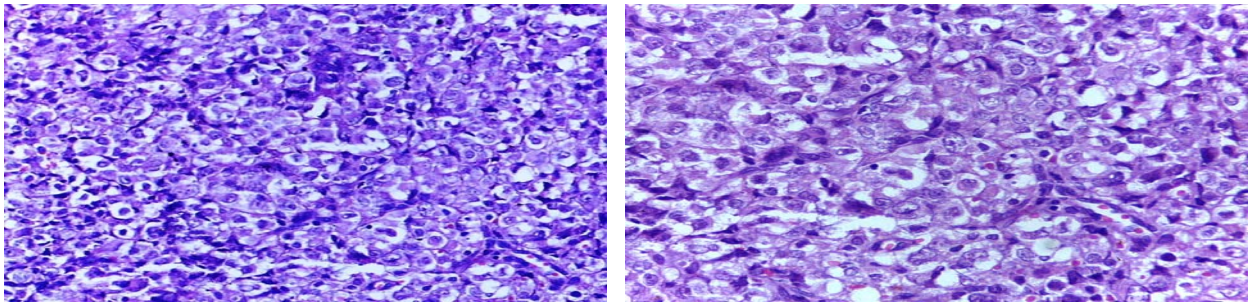
A left renal radical nephrectomy was done. Two lymph nodes were sampled. Since lymph nodes were in close proximity to the aorta, further sampling was not possible.

Pathology

Gross specimen examination revealed gray brown capsulated nephrectomy specimen measuring 8x6.5x6 cm. Cut-section showed grey-brown tumor mass measuring 7.5x 5x6cm filling most of the cortex and renal medulla with capsular invasion at two points. Non-neoplastic renal tissue measuring 0.5x1 cm and 0.5X0.5cm at each pole was present. About 40% was necrotic in the tumor mass.

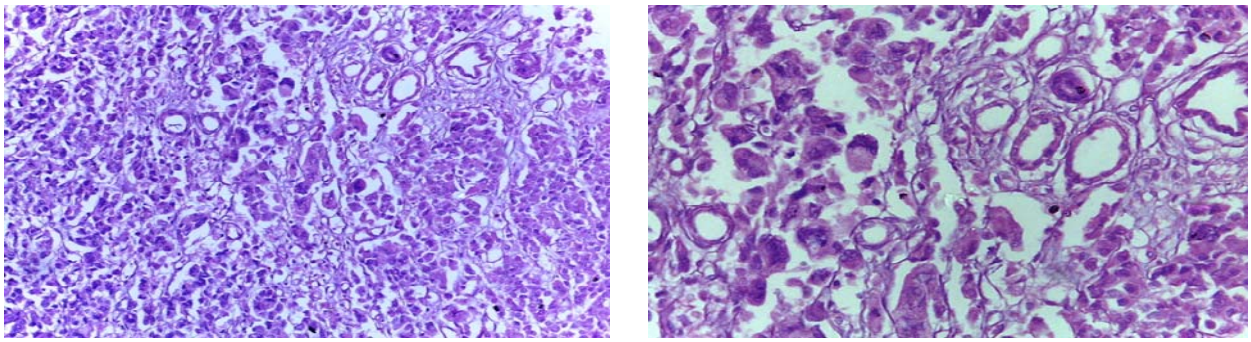
Two gray white para-aortic lymph nodes measured 0.8cm and 0.6cm in greatest dimension.

On histopathological examination, round to oval tumor cells were seen arranged in sheets, having pleomorphic vesicular nuclei and prominent nucleoli (Figure 2A and 2B). These tumor cells showed moderate to abundant amount of eosinophilic cytoplasm and hyaline intracytoplasmic inclusions (Figure 3A and 3B). There was increased mitotic activity with large areas of necrosis. Few sections showed renal tissue with glomeruli and tubules. Histologic sections from the para-aortic lymph nodes showed secondary infiltration by similar tumor cells**.**



A **B**

Figure 2: Rhabdoid tumor of the kidney: (A) round to oval tumor cells arranged in sheets, having pleomorphic vesicular nuclei and prominent nucleoli (hematoxylin & eosin, 200×), (B) High power microscopic findings of tumor cells with acidophilic cytoplasm (hematoxylin & eosin, 400×)



A **B**

Figure 3: (A) Large cells with eccentrically located nuclei and abundant, eosinophilic cytoplasm overrunning tubules (hematoxylin and eosin, 200×), (B) High power microscopic of similar tumor cells (hematoxylin & eosin, 400×).

DISCUSSION

The diagnosis of rhabdoid tumor of the kidney (RTK) was made based on the characteristic findings of vesicular chromatin, prominent nucleoli and hyaline cytoplasmic inclusions. With lung involvement, the infant was staged as stage IV at presentation (advanced disease). Tomlinson et al study showed most infants had advanced disease at presentation (5). In their publication, lung metastasis accounted for 28.9%. Most had multiple lesions in both lung fields and were unresectable as depicted in our report.

The inconsistent availability of proper diagnostics in low income countries makes it challenging to do immunohistochemistry and genetic testing. The RTK immunohistochemistry would be positive for vimentin, EMA and negative for INI1 (8). In addition, RTK and extra-renal rhabdoid tumors all share a SMARCB1 mutation at chromosome 22q11-12 (8, 9). This could be germline or somatic mutation .The median age group for germline mutations was 6 months of age (9). Considering our client's age, germline mutation is a possibility.

Symptoms, signs and laboratory evidence related to RTK include hematuria, hypertension, and hypercalcemia. Amar et al study showed that hematuria (gross/ microscopic) and hypercalcemia were present in 84% (22/37) and 26 %(6/23) of cases, respectively (10). Mary M et al study, hypercalcemia, hypertension and hematuria were present in 19%, 11% and 43 % respectively (11). The presence of hematuria, hypercalcemia and hypertension could be considered as supportive evidence for the disease in our case.

The characteristic radiologic evidence for RTK reported above (6, 7) and consistent intra-operative findings of extensive disease are supportive of the diagnosis.

The prognosis of young infants with advanced disease treated on conventional chemotherapy, in high income countries, has been dismal (5). Though our client died as a consequence of toxicity, she would have had a similar fate if she had survived.

ACKNOWLEDGEMENTS

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Conflict of Interest :
We have no conflict of interest to reveal.

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