

Case Report:

Malignant rhabdoid tumour of kidney - a rare aggressive tumour

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ABSTRACT

Malignant rhabdoid tumour of kidney is a rare highly aggressive neoplasm of childhood. We present the case of a 18-months old girl presenting with decreased appetite, abdominal distention of 20 days duration and 3 episodes of haematuria. The patient underwent left radical nephrectomy and histopathological examination of the excised specimen confirmed the diagnosis of malignant rhabdoid tumour of the kidney.

This case highlights the need to consider malignant rhabdoid tumour of the kidney of possibility young children in presenting with a renal mass.

Key words: Rhabdoid tumor, Malignancy, Childhood renal mass

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INTRODUCTION

Malignant rhabdoid tumour of kidney (MRTK) was first described as a variant of Wilms' tumour of the kidney till 1978. Later rhabdoid tumours outside the kidney were reported in many tissues which includes mainly the liver, soft tissue, and the central nervous system. The term "rhabdoid" was used due to its resemblance with rhabdomyosarcoma under the light microscope. The exact pathogenesis and cell of origin of MRT is unknown. Malignant rhabdoid tumours of kidney are rare and highly aggressive neoplasm of childhood. The clinical characters are non-specific and are diagnosed with histological as well as immunohistochemical characteristics and cytogenetic studies. This condition has very poor prognosis with overall survival rate not more than 20% to 25%.¹⁻⁴

CASE REPORT

An 18-month-old girl born out of non-consanguineous marriage presented to our

outpatient department with decreased appetite, abdominal distention of 20 days duration and 3 episodes of haematuria during that period. There was no history of fever, altered sensorium or seizures. There was no difficulty in accepting feeds, bowel disturbance or lower urinary tract symptoms. She had received 200 mL packed red blood cell transfusion for same ailment one week before coming to us. Physical examination revealed a large palpable mass involving (left hypochondrium, left lumbar, epigastrium and umbilical regions) which was non-tender. There was fullness in the left renal angle, spine was normal. Rectal examination was also normal. Laboratory investigations including haemogram, serum biochemistry including serum calcium were within normal limits. Contrast-enhanced computed tomography (CECT) of the abdomen showed a well defined heterogeneous mass replacing the left kidney displacing the surrounding structures, (Figures 1A and 1B). The clinical

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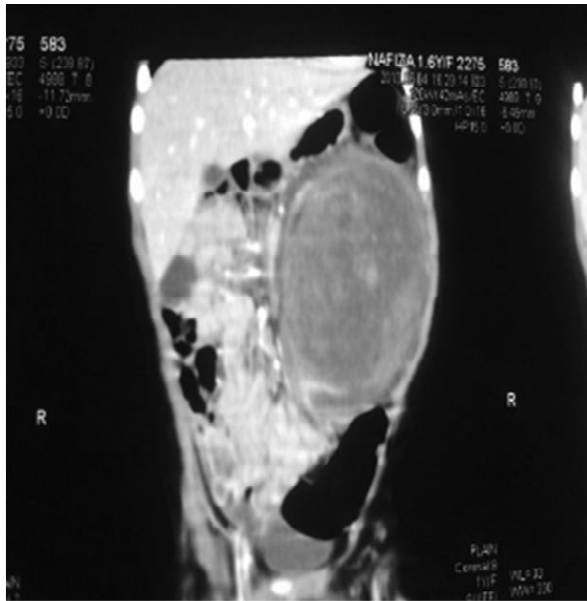


Figure 1A: CECT of abdomen (coronal section) showing a huge left renal mass

differential diagnosis was Wilms' tumour, retroperitoneal soft tissue tumour, and mesoblastic nephroma. Ultrasonography (USG) guided fine needle aspiration cytology (FNAC) from the lesion showed a high grade malignant tumour; the differential diagnosis considered included rhabdoid tumour and germ cell tumour.

She underwent left radical nephrectomy. Per-operatively there was a large left renal mass which was adherent to descending mesocolon and could be dissected freely with blunt dissection, the left adrenal gland was thinned out, one perihilar lymph node was identified, liver appears normal; there were no peritoneal deposits and no ascites. Tumour was resected *en-masse* after mobilizing from adherent descending mesocolon and surrounding structures (Figure 2). The procedure was uneventful and the respected specimen was sent for histopathological examination, immuno-histochemistry and a peripheral venous blood sample was sent for molecular studies. Histopathological examination revealed single oval capsulated grey white mass measuring $12 \times 11 \times 9$ cm and weighs 850 g. Cut-section



Figure 1B: CECT of abdomen (axial section) showing the mass displacing the surrounding structures

of showed grey-brown variegated mass measuring $12 \times 10 \times 8$ cm. Tumour was soft and necrotic. No capsular breach was seen. At the lower pole non-neoplastic renal tissue measuring 0.4×1 cm was present. On histopathological examination (Figures 3A and 3B), round to oval tumor cells were seen arranged in sheets and lobules. These tumour cells show moderate amount of eosinophilic cytoplasm pleomorphic vesicular nuclei and prominent nucleoli. There was increased mitotic activity ($>10/10$ high power fields) with large areas of haemorrhage



Figure 2: Gross specimen photograph of the excised retroperitoneal mass showing a variegated appearance

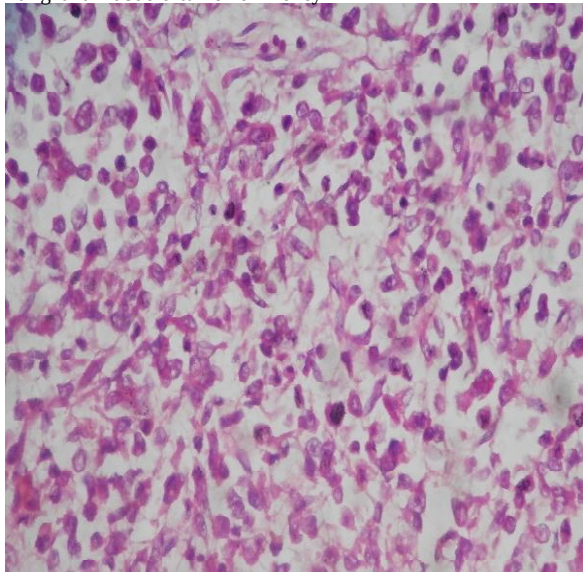


Figure 3A: Photo microphotograph of the tumour showing large cells with eccentrically located nuclei and abundant, eosinophilic cytoplasm with few non-neoplastic cells (Haematoxylin and eosin, × 200)

and necrosis. Few sections showed renal tissue with glomeruli and tubules. Immunohistochemistry (IHC) revealed diffuse nuclear positivity for vimentin and focal positivity for epithelial membrane antigen (EMA) suggestive of malignant rhabdoid tumour of the kidney (Figure 4A and 4B). The child recovered well post-operative period was uneventful and referred to medical oncology for further management.

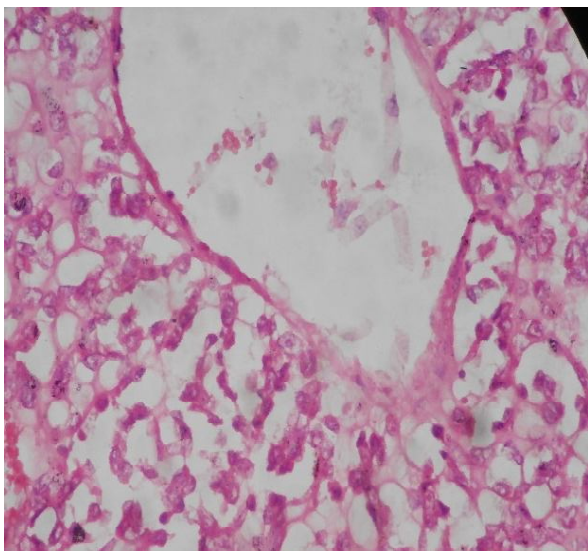


Figure 3B: Photomicrograph of the tumour showing large cells with eccentrically located nuclei and abundant, eosinophilic cytoplasm (Haematoxylin and eosin, × 400)

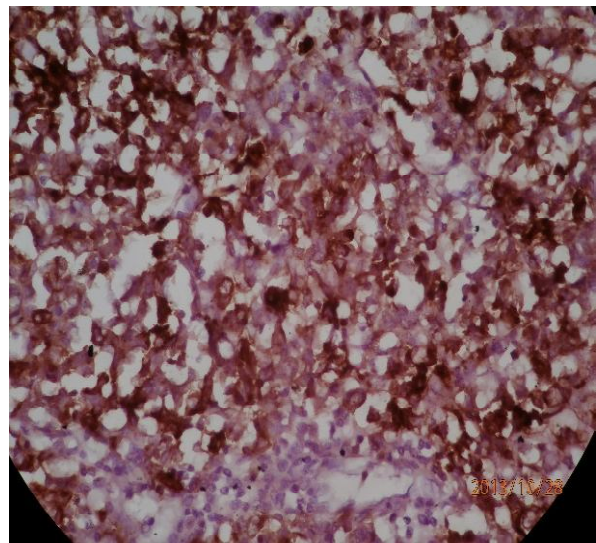


Figure 4A: Photomicrograph showing positivity for EMA (Immunohistochemistry, ×200)
EMA = epithelial membrane antigen

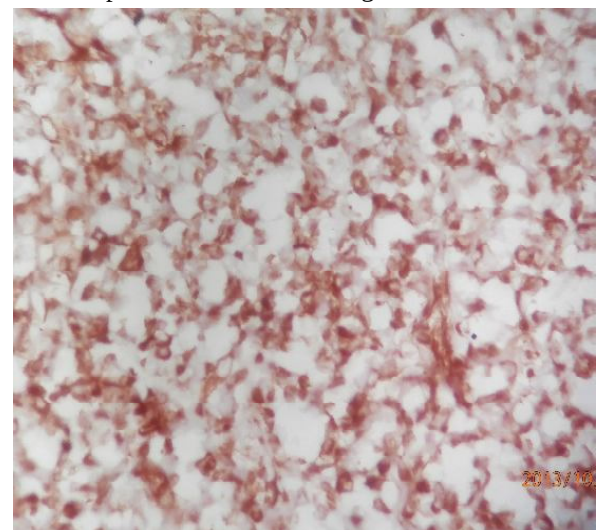


Figure 4B: Photomicrograph showing positivity for vimentin. (Immunohistochemistry, × 200)

DISCUSSION

Rhabdoid tumour of the kidney is a most aggressive and lethal renal tumour of childhood, which accounts for 2% of renal tumours as registered in the National Wilms' Tumor Staging Group (NWTSG).¹ This entity was originally described as a rhabdomyosarcomatoid variant of Wilms' tumour due to its resemblance of the cells to rhabdomyoblasts microscopically.^{2,8} Presently malignant rhabdoid tumour of the kidney is considered as sarcoma of the kidney and the exact cell of origin is unknown.¹

Malignant rhabdoid tumours are usually seen in kidney but have also been reported in extra-renal sites like brain, liver, urogenital system.^{2,9,10} Two cases are reported to arise from oesophagus.³

Patients with malignant rhabdoid tumour of the kidney present with abdominal mass, haematuria, flank pain and may also manifest reversible hypocalcaemia due to hyperparathyroidism. Symptoms are also seen due to metastasis, usually affecting the lungs, liver, brain, bone.² CT findings suggestive of rhabdoid tumour of kidney include calcification, sub-capsular hematoma and the lobular appearance of a large, centrally located heterogeneous mass.^{2,11} In our case, the child presented with haematuria, abdominal distention and imaging study showed huge left renal mass.

Histologically these tumours have a classical appearance that of pattern less sheets of large, non cohesive, ovoid or round to polygonal cells with vesicular nuclei and prominent nucleoli and large round eosinophilic cytoplasmic inclusion-like structures. Ultra structurally, rhabdoid cells have paranuclear intermediate filament aggregates, which are arranged haphazardly. Immuno-histochemically the rhabdoid cells stain diffusely positive for vimentin and focally positive for cytokeratin, EMA neuron-specific enolase (NSE) and *S-100* protein(s-100).²

Integrase interactor 1 (INI 1) gene is associated with chromatin remodelling and is expressed in all tissues. It is a product of a component of SWI/SNF chromatin remodeling complex (hsNF5/INI1) tumour suppressor gene on chromosome 22 which has been demonstrated to be frequently mutated or deleted in malignant rhabdoid tumour.²⁻⁷ Cell lines obtained from rhabdoid tumour of kidney and extra renal rhabdoid tumours have consistently expressed

the c-Abl tyrosine kinase proto-oncogene and showed reduced cellular growth in two cell-lines when treated with the tyrosine kinase inhibitors sorafenib.²

A multimodal approach with tumour resection to the extent possible, radiotherapy and chemotherapy are often employed together in the treatment of the malignant rhabdoid tumour. The prognosis is very poor in spite of much advancements. There is no significant difference in survival due to primary site of tumour, gender or ethnicity in children.² Children aged 2-18 years have a better prognosis compared to those younger than 2 years or who were older than 18 yrs.² Further studies are required to completely understand the natural course of the disease and the need for ideal treatment protocol for these highly malignant, aggressive tumours.

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