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# PAEDIATRIC RENAL PRIMITIVE NEUROECTODERMAL TUMOR: A CASE REPORT WITH LITERATURE REVIEW

Pathology

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## ABSTRACT

Primitive neuroectodermal tumor (PNET) of the kidney is a highly aggressive malignancy that predominantly affects young adults and is rarely seen in children and adolescents. populations. We report a case of a 16-year-old female who presented with an abdominal mass. Contrast-enhanced computed tomography (CECT) revealed a heterogeneously enhancing lesion in the left kidney, causing distortion of the renal contour, suggestive of a malignant renal neoplasm. The patient underwent radical nephrectomy, and histopathological examination supported by immunohistochemistry (IHC) confirmed the diagnosis of renal PNET. Timely diagnosis and early initiation of therapy are essential for improving clinical outcomes. A multidisciplinary approach is essential for an early diagnosis and timely management of the patients. This case highlights the necessity of including renal PNET in the differential diagnosis of renal masses, especially in younger patients.

## KEYWORDS

Kidney, primitive neuroectodermal tumor, small round cell tumor, Paediatric

## INTRODUCTION:

Primitive neuroectodermal tumor (PNET) of kidney is a rare and highly malignant small round cell tumor of neuroectodermal origin. According to literature, less than 200 cases have been reported worldwide. PNET is primarily found in brain, spinal cord and peripheral structures like bones or soft tissues.[1] Primary ES/PNET of the retroperitoneum, pelvis, paraspinal region and visceral organs like bladder, prostate, pancreas, ovaries, etc have been described in the literature.[2] However, it is rarely reported in the kidney. It is usually seen in young age with a median age of presentation around 30 years. Due to its rare nature and overlapping clinical features with other renal malignancies, renal PNET (rPNET) is usually not suspected initially and there is a significant dilemma in reaching the diagnosis. Diagnosis of rPNET is mainly based on histopathology, ancillary techniques like immunohistochemistry (IHC) and cytogenetic analysis. Renal PNET exhibits highly aggressive behaviour with poor prognosis.[1]

## A Case Report:

A 16-year-old female presented to the outpatient department with a complaint of pain in the left flank persisting for the past two months. She did not report any associated symptoms such as fever, loss of appetite or weight loss. Her past medical history was also unremarkable. Both general physical and systemic examinations were within normal limits. Laboratory investigations, including complete blood count, renal function tests, and liver function tests, all showed values within the normal range.

A contrast-enhanced CT scan revealed a heterogeneously enhancing mass in the left kidney, measuring 9.7 x 8.8 x 10 cm, with distortion of the renal outline, suggestive of a malignant renal tumor. The right kidney appeared normal in both size and function. After thorough evaluation, the patient underwent a left radical nephrectomy under general anesthesia. The excised specimen was sent to the Department of Pathology for histopathological analysis. Both the intraoperative and postoperative periods were uneventful.

**Histopathological Examination:** On gross examination, left nephrectomy specimen measured 12x11x8 cm and weighed 530 grams. Cut surface showed a gray white to tan tumor mass measuring 8x9x5cm, reaching upto pelvicalyceal system along with areas of necrosis and hemorrhage. Microscopic examination revealed clusters, sheets and nests of monomorphic round to oval tumor cells with high N:C ratio, scanty cytoplasm, pleomorphic nucleus and inconspicuous nucleoli. Numerous rosettes were also noted. Tumor was infiltrating the renal pelvis, perinephric fat and Gerota's fascia. Sections from the ureter and renal vessels also revealed tumor infiltration. On immunohistochemistry, tumor cells showed membranous positivity for CD99 and NSE, patchy positive for Vimentin and were negative for CK, CD10, CD56, Synaptophysin, WT1, PAX-8. Based on histopatholomorpholical evaluation and IHC findings, the diagnosis of rPNET was made. (Figure 1)

The patient recovered well in the postoperative period and was discharged. After discharge the patient died after 6 months.

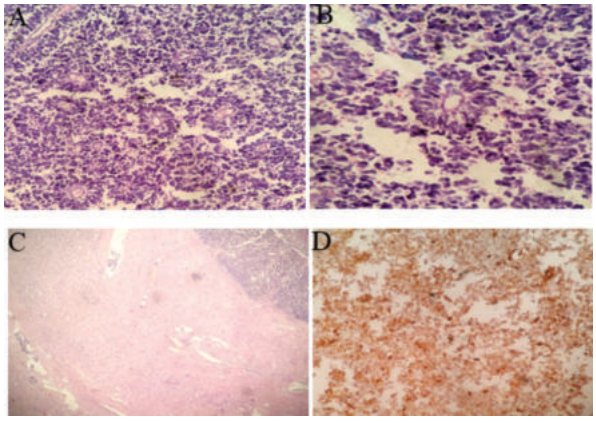


**Figure 1.** A) Section showing round to oval tumor cells having pleomorphic nucleus, scant cytoplasm and forming rosettes at place [Hematoxylin and Eosin stain (200X)]. B) Tumor cells forming rossetes (Hematoxylin and Eosin stain, 400X). C) Vascular and capsular invasion with neoplastic cells (Hematoxylin and Eeosin stain, 100X). D) Tumor cells showing CD99 cytoplasmic positivity (IHC,200X).

## DISCUSSION:

Pediatric renal tumors account for about 6–7% of all solid neoplasms in children, with Wilm's tumor being the most common type. PNET represent fewer than 5% of these renal malignancies.[1]Renal PNET was

first identified by Arthur Purdy Stout in 1918, as a "small round tumor." Although peripheral PNET usually originate in bones or soft tissues, their recurrence in kidney is uncommon.[3] These tumors are more frequently observed in young adults, with only a limited number of cases documented in individuals under 18 years old.[4] The condition shows a male predominance, with a male-to-female ratio of 3:1.

Although the origin of the tumor remains uncertain, studies suggest that it arises from cells that have migrated from the neural tube and possess varying abilities to differentiate into ectodermal or neuronal tissues. These tumors can develop anywhere along the central nervous system, from the brain to the sacrum.[3] The hallmark feature of these tumors is specific chromosomal translocation t(11;22)(q24;q12), which results in the fusion of the EWS1 and FLI1 genes.[5]

The clinical presentation of rPNET is non-specific and show similarity with other renal tumors. Flank pain is the most common manifestation (67.5%) followed by hematuria (33.8%), mass (33.8%), inferior vena cava thrombosis (25%) and weight loss (16%).[4] These tumors mostly arise from medullary or pelvic region and can infiltrate the entire kidney. There might be invasion into the renal vein and inferior vena cava in several cases.[2]

Radioimaging techniques (USG, CECT, magnetic resonance imaging etc.) plays crucial role in detecting, localizing and extent of invasion of tumor to help the operating surgeon to frame better operative plan. However, imaging is not helpful for the diagnosis of rPNET because there are no distinctive radiological findings and other renal tumors like Wilm's tumor may also show similar findings.[1]

The diagnosis of PNET mainly rely on histological and IHC findings. Microscopically, sections examined show primitive small, uniform, round to oval tumor cells with high N:C ratio, inconspicuous nucleoli, scant cytoplasm arranged in sheets and nests. Homer-Wright rosettes seen in rPNET are the important diagnostic histological features of renal PNET.[1]

Considering the age group, overlapping clinical manifestation and morphological features, other differentials include blastemal predominant Wilm's tumor, renal cell carcinoma, malignant lymphoma, neuroblastoma and desmoplastic small round cell tumor (DSRCT) tumor for which a panel of IHC markers needs to be applied.[4] In our case positive expression of CD99, NSE and vimentin was noted. There was negative expression of WT1 and PAX8 ruling out Wilm's tumor and renal cell carcinoma respectively. Further IHC applied were CD10, synaptophysin and CD56, which came out to be negative and differentiating rPNET from lymphoma and neuroblastoma. Based on morphology and IHC findings a diagnosis of rPNET was made.

The diagnosis of rPNET can be confirmed by cytogenetic analysis expressing t(11:22)(q24;q12) [EWS-FLI-1 gene fusion] either by fluorescent in situ hybridization technique or reverse transcriptase-polymerase chain reaction.[2] Molecular markers (EWS-FLI chromosomal translocation) have proven to be extremely helpful when histology and immunohistochemistry fails to provide a definite diagnosis, particularly in situations of small biopsies/atypical morphology in renal mass.[4]

The PNET tumor family is classified into three groups based on their tissue of origin.[6] The classification is as follows:
**Group I:** Central nervous system (CNS) PNETs – originating from the CNS
**Group II:** Neuroblastomas – arising from the autonomic nervous system
**Group III:** Peripheral PNETs (pPNETs) – originating from tissues outside both the central and autonomic nervous systems. Our patient falls under Group III, which includes renal PNETs.

Renal PNETs are notably more aggressive than other renal tumors, characterized by a high propensity for metastasis. The lungs and bones are the most frequent sites of dissemination, with secondary involvement of the liver, bone marrow, and leptomeninges. The metastatic potential highlights the tumor's aggressive nature. Prognosis is closely linked to disease stage, with a 5-year survival rate of approximately 70% in localized cases, which declines to about 40% in those with metastatic disease.[1]

Treatment options include combination of surgery, chemotherapy, and radiotherapy. The preferred surgical approach is radical nephrectomy. Postoperative chemotherapy is advised when there is local invasion into the perinephric tissue or when frozen section analysis reveals positive surgical margins. For patients with unresectable tumors, positive margins, or residual disease, radiotherapy can be beneficial.[1] Disease-free survival rate (5 years)is around 45–55% in kidney-confined cases, whereas, these tumors have a 5 year survival rate of 20-30% in cases with advanced metastatic disease and median relapse-free survival of 2 years.[2]

These tumors are quite rare, hence no definite management protocols have been documented . A multidisciplinary approach is recommended for treatment of such patients in view of the poor prognosis of the disease.[2] Despite the use of various treatment approaches, including high-dose chemotherapy and autologous stem cell rescue, the outcome for metastatic rPNET still remains poor. However, children and young adults tend to have a more favorable prognosis.[4]

## CONCLUSION:

Peripheral neuroectodermal tumor (PNET) of the kidney is a rare and aggressive malignancy that should be considered in the differential diagnosis of large renal masses in adolescents and young adults. Definitive diagnosis is established through histopathological analysis and immunohistochemistry. Optimal outcomes are achieved with multimodal treatment approach incorporating surgery, chemotherapy, and radiotherapy delivered in specialized tertiary care centers with appropriate expertise and facilities.

**Conflicts Of Interest:** None

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