ABSTRACT

Ewing sarcoma/primitive neuroectodermal tumor (PNET) is a high-grade malignant neoplasm commonly affecting bones of the thoracic region. Primary Ewing sarcoma/PNET of the kidney is exceptional; it commonly affects young adults and is rarely reported in children.

Here we describe a case of renal PNET in a 10-year-old girl who presented at our institute with haematuria and abdominal lump. Computer tomography scan revealed a huge mass in the right kidney. Computer tomography-guided aspiration from the mass showed cytomorphological features of a small blue round cell tumor. The patient underwent right radical nephrectomy. Histopathology with supplementary immunohistochemistry confirmed the diagnosis of PNET. She has been receiving treatment with adjuvant chemotherapy post-surgery and is currently disease free.

Primary renal PNET is a distinct and rare entity. This tumor is very aggressive with low survival rate, even with a multi-disciplinary approach. We reported the case because it is rare in children, poses diagnostic challenges, is aggressive in behaviour and responds poorly to treatment.

Key Words: Ewing sarcoma, Kidney, Primitive neuroectodermal tumor

INTRODUCTION

Primitive neuroectodermal tumor (PNET) is a malignant neoplasm that originates from neural crest cells and the neuroectoderm that are found during normal human development. These tumors are broadly classified as central (CNS PNET) and peripheral, which is also known as Ewing sarcoma/primitive neuro-ectodermal tumor (ES/PNET) due to its signature genetic alteration. ES/PNET is primarily a bone-soft tissue neoplasm. Primary ES/PNET of the kidney is an exceptionally rare malignant lesion (1). Median age of presentation is 27 years; it is extremely uncommon in <15 years and only a few sporadic cases have been reported so far (2). Conclusive diagnosis is done by morphology, immunohistochemistry (IHC) or molecular techniques. Patients are managed aggressively with surgery, chemotherapy with/without adjuvant radiotherapy. A commonly employed regimen includes combination chemotherapy with vincristine, adriamycin, cyclophosphamide (VAC) with alternating ifosfamide and etoposide (IE) (3). Response to treatment is often poor even with multimodality treatment. Overall prognosis and survival is mostly bleak though younger patients often have better survival than older ones (4). Here we report a case of primary ES/PNET of the kidney in a 10-year-old female.

CASE REPORT

A 10-year-old girl presented with intermittent haematuria for one week. Detailed clinical history revealed complaints of right-sided abdominal discomfort and generalised weakness for three months prior to the onset of haematuria. Examination revealed a palpable mass in the right lumbar region. Blood and urine examination showed mild anemia and presence of RBCs in the urine. Urine culture was negative. Serum creatinine was within normal limits. Contrast-enhanced Computer Tomography (CECT) Scan showed loss of normal right renal architecture and replacement with a huge mass (10.44x9.02 cm), displaying hyper-dense enhancing and hypo-dense non-enhancing areas, without any excretion of contrast, suggesting a neoplastic lesion (Figure 1A). The left kidney was normal. A CT-scan guided aspiration from the mass showed cytomorphological features of a small blue round cell tumor. The patient subsequently underwent right radical nephrectomy.

Grossly, the specimen measured 14.5x10x8 cm and weighed 700 gm. The cut surface showed that almost the entire renal parenchyma was replaced by a solid, grey white tumor with areas of haemorrhage, necrosis, and cystic degenerations.
Radiotherapy for further management. She was started on chemotherapy four weeks post-surgery and has completed two cycles of chemotherapy without any obvious metastasis or recurrence.

DISCUSSION

PNET/ES covers one percent of all sarcomas (5). It is a malignant small round cell tumor of neural crest origin, first described in the ulnar nerve by Arthur Pourdy Stout in 1918 (6). Commonly, PNETs arise in the ribs and paraspinal areas; involvement of skin, soft tissue, lungs, kidney, and retroperitoneum is rare. Renal PNET was first described almost sixty years later than the first report of PNET, in 1975; few cases have been reported so far (7). Till now, around 100 cases have been reported worldwide, with very few from India (8).

Renal PNET commonly affects young adults, though the age range is 4 - 61 years in reported cases. It more commonly affects males, with a male to female ratio of 3:1 (8). Most cases of renal PNET are diagnosed on resected specimens, based on histopathology and immunohistochemistry. The differential diagnosis includes other small round cell tumors, including Wilms’ tumor, neuroblastoma, and lymphoma. These tumors are composed of primitive-appearing round cells with a high nucleo-cytoplasmic ratio. Perivascular pseudo-rosettes are usually identified; Homer-Wright rosettes are less frequently seen. A commonly appreciated feature on electron microscopy is aggregates of cytoplasmic glycogen granules though sometimes polar processes, microtubules or neurosecretory granules are seen, suggesting a neuronal differentiation (9). Tumor cells...
In conclusion, primary renal PNET is a distinct and rare entity, typically affecting young adults. This tumor is very aggressive with a low survival rate, even with multi-modality treatment. Although the incidence of renal PNET in children is low, oncologists and pathologists need to be aware of this tumor and every attempt should be made to differentiate it from other more common tumors as it carries very poor prognosis. Morphology alone can only suggest PNET as an important differential of small round cell tumors; ancillary techniques and immunohistochemistry for CD99 with or without a molecular test are vital to establish a correct diagnosis. We reported this case for its aggressive nature and rarity in children.

REFERENCES


