Malignant Rhabdoid Tumour of the Perineum in an Adult—A Rare Presentation

Rajdeep Singh · Baljeet Kumar Basra · Nita Khurana · Anil Kumar Sarda

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Abstract Rhabdoid tumors of extrarenal origin are uncommon. This is the first report of rhabdoid tumor presenting as a perineal mass. Furthermore, it is a disease of childhood, with adult presentation being rare. We report a case of extrarenal rhabdoid tumor and discuss its presentation and possible treatment options.

Keywords Rhabdoid tumor · Extrarenal rhabdoid · Perineal tumor

Introduction

A malignant rhabdoid tumor was first described as a variant of Wilms' tumor with poor prognosis [1]. Having been described at extrarenal sites also [2, 3], they are tumors of childhood with a mean age of presentation at 6 years [3, 4].

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Presentations This case report has never been presented at a meeting.

R. Singh · B. K. Basra · A. K. Sarda
Department of Surgery,
Maulana Azad Medical College & Lok Nayak Hospital,
New Delhi 110002, India

N. Khurana
Department of Pathology,
Maulana Azad Medical College & Lok Nayak Hospital,
New Delhi 110002, India

R. Singh (✉)
Department of Surgery, BL Taneja Block,
Maulana Azad Medical College,
New Delhi 110002, India
e-mail: rajdeep.mamc@gmail.com

We report the first case of malignant extrarenal rhabdoid tumor presenting with a perineal mass during adulthood.

Case Report

A 25-year-old man presented with a progressively increasing perineal mass for 2 years and urinary retention for 1 day.

General and systemic examination was essentially normal. Apart from a distended urinary bladder, abdominal examination was normal. Local examination of perineum revealed a large (15 cm×15 cm) hard mass at the root of penis, lying posterior to the scrotal sac, stretching the skin of the posterior wall of the scrotum and pushing both the testes and the shaft of the penis anteriorly. The mass extended anterosuperiorly, involving the bulbar urethra, and fixed to the bone. On digital rectal examination, the rectum was spared but the prostate and seminal vesicles could not be felt separate from the mass. Overlying skin was thickened, indurated, and ulcerated. Multiple bilateral inguinal lymph nodes were present, largest measuring 2.5 cm×2.5 cm, fixed to underlying muscles, ligaments, and overlying skin.

A provisional diagnosis of urethral/periurethral gland malignancy or a soft tissue sarcoma was considered. Contrast-enhanced CT defined a solid mass lesion at the root of the penis and the base of the scrotum, involving bilateral pubic rami with osteolytic destruction of the bone (Fig. 1). Enlarged lymph nodes were present along both iliac arteries. Multiple pulmonary lesions suggestive of metastases were also noted. Bilateral testes, kidneys, ureters, liver, and retroperitoneum were unremarkable. A diagnostic cystourethroscopy was attempted but the scope could be passed till the middle of penile urethra only.

Fine-needle aspiration cytology (FNAC) from the mass suggested a malignant mixed germ cell tumor, possibly

Fig. 1 Contrast-enhanced CT image shows invasion and destruction of pubic bones and symphysis. The mass is infiltrating and encasing the urethra

seminoma and embryonal carcinoma, or a malignant teratoma. However, testicular tumor markers were within normal limits: LDH 289 U/l ($n=200\text{--}400$), α FP 1.9 ng/ml ($n=2\text{--}10$), β HCG 0.8 mIU/ml ($n<1.2$). FNAC from the left inguinal lymph nodes showed evidence of metastasis, but the type could not be ascertained.

Trucut biopsy of the mass revealed sheets of large polygonal cells with eccentric nuclei, prominent nucleoli and abundant dense eosinophilic cytoplasm with round cytoplasmic inclusions (Fig. 2a). Immunohistochemistry demonstrated expression of vimentin (Fig. 2b) and cytokeratin (Fig. 2c). These features were consistent with a diagnosis of malignant rhabdoid tumor.

In view of extensive local involvement and metastasis, surgical resection was ruled out. Suprapubic cystostomy for relief of urinary obstruction was done. The patient refused trial of chemotherapy.

Fig. 2 Photomicrograph of trucut biopsy: **a** sheets of large polygonal cells with eccentric nuclei, prominent nucleoli and abundant eosinophilic cytoplasm with round cytoplasmic inclusions (H&E $\times 400$), **b** immunohistochemistry showing expression of cytokeratin (streptavidin peroxidase stain $\times 400$), and **c** immunohistochemistry showing expression of vimentin (streptavidin peroxidase $\times 400$)

Discussion

Patients of Wilms' tumor having a rhabdoid (rod-like) pattern had a poor outcome irrespective of the treatment given, which were then classified as a distinct tumor [5]. Under light microscopy, rhabdoid tumors are characterized by diffuse growth of round to polygonal cells, with ample cytoplasm and large nuclei. The characteristic feature is presence of eosinophilic inclusions in the cytoplasm, composed of whorls of intermediate filaments [5]. Immunohistochemically, rhabdoid tumor cells stain with vimentin, cytokeratin, neuron-specific enolase (NSE), and epithelial membrane antigen (EMA). Although the precursor cell is not known, the presence of vimentin suggests a mesenchymal origin.

These tumors may exist in a pure or composite form, composite form being characterized by the presence of other tissue elements such as renal cell carcinoma, transitional cell carcinoma, squamous cell carcinoma, sarcomas, or malignant melanoma [2]. Genetic analysis has shown that composite tumors are distinct from pure rhabdoid tumors inasmuch as they do not have mutations of INI1 gene at 22q11 [6]. This gene may be a tumor-suppressor gene [7] and its abnormality is uniformly found in all pure rhabdoid tumors.

That rhabdoid tumor is rarely the first clinical diagnosis is evident from the present case. We believe that FNAC is misleading, and a trucut biopsy should be the modality of choice in all palpable tumors. Imaging serves to define the extent of disease, which is vital since adequate resection provides the best chance for control of disease. Aggressive chemotherapy and radiotherapy have improved short-term survival [8], but overall survival remains dismal. Chemotherapeutic agents used for rhabdoid tumors include actinomycin D, vincristine, cisplatin, adriamycin, cyclophosphamide, ifosfamide, and etoposide, with no definite advantage of one over the other [8]. Due to rarity of this disease, no treatment protocols have been defined. The disease is uniformly fatal.

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