Case Report

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Pathology Section

Primary Malignant Rhabdoid Tumour of the Urinary Bladder Presenting in an Adolescent: A Case Report

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ABSTRACT

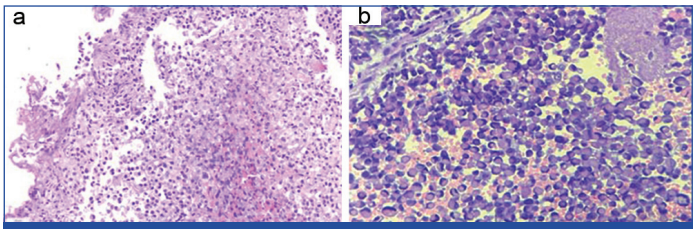
Primary Malignant Rhabdoid Tumour (PMRT) of the urinary bladder is an extremely rare, undifferentiated malignant tumour. Most cases of rhabdoid tumours present below three years of age and presentation during adolescence is extremely unusual. The common sites of occurrence for PMRTs are kidneys and the central nervous system. PMRTs are highly aggressive tumours with poor prognosis and a propensity for metastasis. A 15-year-old female presented with haematuria and Lower Urinary Tract Symptoms (LUTS) since one year. She developed severe anaemia for which two units of packed red cells were recently transfused. Contrast Enhanced Computed Tomography (CECT) showed a large, enhancing, exophytic mass arising from the right lateral wall of the urinary bladder, measuring 8.8×6.3×6.1 cm. A Transurethral Resection of Urinary Bladder Tumour (TURBT) was done which showed a high-grade malignant tumour with large, non-cohesive cells with a rhabdoid morphology. Immunohistochemistry (IHC) was positive for EMA, vimentin and pancytokeratin. The tumour was negative for LCA, CD20, CD3, MPO, CD34, myogenin, desmin, GATA3 and uroplakin III. INI1 showed complete loss of nuclear expression in the tumour cells and a diagnosis of PMRT was given. A month later, a radical cystectomy with urethrectomy and lymph node dissection was received. A large fungating mass was seen on the right lateral wall of the urinary bladder. Histopathology showed similar features of malignant rhabdoid tumour similar to the TURBT, with invasion into lamina propria as well as the muscularis propria. No lymph node metastasis was seen. No CNS or renal tumour was seen. A positive vimentin and loss of INI1 (SMARCB1) is diagnostic of rhabdoid tumour. Differential diagnosis of PMRT includes rhabdoid variant of urothelial carcinoma, plasmacytoid variant of urothelial carcinoma, rhabdomyosarcoma, and haematolymphoid malignancies. Loss of INI1 establishes the diagnosis. Although rare in adolescents and rare in location, PMRTs of the urinary bladder should not be overlooked and an early, accurate diagnosis is warranted in view of its aggressive nature.

Keywords: Haematuria, INI1, SMARCB1, Urinary bladder neoplasms, Urogenital neoplasms

CASE REPORT

A 15-year-old presented at a tertiary care centre with haematuria and LUTS since one year. She also had severe anaemia for which two units of packed red cells were recently transfused. CECT scans showed a large enhancing exophytic mass measuring 8.8×6.3×6.1 cm arising from the right lateral wall of the urinary bladder. No lump/tumour was seen anywhere in the body.

A TURBT was done which showed a high-grade malignant tumour arranged in sheets and solid nests with large non-cohesive cells with rhabdoid morphology. The tumour also showed focal microcystic areas and perivascular pattern of growth. Individual cells were oval to polygonal with large eccentric nuclei and prominent nucleoli. Areas of necrosis and brisk mitosis were seen in the tumour. On light microscopy, differential diagnosis of malignant rhabdoid tumour, rhabdoid variant of urothelial carcinoma, rhabdomyosarcoma and haematolymphoid malignancies were contemplated [Table/Fig-1].

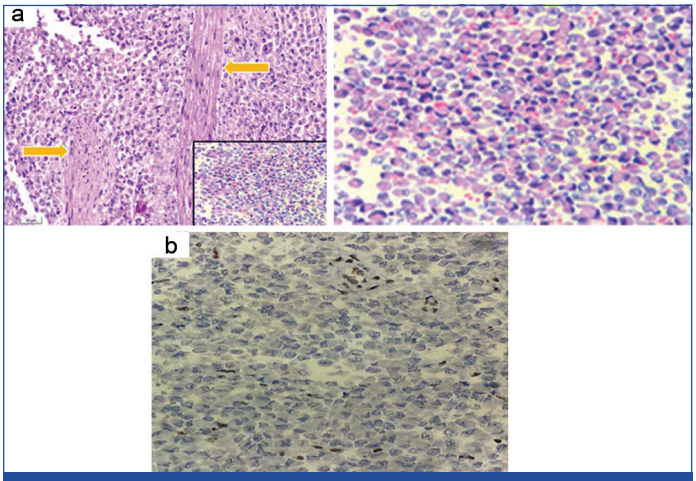


[Table/Fig-1]: a) TURBT showing a tumour with sheets of large, discrete malignant cells (H&Ex100); b) Tumour cells showing rhabdoid morphology with eccentric nuclei, prominent nucleoli and abundant eosinophilic cytoplasm (H&E 400x).

A month later a radical cystectomy with urethrectomy with a lymph node dissection was done and the specimen sent for histopathology [Table/Fig-2]. Histopathological examination of the tumour confirmed a malignant rhabdoid tumour of the urinary bladder. The tumour invaded deep into the muscularis propria (outer half) and was staged as pT2b [Table/Fig-3a]. None of the lymph nodes showed metastasis. The patient did not have any central nervous system or renal tumour.

[Table/Fig-2]: Cystectomy specimen showing a large tumour measuring 8.8×6.3×6.1 cm, arising from the right lateral wall of the bladder.

IHC was performed, which showed that the tumour cells were strongly positive for EMA and vimentin, with focal positivity for Pan-Cytokeratin (PanCK). BRG1 expression was retained in the tumour cells. The tumour cells were negative for LCA, CD20, CD3, MPO, CD34, myogenin, desmin, GATA3, uroplakin III, SALL4, MyoD1, and SOX10. INI1 showed complete loss of expression in the tumour cells, supporting a diagnosis of PMRT [Table/Fig-3b]. Patient was on follow-up till the writing of this report and was declared tumour free.



[Table/Fig-3]: a) Cystectomy specimen showing sheets of rhabdoid cells extensively involving the muscularis propria. (Arrows showing muscularis propria) (H&Ex200); Inset showing rhabdoid cells (40x); b) INI1 immunostain showing complete loss of nuclear expression in the tumour cells, indicative of a rhabdoid tumour (INI1x400).

DISCUSSION

The PMRTs of the urinary bladder are extremely rare and highly aggressive neoplasms that have only been described in a small number of cases worldwide. Although rhabdoid tumours more commonly arise in the kidney or central nervous system in infants and young children, pure rhabdoid tumours confined to the bladder have been reported sporadically, with only a couple of dozen documented cases across all age groups. These tumours display a broad age spectrum, the majority occur in young paediatric patients, particularly those under five years of age, but older children and even adults (e.g., adolescents and individuals in their 20s to 40s) have also been affected [1-4]. The documented tumour size ranges widely, from small lesions of around 2 cm to large masses exceeding 15 cm, and a female preponderance has been noted among reported cases, with markedly more females affected than males [1,2,5,6].

Clinically, PMRTs of the bladder often present with symptoms related to intravesical mass effects. Gross haematuria is one of the most common initial manifestations, accompanied in many cases by irritative LUTS and occasionally nonspecific complaints such as pelvic discomfort or decreased appetite. Imaging studies such as ultrasound and CT scans typically reveal a solid, heterogeneous bladder mass that may project into the lumen and involve any bladder wall region [7-9]. Given the propensity of rhabdoid tumours to disseminate early, staging with full-body imaging is essential to assess for metastatic disease, including evaluation of the lungs, liver, bones and, in some settings, central nervous system involvement [4,5,7,10,11].

Accurate diagnosis hinges on a combination of histopathologic evaluation, IHC, and molecular analysis. Histologically, PMRTs are composed of sheets of neoplastic cells that feature eccentric nuclei, prominent nucleoli, and abundant eosinophilic cytoplasm, often with cytoplasmic inclusions characteristic but not wholly specific features of rhabdoid morphology [5,6,8,12,13]. The cornerstone of definitive diagnosis is immunohistochemical loss of nuclear INI1 (SMARCB1) expression, reflecting underlying biallelic inactivation of the SMARCB1 tumour suppressor gene on chromosome 22q11.2. This loss of INI1 expression is the genetic hallmark in approximately 98% of classic malignant rhabdoid tumours and strongly supports the diagnosis when correlated with morphology [1,2,12-15]. While rhabdoid-like cells may occasionally be seen in other bladder malignancies such as urothelial carcinomas with rhabdoid differentiation, these generally retain INI1 expression, underscoring the importance of IHC and, when necessary, molecular testing to distinguish true PMRT from other entities [15-17].

The differential diagnosis for a poorly differentiated bladder mass with rhabdoid features is broad and includes tumours such as rhabdomyosarcoma, poorly differentiated urothelial carcinoma with rhabdoid variant, and Primitive Neuroectodermal Tumours (PNET). Distinguishing these entities is critical because their management and prognosis differ significantly. For example, rhabdomyosarcomas typically express muscle markers like desmin and myogenin, while true rhabdoid tumours lack these markers and instead show loss of INI1. Similarly, while high-grade urothelial carcinomas may display focal rhabdoid morphology, they usually retain INI1 nuclear expression and demonstrate typical urothelial markers on IHC [5,8,9,13,14,16].

Management of PMRT of the bladder is multimodal and tailored to the individual patient due to the rarity of these tumours and absence of standardised treatment protocols. Surgical intervention ranges from bladder-preserving approaches such as transurethral resection or partial cystectomy to more extensive procedures like radical cystectomy when appropriate. Given the systemic aggressiveness of rhabdoid tumours, most patients receive combination chemotherapy regimens incorporating agents such as vincristine, cyclophosphamide, doxorubicin, etoposide, ifosfamide, and platinum compounds, often adapted from protocols used in other paediatric sarcomas or rhabdoid tumour protocols. In selected cases, radiotherapy has been added for local control, particularly when complete surgical resection is not feasible or when there are high-risk features. Notably, some case reports describe successful outcomes with bladder-preserving strategies combined with intensive chemotherapy and radiotherapy, highlighting the potential for long-term remission with early and aggressive multimodal therapy in select patients [9,10,14,16,17]. [Table/Fig-4] presents the list of 16 cases of PMRTs of the urinary bladder reported in literature.

S. No.	Year/Reference	Age/Gender	Site	Size (cm)	IHC positive	IHC negative	Molecular testing	Outcome
1.	2024, Li J et al., [1]	4 years/F	Bladder wall	2.8	NA	INI1	+	Alive 15 months after diagnosis
2.	2024, Huang HC et al., [2]	1-year 9 months/F	Bladder wall	3.2	AE1/AE3, CAM5.2, glypican-3, CD34, SALL4	INI1, myogenin, MyoD1, S100 and SOX10	-	Recurrence and symptom free at follow-up
3.	2020, Tang V et al., [3]	1.5 months/ F	Bladder (posterior inferior wall)	2	NA	INI1, Melan-A, synaptophysin	+	Died of disease
4.	2020, Hecht SL et al., [4]	18 months/F	Left postero-lateral wall of bladder	5.4	NA	INI1	-	Alive 6 weeks after diagnosis
5.	2019, Hoare DT et al., [5]	Antenatal	Bladder	1.8	AE1/AE3, vimentin, MIB-1	INI1	-	Alive 18 months after diagnosis with no recurrence

6.	2017, Devnani B et al., [6]	20 years/M	Bladder	8.6	CK, EMA, vimentin, synaptophysin, MIC-2 (CD 99)	INI1, SMA, myogenin, GFAP, Bcl-2, CD 34, brachyury	+	Alive 2 yrs after diagnosis
7.	2015, Sterling ME et al., [7]	14 years/M	Bladder wall	5.5	NA	INI1	-	Died of disease after 3 months from partial cystectomy
8.	2014, Warren KS et al., [8]	17 years/F	Bladder	5	Pan CK, CD34, WT1, CD56, vimentin	INI1	+	Alive with no recurrence post treatment
9.	2012, Savage N et al., [9]	3 years/M	Bladder (anterior wall)	2.3	LMWK, EMA, CD99	Pankeratin, EBV, desmin, myogenin, S-100, WT1, CD31, CD34, CD45/LCA, CD43, CD20, CD3, CD5, CD30, ALK1, TdT, MPO, CD68, CD4, CD8	+	Alive 6 months after diagnosis
10.	2012, Tam HKY et al., [10]	5 months/F	Bladder (dome)	2	NA	INI1	-	Alive 1 month after diagnosis
11.	2011, Eaton KW et al., [11]	5 months/F	Bladder	NA	NA	NA	+	Died of disease
12.	2008, Bourdeaut F et al., [12]	6 months/F	Bladder	NA	NA	INI1	+	Died of disease
13.	2004, Chang JH et al., [13]	4 years/F	Bladder (anterior/ superior wall)	5	Vimentin	SMA, desmin, myoglobin	-	Alive 9 year after diagnosis
14.	2001, Duvdevani M et al., [14]	4 years/F	Bladder (dome)	2.4	Vimentin, SMA, EMA	Desmin, myoglobin	-	Alive 2 year after diagnosis
15.	1989, Carter RL et al., [15]	6 years/F	Bladder (posterior wall)	NA	Vimentin, EMA	Desmin, myoglobin	-	-
16.	1987, Harris M et al., [16]	46 years/F	Bladder	10-15	Vimentin, CK, NSE and EMA	α1- antitrypsin, α1-antichymotrypsin, myoglobin, desmin	-	Died of disease
17.	Present case	15 years/F	Bladder (right lateral wall)	8.8	EMA, vimentin, Pan-CK, BRG1	INI1, LCA, CD20, CD3, MPO, CD34, myogenin, desmin, GATA3, uroplakin III, SALL4, MyoD1, SOX10	-	Lost to follow-up

[Table/Fig-4]: List of 16 cases of PMRTs of the urinary bladder reported in literature [1-16].
F: Female; M: Male; IHC: Immunohistochemistry

Despite advances in diagnosis and treatment, the prognosis for PMRT of the bladder remains guarded to poor, reflecting the inherent biological aggressiveness of rhabdoid tumours. Historical data on extrarenal malignant rhabdoid tumours show low overall survival rates, often quoted at around 20% at five years, and many patients experience rapid progression even after surgical resection and adjuvant therapy. There are documented cases of rapid systemic dissemination shortly after surgery despite initial absence of metastasis, illustrating the aggressive nature of these tumours and the challenge in achieving durable control. Nonetheless, emerging evidence suggests that younger children treated with high-dose, multimodal therapy may achieve longer disease-free intervals, underscoring the importance of early diagnosis, comprehensive staging, and tailored treatment planning [1,3,7,9,15-17].

CONCLUSION(S)

A positive vimentin and loss of INI1 (SMARCB1) is diagnostic of rhabdoid tumour. Differential diagnosis of PMRT includes rhabdoid variant of urothelial carcinoma, plasmacytoid variant of urothelial carcinoma, rhabdomyosarcoma, and haematolymphoid malignancies. Loss of INI1 establishes the diagnosis. Although rare in adolescents and rare in location, PMRTs of the urinary bladder should not be overlooked and an early, accurate diagnosis is warranted in view of its aggressive nature. The unusual features of this case include an older age of presentation, the large size of tumour and an unusual location.

REFERENCES

[1] Li J, Bao J, Hu T, Zhao Q, Zhou J, Zhou L, et al. Successful treatment of a 4-year-old girl with pure malignant rhabdoid tumour of the bladder: A case report. Transl Pediatr. 2024;13(10):1879-84. Doi: 10.21037/tp-24-255.

[2] Huang HC, Huang SC, Ming YC, Lai JY, Wang YL, Jaing TH. Malignant rhabdoid tumour of the urinary bladder in a 1-year and 9-month-old girl: A case report and literature review. Pediatr Neonatol. 2024;65(6):597-98. Doi: 10.1016/j.pedneo.2023.12.012.

[3] Tang V, Conner PM, Tovar JP, Gandour-Edwards RF, Antony R, Bobinski M, et al. Congenital presentation of synchronous atypical teratoid rhabdoid tumour and malignant rhabdoid tumour of the urinary bladder in a term infant. Autops Case Rep. 2020;10(4):e2020205. Doi: 10.4322/acr.2020.205.

[4] Hecht SL, Walker JP, Treece AL, Cost NG. Isolated pure malignant rhabdoid tumour (MRT) of the bladder: Case report and lessons learned. Urology. 2020;137:164-67. Doi: 10.1016/j.urology.2019.10.023.

[5] Hoare DT, Kiddoo D, Stryker T. An antenatally detected pure malignant rhabdoid tumour of the bladder. Urology. 2019;123:221-23. Doi:10.1016/j.urology.2018.10.019.

[6] Devnani B, Biswas A, Bakhshi S, Kaushal S, Nakra T. Extrarenal extracranial rhabdoid tumour of the pelvis in a young adult – management of a challenging case. Indian J Med Paediatr Oncol. 2017;38(3):383-86. Doi: 10.4103/ijmpo.ijmpo_108_17.

[7] Sterling ME, Long CJ, Bosse KR, Bagatell R, Shukla AR. A rapid progression of disease after surgical excision of a malignant rhabdoid tumour of the bladder. Urology. 2015;85(3):664-66. Doi: 10.1016/j.urology.2014.11.011.

[8] Warren KS, Oxley J, Koupparis A. Pure malignant rhabdoid tumour of the bladder. Can Urol Assoc J. 2014;8(3-4):E260-E262. Doi: 10.5489/cuaj.1633.

[9] Tam HKY, Li A, Lee PSF, Tse VPW, Pang KKY. Infantile intravesical malignant rhabdoid tumour. Hong Kong J Radiol. 2013;16(3):e21-e25. Doi: 10.12809/hkjr1312130.

[10] Savage N, Linn D, McDonough C, Donohoe JM, Franco A, Reuter V, et al. Molecularly confirmed primary malignant rhabdoid tumour of the urinary bladder: Implications of accurate diagnosis. Ann Diagn Pathol. 2012;16(6):504-07. Doi: 10.1016/j.anndiagpath.2011.04.008.

[11] Eaton KW, Tooke LS, Wainwright LM, Judkins AR, Biegel JA. Spectrum of SMARCB1/INI1 mutations in familial and sporadic rhabdoid tumours. Pediatr Blood Cancer. 2011;56(1):07-15. Doi: 10.1002/pbc.22831.

[12] Bourdeaut F, Fréneaux P, Thuille B, Bergeron C, Laurence V, Brugières L, et al. Extra-renal non-cerebral rhabdoid tumours. Pediatr Blood Cancer. 2008;51(3):363-68. Doi: 10.1002/pbc.21632.

[13] Chang JH, Dikranián AH, Johnston WH, Storch SK, Hurwitz RS. Malignant extrarenal rhabdoid tumour of the bladder: 9-year survival after chemotherapy and partial cystectomy. J Urol. 2004;171(2):820-21. Doi: 10.1097/01.ju.0000107014.40195.cf.

[14] Duvdevani M, Nass D, Neumann Y, Leibovitch I, Ramon J, Mor Y. Pure rhabdoid tumour of the bladder. J Urol. 2001;166(6):2337. Doi: 10.1016/S0022-5347(05)65582-6.
[15] Carter RL, McCarthy KP, al-Sam SZ, Monaghan P, Agrawal M, McElwain TJ. Malignant rhabdoid tumour of the bladder with immunohistochemical and ultrastructural evidence suggesting histiocytic origin. Histopathology. 1989;14(2):179-90. Doi: 10.1111/j.1365-2559.1989.tb02127.x.
[16] Harris M, Eyden BP, Joglekar VM. Rhabdoid tumour of the bladder: A histological, ultrastructural and immunohistochemical study. Histopathology. 1987;11(10):1083-92. Doi: 10.1111/j.1365-2559.1987.tb01847.x.
[17] Zangari A, Zaini J, Gulia C. Genetics of bladder malignant tumours in childhood. Curr Genomics. 2016;17(1):14-32.

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