BLADDER EMBRYONAL RHABDOMYOSARCOMA IN AN 18-MONTHS-OLD BOY: A CASE REPORT

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ABSTRACT

Objective: *This study aims to report a case of bladder embryonal rhabdomyosarcoma in an 18-months-old boy.* ***Case(s) Presentation:*** *An 18-months-old boy presented with complaint of difficult urinating since the last 2 months before presentation. It was accompanied with hematuria, blood clot and palpable mass in the lower abdomen. Magnetic resonance imaging (MRI) of lower abdomen showed a solid mass in the inferior bladder wall, involved bilateral ureterovesical junction (UVJ) that caused bilateral severe hydroureteronephrosis, lymphadenopathy, and multiple nodul in both lungs. Cystoscopy was performed and found intrabladder tumor near the bladder neck and trigonum. Then biopsy of the bladder tumor was performed. Pathology result of the specimens showed bladder embryonal rhabdomyosarcoma. Then immunohistochemistry examination was performed with desmin and myogenin antibody to confirm bladder embryonal rhabdomyosarcoma diagnosis.* ***Discussion:*** *Bladder embryonal rhabdomyosarcoma often involves the bladder neck and trigone. In general, clinical symptoms relate to mass and blockage caused by mass. Ultrasound is the first imaging examination performed. Magnetic resonance imaging (MRI) helps depict the mass, usually hyperintense on the T2 image. The immunohistochemical examination is an important examination of skeletal muscle markers. Desmin found in muscle is used to diagnose rhabdomyosarcoma.* ***Conclusion:*** *Bladder embryonal rhabdomyosarcoma is a diagnostic challenge as it is a rare malignant tumor. The clinician needs to be aware of this entity and performed immunohistochemistry as an important examination to accurately diagnose embryonal rhabdomyosarcoma of the bladder.*

Keywords: *Bladder, immunohistochemistry, rhabdomyosarcoma.*

ABSTRAK

Tujuan: *Penelitian ini bertujuan untuk melaporkan kasus rhabdomyosarcoma embrional kandung kemih pada anak laki-laki usia 18 bulan.* ***Presentasi Kasus:*** *Seorang anak laki-laki berusia 18 bulan datang dengan keluhan sulit buang air kecil sejak 2 bulan terakhir sebelum datang. Disertai dengan hematuria, bekuan darah dan teraba massa di perut bagian bawah. Magnetic resonance imaging (MRI) abdomen bagian bawah menunjukkan massa padat di inferior dinding kandung kemih, melibatkan bilateral ureterovesical junction (UVJ) yang menyebabkan hidroureteronefrosis berat bilateral, limfadenopati, dan nodul multiple di kedua paru. Dilakukan sistoskopi dan ditemukan tumor intrabladder di dekat kandung kemih dan trigonum. Kemudian dilakukan biopsi tumor kandung kemih. Hasil patologi spesimen menunjukkan rhabdomyosarcoma embrional kandung kemih. Kemudian dilakukan pemeriksaan imunohistokimia dengan desmin dan antibodi miogenin untuk memastikan diagnosis rhabdomyosarcoma embrional kandung kemih.* ***Diskusi:*** *Rhabdomyosarcoma embrional kandung kemih sering melibatkan leher kandung kemih dan trigonum. Secara umum, gejala klinis berhubungan dengan massa dan penyumbatan yang disebabkan oleh massa. Ultrasonografi adalah pemeriksaan gambar pertama yang dilakukan. Magnetic resonance imaging (MRI) membantu menggambarkan massa, biasanya hiperintens pada gambar T2. Pemeriksaan imunostokimia merupakan pemeriksaan penting dari penanda otot rangka. Desmin ditemukan di otot digunakan untuk mendiagnosis rhabdomyosarcoma.* ***Simpulan:*** *Rhabdomyosarcoma embrional kandung kemih merupakan tantangan diagnostik karena merupakan tumor ganas yang langka. Dokter perlu menyadari entitas ini dan melakukan imunohistokimia sebagai pemeriksaan penting untuk mendiagnosis rhabdomyosarcoma embrional kandung kemih secara akurat.*

Kata Kunci: *Kandung Kemih, imunohistokimia, rhabdomyosarcoma.*

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INTRODUCTION

Rhabdomyosarcoma is a malignant, progressive solid tumor originating from the mesenchymal tissue. The most common sites are head and neck, followed by the genitourinary tract.[1] The incidence of genitourinary rhabdomyosarcoma ranges from 0.5 to 0.7 cases per million children under 15 years of age. Tumors are often large and involve adjacent organs and blood vessels.[2] Bladder rhabdomyosarcoma most commonly occurs in children under 10 years of age and the median age of children with bladder rhabdomyosarcoma is 5 years. The male to female ratio is (1.4: 1) and is more common in Caucasians than African-Americans (2.3: 1).[3] The clinician must know the histological diagnosis in order to establish the most appropriate therapeutic regimen for embryonal rhabdomyosarcoma of the bladder.

CASE PRESENTATION

An 18 month old boy presented with complaints of difficult to urinate since the last 2 months before presentation. It was associated with hematuria and blood clots. Body weight had also decreased by about 1 kg over the last 2 months before presentation. His family history of tumors was denied.

The patient's vital signs were within normal limits. Physical examination revealed anemic conjunctiva and palpable mass in the lower abdomen. Patient had history of circumcision and insertion of 10fr silicone catheter. Urine output was 1200 cc / 4 hours, hematuria. There was no enlargement of the inguinal lymph nodes.

Laboratory findings indicated moderate anemia with increased ureum and creatinine. Ultrasound of the abdomen (Figure 1) showed a mass inside the bladder, in the bladder neck projection until trigone, suspiciously malignant, causing bilateral severe hydronephrosis and multiple lymphadenopathy of the right parailiac with malignant characteristics.

On the AP/lateral chest X-ray (Figure 2), there were multiple infiltrates and nodules in both lungs, which was suspected as a metastatic process.

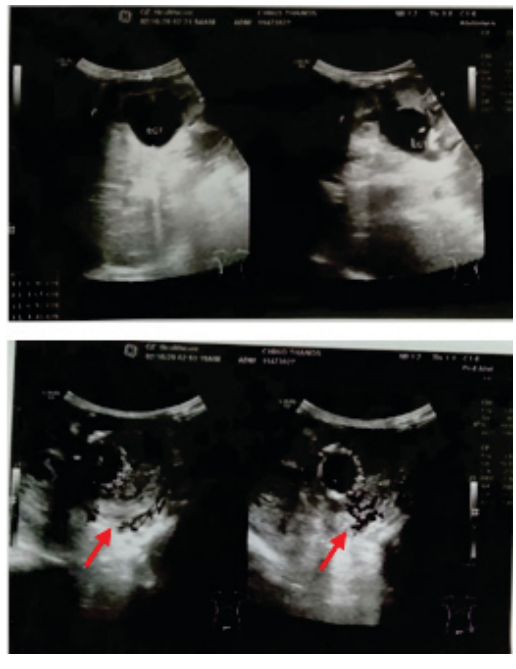

Figure 1. Ultrasound of the abdomen showed a mass inside the bladder, in the bladder neck projection–trigone, suspiciously malignant causing bilateral severe hydronephrosis, and multiple lymphadenopathy of the right parailiac with malignant characteristics.

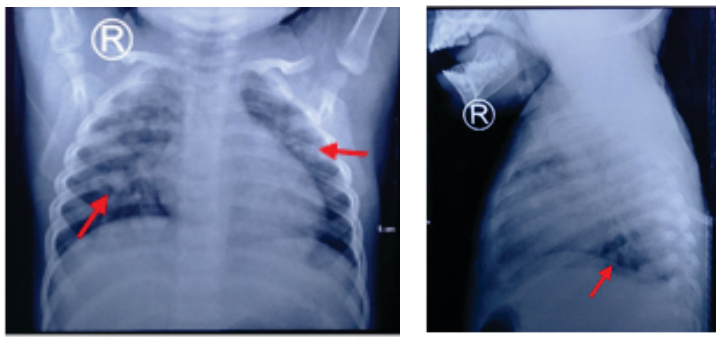

Figure 2. AP / lateral chest X-ray showed multiple infiltrates and nodules in both lungs.

X-ray of left femur AP / lateral (Figure 3) showed destructive osteolytic lesion on left femur, suspected as a metastatic process.

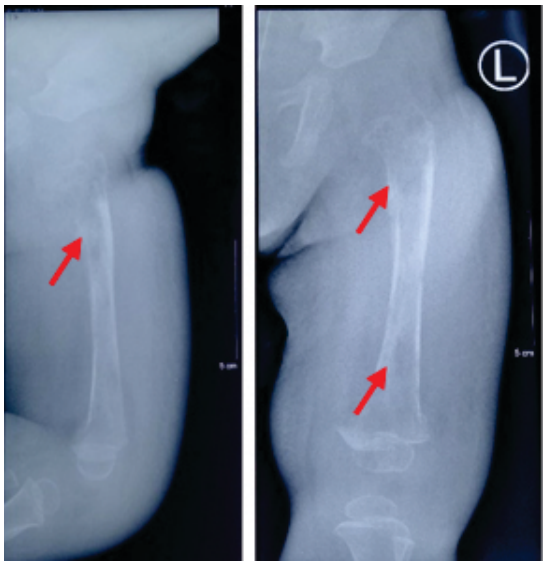

Figure 3. AP / Lateral left femur X-ray showed destructive osteolytic lesion on left femur.

Magnetic resonance imaging (MRI) of abdomen (Figure 4) showed a solid mass inferior to the bladder (2.6 cm) involving a bilateral ureterovesical junction (UVJ) with malignant characteristics leading to bilateral severe hydroureteronephrosis. In addition, there was right parailiac-paraaortic lymphadenopathy. Multiple nodules were also found in both lungs, which was suspected as a metastasis.

Cystoscopy (Figure 5) was performed and showed a mass in the bladder pressing on the superior and posterior side of the bladder wall. The outlet of right and left ureters were difficult to evaluate.

Based on the results of history taking, physical examination and radiological diagnostics, patient was diagnosed with a primary diagnosis of suspected malignant bladder tumor cT2N2M1b and complication diagnosis of bilateral severe hydroureteronephrosis. Patient underwent bladder biopsy and the specimen biopsy was sent to the anatomical pathology department. Pathology results (Figure 6) showed embryonal rhabdomyosarcoma of bladder, a botryoid subtype.

Further examination with immuno-histochemistry (Figure 7) was performed to confirm

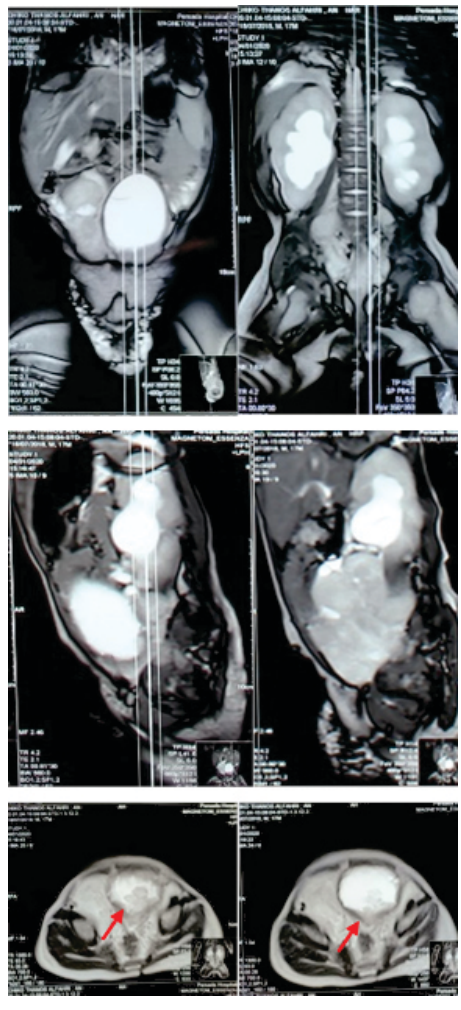

Figure 4. Magnetic resonance imaging (MRI) of the abdomen showed solid masses inferior to the bladder wall, bilateral severe hydro-ureteronephrosis, lymphadenopathy, as well as multiple nodules in both lungs.

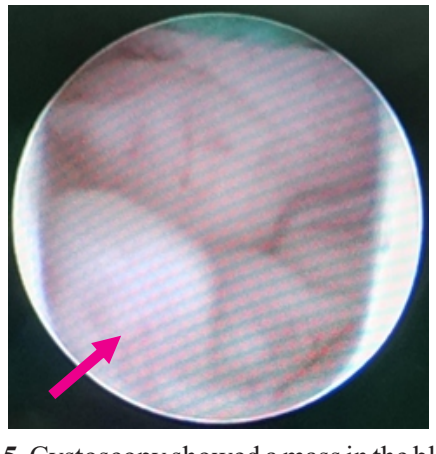

Figure 5. Cystoscopy showed a mass in the bladder.

the diagnosis, using desmin and myogenin antibodies. From the results, desmin was found to be positive in the tumor cell cytoplasm (Figure 7a), and myogenin was found to be positive in tumor cell nucleus (Figure 7b). The immunophenotype was consistent with embryonal rhabdomyosarcoma.

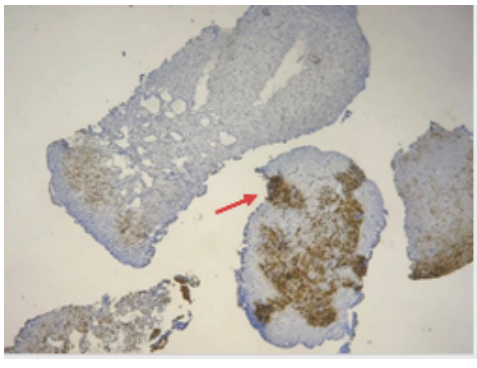

Figure 6. Pathology results at 40x magnification, showed embryonal rhabdomyosarcoma of bladder, a botryoid subtype.

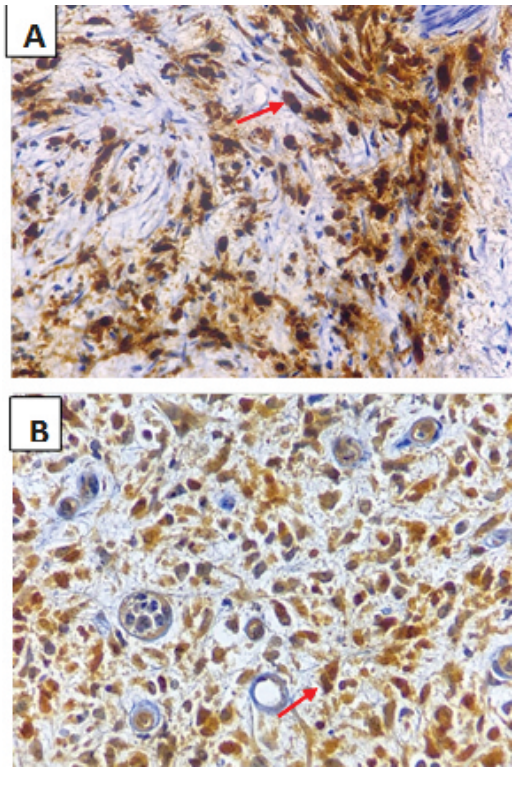

Figure 7 a. Desmin was found to be positive in the cytoplasm of tumor cells (magnification 400x); b. Myogenin was found to be positive in tumor cell nuclei (400x magnification).

DISCUSSION

Rhabdomyosarcoma (RMS) is a rare, progressive malignant solid tumor that occurs in approximately 5% of all soft tissue carcinoma cases. About 10-15% of all cases of rhabdomyosarcoma arise from the pelvic organs. Tumors are often large and involve adjacent organs and blood vessels.[3]

Rhabdomyosarcoma of the genitourinary tract is a rare tumor almost exclusive to the pediatric population. Cases that occur in the bladder and prostate account for 50% of genitourinary sarcomas. Rhabdomyosarcoma usually arises from the prostate or trigone of the bladder in boys, and from the vagina or uterus in girls. Nearly 75% of cases of genitourinary rhabdomyosarcoma (RMS) are diagnosed before the age of 5 years, with a male to female ratio of 3.3:1. Rhabdomyosarcoma of the bladder generally occurs on the posterior wall and often involves the bladder neck and trigone.[4-5]

In the 2020 WHO classification, rhabdomyosarcoma (RMS) has been subdivided into few types, including embryonal, alveolar, pleomorphic, and spindle cell/sclerosing RMS.[6] Bladder rhabdomyosarcoma is usually embryonal. Embryonal cell types represent more than half of all histological subtypes.[7]

In embryonal rhabdomyosarcoma (ERMS), the clinical symptoms vary. In general, clinical symptoms relate to the mass and blockage caused by the mass. Symptoms are caused by displacement or blockage of normal structures. Children with rhabdomyosarcoma (RMS) bladder are usually under 4 years of age and present with hematuria and urinary tract obstruction. In general, the tumor is more than 5 cm in size and is invasive. Patients usually present with urinary symptoms (hematuria, dysuria and urinary retention).[2] In this case report, patient presented with complaint of difficult to urinate. Patient also had a history of hematuria, often with blood clots.

Ultrasound is the first imaging examination performed and usually shows an intraluminal polypoid mass with solid and cystic elements and variable internal flow.[4,8] In this case report, the abdominal ultrasound showed mass on the bladder neck projection until bladder trigone causing bilateral severe hydronephrosis and multiple lymphadenopathy of the right parailiac with malignant characteristics.

Rhabdomyosarcoma (RMS) bladder in children tends to remain localized. Metastasis to

regional lymph nodes and to distant sites is a relatively late occurrence. Metastases occur most often in the lymph nodes, lungs and cortical bones, but also in the liver and bone marrow.[9]

Magnetic resonance imaging (MRI) helps depict the mass, usually hyperintense on the T2 image because of the histological pattern of the myxoid.[5,10] In this case, MRI of the abdomen showed asolid mass inferior to the bladder (2.6 cm) involing bilateral ureterovesical junction (UVJ) with malignant characteristics, causing bilateral severe hydroureteronephrosis. There were also right parailiac-paraaortic lymphadenopathy, and several nodules in both lungs, suspicious of metastasis.

These tumors are often found in mucous-lined hollow visceral organs such as bladder, with preference for the regions of bladder neck and trigone. Tumors show intra-luminal polypoid growth. Macroscopically the mass consists of a fine, grape-like cluster of several soft polypoid gray nodules with a translucent and gelatinous appearance. Microscopically the tumor has a histological myxoid pattern and a submucosal zone characteristic of increased cellularity.[6,11]

Immunistochemical examination of embryonal rhabdomyosarcoma (ERMS) is an important examination of skeletal muscle markers. These markers are related to the degree of differentiation of the tumor. Strong stain markings on myogen antigens, especially on cells that show myoblast differentiation, as well as in desmin will give a positive result on embryonal rhabdo-myosarcoma (ERMS). Desmin found in muscle is used to diagnose rhabdomyosarcoma. Desmin is an intermediate filamentous protein found in smooth muscle cells, striated muscle cells, and myocardium.[9]

In this case report, immunohistochemistry of patient's tumor showed positive desmin in several cells, indicating that these cells contain smooth and striated muscle cells. Myogenin examination was positive in almost all cell nuclei. Myogenin is a primitive transcription factor that regulates desmin production. So it can be concluded that the results of immunistochemical examinations support the diagnosis of embryonal rhabdomyosarcoma of the bladder.

Patient was managed with VAC chemo-therapy regimen, which consist of vincristine, actinomycin D, and cyclophosphamide. Prognosis of the tumor is difficult to conclude as very few cases have been reported. However, in patients with metastatic disease, little progress has been made in survival rates, with a 5-year, event-free survival rate of less than 30%.[12] Hence, the patient must be kept on follow-up.

CONCLUSION

Bladder embryonal rhabdomyosarcoma is a diagnostic challenge as it is a rare malignant tumor. Clinician needs to be aware of this entity and performed immunohistochemistry as an important examination to accurately diagnose embryonal rhabdomyosarcoma of the bladder.

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