



An Antenatally Detected Pure Malignant Rhabdoid Tumor of the Bladder

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Pediatric malignant rhabdoid tumors are a rare entity with poor prognosis. The extrarenal variants of this disease are sparsely presented within the literature. Our case represents, to our knowledge, the first antenatally detected bladder variant. At 19 months of age, this patient has benefited from early detection and intervention and remains disease free following partial cystectomy and chemotherapy. UROLOGY 123: 221–223, 2019. © 2018 Elsevier Inc.

Pure malignant rhabdoid tumors of the bladder are exceedingly rare. We present the novel case of an antenatally detected bladder mass and the interventions that ensued.

CASE

This patient was born to a G2T1P0A0L1 healthy 34-year-old mother with an antenatally detected bladder mass. The mother had undergone both a first trimester dating ultrasound and second trimester anatomy scan, revealing no fetal abnormalities. At 37 weeks, 5 days, decreased fetal movements triggered a repeat ultrasound and biophysical profile. This scan indicated that the fetus, was symmetrically small for gestational age (5th–10th %) with a normal amniotic fluid volume. The biophysical profile was 8 out of 8. There was increased umbilical arterial resistance in keeping with placental insufficiency. Of note, an echogenic 1.6×1.8 cm mass within the urinary bladder was detected (Fig. 1). It was apparently avascular. Based on the findings of placental insufficiency and intrauterine growth retardation, the mother was induced and the patient was delivered on February 8th (gestational age 37 weeks, 6 days).

At birth, the patient's Apgar scores were 8 and 9 at 1 and 5 minutes respectively. Initial birth weight was 2570 g, head circumference was 32 cm, and length was 47 cm. There were no dysmorphic features and the patient's genitalia were in keeping with a normal male phenotype. A postnatal ultrasound confirmed the presence of a $1.6 \times 2.2 \times 3.1$ cm mass occupying most of the bladder. It was mildly vascular on Dopplers

and was associated with mild bilateral hydronephrosis. Creatinine was elevated on initial bloodwork to 55 $\mu\text{mol/L}$ and there were 6–10 RBCs/high-power field (HPF) on urinalysis.

At 6 days of age, we proceeded with cystoscopic evaluation and biopsy of the bladder tumor (Fig. 2). On direct visualization with a 9-French cystoscope, the mass was easily identifiable. It appeared free floating, originating from an apparently small pedicle, although we were unable to negotiate the mass to directly identify the stalk. Initial attempts with the electrocautery resectoscope to attain tissue were unsuccessful given an outer calcified layer. Laser resection was attempted. We were able to pierce the calcified layer with this modality, but simultaneously induced bleeding. Further resection and alligator forceps were successful in attaining sufficient tissue. Once again, we were unable to navigate to the stalk.

Pathology was sent on rush and culminated in diagnosis of a pure malignant rhabdoid tumor of the bladder. Microscopic results showed irregular fragments of soft tissue composed of spindle cells arranged in ill-defined fascicles. Focal cells clustered into nests with medium sized nuclei and inconspicuous nucleoli. No epithelium was seen. There were prominent mitotic figures. There was 1 small focus of loosely cohesive large cells with eccentric nuclei and inclusion-like nucleoli. Immunohistochemical staining demonstrated AE1/AE3 and vimentin strongly expressed by the epithelioid large cells. MIB-1 was expressed by stromal and epithelioid cells. INI-1 was expressed by stromal cells only and was absent from tumor cells.

A multidisciplinary conference was held and the decision to proceed with partial cystectomy was made. A pre-operative CT chest confirmed the absence of thoracic metastases. Given the interval increase in size between ultrasounds and the tumor's confinement to the bladder, surgery proceeded without neoadjuvant chemotherapy. Radiation was avoided due to the high morbidity and organ-confined disease state. Surgery was performed on day 14 of life. Prior to bladder resection, general surgery was asked to place a 4-French Broviac catheter to facilitate postoperative chemotherapy. Once completed, we

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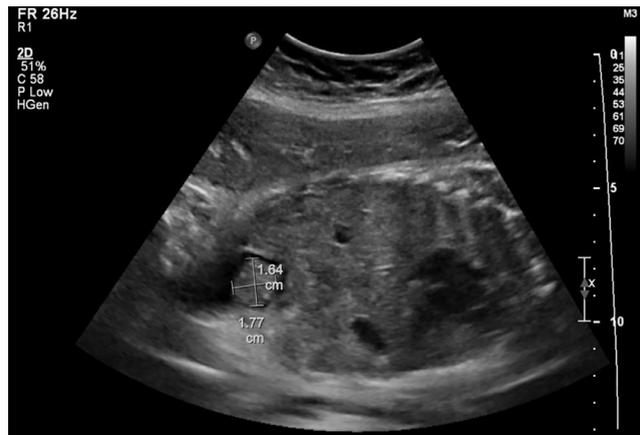


Figure 1. Antenatal ultrasound at 37 weeks, 5 days gestation demonstrating a 1.6 × 1.8 cm echogenic fetal bladder mass.



Figure 2. Cystoscopic visualization of the right and left extent of the intravesical bladder mass. (Color version available online).

proceeded with resection. Dissection to the bladder neck was completed. We placed our cystotomy anterior to the neck in a location we were confident there was no tumor as per our cystoscopic examination. We proceeded to incise superiorly and laterally to the right at which point we could easily identify the tumor (Fig. 3). A small stalk was found protruding from the left, superior aspect of the bladder. We resected around the stalk and removed the tumor en bloc. Repeat resection of the bladder edge was performed and sent for pathology as well. The bladder was

closed primarily and the field irrigated. There were no intraoperative complications.

Immediate postoperative CT chest/abdomen and magnetic resonance (MR) pan-scan confirmed an absence of metastases. In addition, an ultrasound 1 week post-op showed significantly improved bilateral hydronephrosis which had completely resolved by week 2. Final surgical pathology was once again consistent with a rhabdoid tumor. Microscopic tumor extension into the outer half of the deep muscularis propria was noted. Margins on the



Figure 3. Gross pathology before and after surgical resection of the bladder mass. (Color version available online).

primary section were uninvolved by tumor, although margin size was not adequately assessed. The margins for the additional resection of the bladder edge were also negative.

The baby began a patient-specific protocol for his confirmed rhabdoid malignancy. This consisted of alternating courses of vincristine, doxorubicin, cyclophosphamide (VDC) and ifosfamide, carboplatin, etoposide (ICE) chemotherapy as per the EPSSG NRSTS 2005 protocol. Following the initiation of this regime, a 3-month follow-up ultrasound and CT chest/abdomen/pelvis were attained. Outside of clear postoperative changes to the urinary bladder, no disease recurrence was noted. The patient, now post-op 18 months, has not demonstrated any signs of disease recurrence.

DISCUSSION

Once described as a rhabdomyosarcomatoid variant of Wilms, malignant rhabdoid tumors were reclassified in the 1980s as a distinct clinical and histopathologic entity.¹ Initially identified in the kidney, primary rhabdoid tumors have since been found to originate in a number of extra-renal locations. Regardless of site, however, 5-year overall survival rate is poor, with tumor stage and age of detection representing the only clear prognostic factors.²

Common extra-renal locations include the spinal cord and central nervous system.³ A number of distinct bladder tumor types with rhabdoid features exist, but as they are not pure lesions, they are rather characterized by their primary cell type.^{4,5} Pure isolated malignant rhabdoid tumors of the bladder are exceedingly rare, and unlike kidney primaries which occur in infants, these extrarenal malignant rhabdoid tumors often present in older children.⁶⁻⁹ Our case highlights what we believe to be the first antenatally detected malignant rhabdoid of the bladder. Given the potential rapid progression from localized to metastatic disease despite aggressive surgical and chemotherapeutic intervention, we are optimistic that our early detection will lead to a more favourable long-term prognosis. The antenatal identification permitted a focused, multidisci-

plinary approach including rapid partial cystectomy and adjuvant chemotherapy. The use of neoadjuvant treatment and the implementation of radiation therapy are all variables in the current approach to the bladder rhabdoid, and were not employed in our patient.

Genetic links for pure malignant rhabdoid tumors have been established, with deletion and/or mutation of the INI1 gene and subsequent inactivation of the SMARCB1 tumor suppressor gene, representing a common pathophysiology.³ The pathologic evaluation of our patient confirmed the absence of functional INI1, and genetic evaluation confirmed the absence of a germline mutation, suggesting a potential de novo mutation or epigenetic cause.

Pediatric malignant rhabdoid tumors are a rare entity with poor prognosis. The extra-renal variants of this disease are sparsely presented within the literature. Our case represents, to our knowledge, the first antenatally detected bladder-variant. At 19 months of age, this patient has benefited from early detection and intervention.

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