

Botryoid Rhabdomyosarcoma of the Vagina in a Three-Year-Old Girl: A Case Report and Literature Review

Benkaddour Rania*, Dhamnia El Mehdi, Yassine Kassab Alia, Allali Nazik, Chat Latifa and El Haddad Siham

Department of Radiology, Children Hospital, Ibn Sina University Hospital Center, Rabat, Morocco

***Corresponding Author:** Benkaddour Rania, Department of Radiology, Children Hospital, Ibn Sina University Hospital Center, Rabat, Morocco.

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Abstract

Botryoid rhabdomyosarcoma (BRMS) is a rare variant of embryonal rhabdomyosarcoma (eRMS) typically occurring in the genitourinary tract of young children. We report the case of a three-year-old girl presenting with intermittent vaginal bleeding and a “grape-like” protruding mass. Imaging (MRI) showed a localized vaginal tumor without distant metastasis. Diagnosis was confirmed by histopathology showing a characteristic cambium layer and positive immunohistochemical staining for desmin and myogenin. Management involved a multidisciplinary approach combining chemotherapy and conservative surgery. This report highlights the importance of early diagnosis, the role of MRI in monitoring, and the necessity of balancing oncological cure with organ preservation.

Keywords: Botryoid Rhabdomyosarcoma; Vaginal Tumor; Pediatric Oncology; WHO Classification; MRI

Introduction

Rhabdomyosarcoma (RMS) is the most common soft-tissue sarcoma in children. Embryonal rhabdomyosarcoma represents a major histological subtype and frequently arises in the head and neck or genitourinary tract [1,2]. According to the fifth edition of the World Health Organization Classification of Soft Tissue and Bone Tumours, botryoid rhabdomyosarcoma is considered a morphological variant of embryonal rhabdomyosarcoma developing beneath the mucosal surfaces of hollow organs [1].

Vaginal botryoid rhabdomyosarcoma is rare and primarily affects infants and young children. It typically presents with vaginal bleeding, blood-stained discharge, or a polypoid mass protruding through the vaginal introitus. Its characteristic multilobulated “grape-like” appearance results from the submucosal growth of the tumor [2,3]. Nevertheless, this presentation may initially be mistaken for benign conditions, such as vaginal polyps, urethral prolapse, or inflammatory lesions, potentially delaying diagnosis [3].

Magnetic resonance imaging (MRI) plays a central role in local staging by defining tumor size and extent and evaluating possible involvement of adjacent pelvic organs. It is also useful for treatment planning and follow-up while avoiding exposure to ionizing radiation [2,3]. Definitive diagnosis relies on histopathological examination, particularly the identification of a dense subepithelial cambium layer, supported by immunohistochemical expression of skeletal muscle differentiation markers such as desmin and myogenin [1,2].

The management of vaginal rhabdomyosarcoma has progressively shifted from radical surgery toward risk-adapted multimodal treatment combining chemotherapy with conservative surgery and selective radiotherapy. This approach aims to maintain favorable oncological outcomes while preserving pelvic organ function and reducing long-term morbidity [2,4].

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We report the case of a three-year-old girl with vaginal botryoid rhabdomyosarcoma, focusing on its clinical presentation, MRI findings, histopathological characteristics, therapeutic management, and radiological response.

Case Presentation

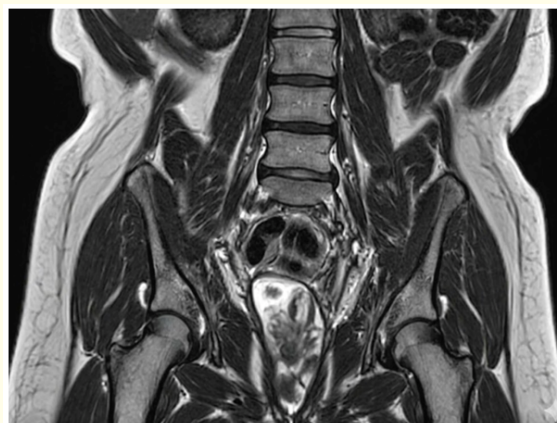
A three-year-old girl presented with a three-week history of intermittent vaginal bleeding and a visible mass. Physical examination revealed a soft, polypoid, 3 cm mass protruding from the vaginal introitus with a characteristic “grape-like” appearance (Figure 1).



Figure 1: Clinical appearance of the vaginal polypoid mass protruding from the vaginal introitus with a characteristic “grape like cluster” appearance.

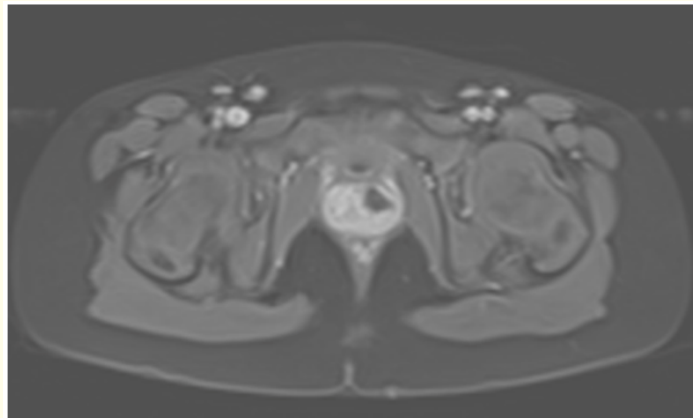
Diagnostic workup:

- **Imaging:** Pelvic MRI demonstrated a well-defined T2-hyperintense intravaginal mass with heterogeneous enhancement (Figure 2). No invasion of the bladder or rectum was noted.
- **Histopathology:** Biopsy showed a malignant mesenchymal proliferation with a dense subepithelial cambium layer. Tumor cells were small and spindle-shaped with rhabdomyoblastic differentiation.
- **Immunohistochemistry:** Strong nuclear positivity for myogenin and cytoplasmic staining for desmin confirmed the diagnosis (Figure 3 - Placeholder for IHC Image).

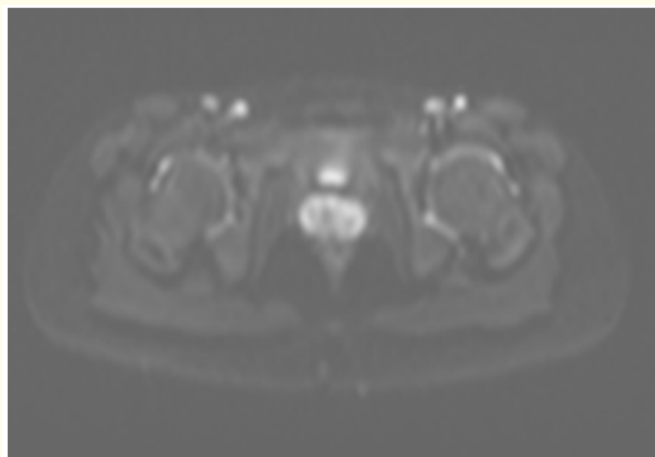




(B) Sagittal T2



(C) Post Gado



(D) Diffusion

Figure 2: Pelvic magnetic resonance imaging demonstrating the vaginal tumor at diagnosis.

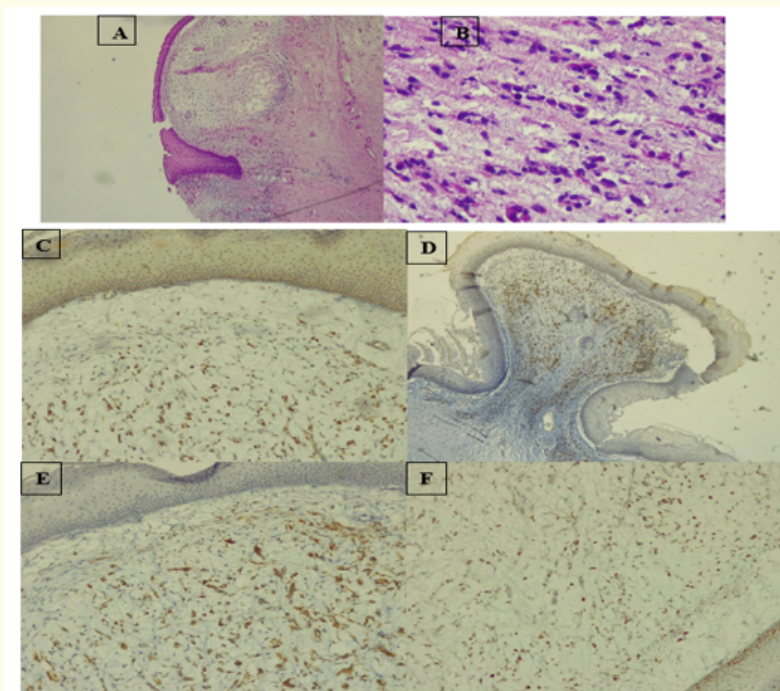


Figure 3: Histopathological and Immunohistochemical Features of the Vaginal Botryoid Rhabdomyosarcoma.

(A) Low-power histopathology (H&E, x25): Low-magnification view showing the pathognomonic dense subepithelial cambium layer epithelium, with a looser, myxoid tumor stroma deeper in the tissue.

(B) High-power histopathology (H&E, x400): High-magnification detailed view of the cambium layer. The neoplastic proliferation consists of small, spindle-to-oval-shaped mesenchymal cells with hyperchromatic, atypical nuclei and high nuclear-to-cytoplasmic ratios, with occasional rhabdomyoblasts showing eccentric eosinophilic cytoplasm.

(C) Immunohistochemistry for Desmin: Demonstrates strong, diffuse cytoplasmic immunoreactivity (brown staining) within the tumor cells, confirming their skeletal muscle origin.

(D) Immunohistochemistry for Myogenin: Confirms strong, diffuse nuclear positivity (brown staining) within the neoplastic cells, which is particularly dense in the subepithelial zone.

(E) Immunohistochemistry for MyoD1: Shows strong, specific nuclear staining of the neoplastic mesenchymal cells, further supporting rhabdomyoblastic differentiation.

(F) Immunohistochemistry for Ki-67: Displays a moderate proliferative index (approximately 30 - 40%), with nuclear positivity concentrated primarily within the active cambium layer.

Therapeutic regimen: The patient was treated according to pediatric oncology protocols with a chemotherapy regimen consisting of Vincristine, Actinomycin-D, and Cyclophosphamide (VAC). Following treatment, follow-up MRI showed a marked regression of the mass with no residual tumor (Figure 4).

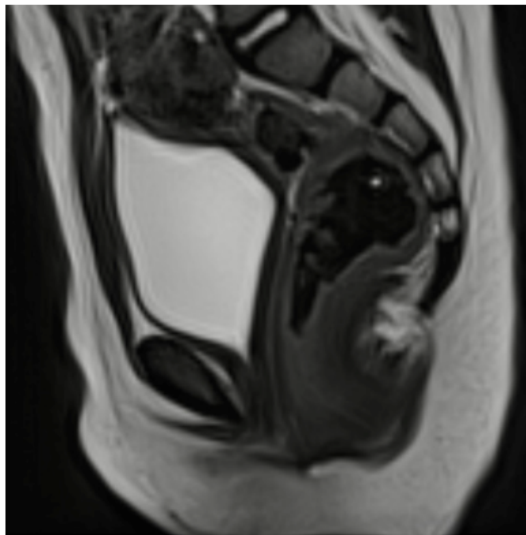


Figure 4: Follow-up pelvic MRI showing regression of the vaginal mass after treatment.

Discussion

Botryoid rhabdomyosarcoma (BRMS) is a rare clinicopathological variant of embryonal rhabdomyosarcoma that predominantly arises beneath the mucosal surfaces of hollow organs, particularly the vagina and urinary bladder in infants and young children. Although uncommon, it represents the most frequent malignant vaginal tumor in childhood and should always be considered in the differential diagnosis of vaginal bleeding associated with a protruding polypoid mass [1,2].

The characteristic “grape-like” appearance observed on clinical examination is highly suggestive of BRMS; however, this feature is not pathognomonic. Benign entities such as urethral prolapse, vaginal polyps, prolapsed ureterocele, and inflammatory lesions may present with similar findings, which may delay diagnosis. Therefore, prompt recognition and referral to a specialized pediatric oncology center are essential to optimize patient outcomes [2,3].

To place our observation into context, we reviewed previously published pediatric cases of vaginal botryoid rhabdomyosarcoma. As summarized in table 1, most patients presented during early childhood with vaginal bleeding and/or a protruding grape-like mass. Despite variations in tumor size and initial presentation, multimodal treatment based on chemotherapy followed by conservative surgery whenever feasible was associated with favorable oncological outcomes in the majority of reported cases.

MRI is the imaging modality of choice for local staging because of its excellent soft-tissue contrast and multiplanar capabilities. It accurately defines tumor size, assesses extension to adjacent pelvic organs, evaluates lymph node involvement, and guides therapeutic planning without exposing children to ionizing radiation. In our patient, MRI accurately demonstrated a localized vaginal lesion without bladder or rectal invasion and proved valuable for evaluating treatment response following chemotherapy [2].

Author	Age	Main symptom	MRI findings	Treatment	Outcome
Martelli., <i>et al.</i>	Infant	Vaginal bleeding	Localized vaginal mass	VAC + conservative surgery	Disease-free
Andrassy., <i>et al.</i>	2 years	Protruding mass	Vaginal lesion	Chemotherapy + conservative surgery	Disease-free
Walterhouse., <i>et al.</i>	3 years	Vaginal bleeding	No pelvic invasion	VAC	Complete response
Ferrari., <i>et al.</i>	4 years	Grape-like mass	Localized disease	Multimodal treatment	Favorable outcome
Present case	3 years	Vaginal bleeding + protruding grape-like mass	Localized vaginal mass without bladder or rectal invasion	VAC chemotherapy followed by conservative management	Marked radiological response

Table 1: Summary of published pediatric cases of vaginal botryoid rhabdomyosarcoma.

Histopathological examination remains the cornerstone of diagnosis. The presence of a subepithelial cambium layer together with diffuse immunohistochemical expression of desmin and myogenin confirms skeletal muscle differentiation and establishes the diagnosis. According to the latest WHO Classification of Soft Tissue and Bone Tumours, botryoid rhabdomyosarcoma is currently regarded as a morphological variant of embryonal rhabdomyosarcoma rather than a separate histological subtype [1,5].

The management of vaginal BRMS has changed considerably over recent decades. Historically, radical surgical procedures frequently resulted in significant urinary, gynecological, and reproductive sequelae. Current international protocols developed by the Children’s Oncology Group (COG) and the European paediatric Soft tissue sarcoma Study Group (EpSSG) advocate risk-adapted multimodal therapy combining systemic chemotherapy with conservative surgery whenever feasible. This strategy has substantially improved survival while preserving pelvic organ function and quality of life [2,6].

The VAC regimen (vincristine, actinomycin D, and cyclophosphamide) remains one of the standard first-line chemotherapy protocols and usually induces marked tumor regression, facilitating organ-sparing surgery. Radiotherapy is reserved for selected cases with residual disease or local recurrence in order to minimize long-term complications in growing children [6].

From a molecular perspective, embryonal and botryoid rhabdomyosarcomas differ from the alveolar subtype by the absence of recurrent PAX3–FOXO1 or PAX7–FOXO1 fusion genes. Although molecular analysis is not mandatory for diagnosis in every case, it has become increasingly important for subtype classification, prognostic stratification, and future targeted therapeutic approaches [5].

Long-term surveillance remains mandatory despite the excellent prognosis of localized disease. Clinical follow-up combined with serial MRI examinations allows early detection of local recurrence and monitoring of treatment-related complications. In addition to oncological surveillance, long-term assessment should include urinary, gynecological, reproductive, and psychosocial outcomes, particularly in very young children.

Our case illustrates the typical clinical presentation, characteristic MRI findings, and classical pathological features of vaginal botryoid rhabdomyosarcoma in a three-year-old girl. Early diagnosis, multidisciplinary management, and favorable response to chemotherapy allowed conservative treatment while preserving pelvic anatomy. This observation further supports the current trend toward organ-preserving strategies for localized vaginal BRMS.

Conclusion

Botryoid rhabdomyosarcoma of the vagina is a rare but highly curable pediatric malignancy when recognized early and managed within a multidisciplinary setting. Because its initial presentation may mimic benign gynecological conditions, clinicians should maintain a high index of suspicion when confronted with vaginal bleeding or a protruding grape-like mass in young girls.

MRI plays a pivotal role in local staging, treatment planning, and post-treatment surveillance, whereas histopathological examination with immunohistochemical confirmation remains essential for establishing the diagnosis. Current risk-adapted multimodal treatment protocols combining chemotherapy with conservative surgery have markedly improved survival while minimizing long-term functional morbidity.

This case highlights the importance of early diagnosis, multidisciplinary collaboration, and organ-preserving therapeutic strategies. It also reinforces the value of MRI in both the initial assessment and longitudinal follow-up of pediatric patients with vaginal botryoid rhabdomyosarcoma.

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