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Case Report

Vaginal botryoid rhabdomyosarcoma in an infant: A case report and review of the literature ☆

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ABSTRACT

Botryoid rhabdomyosarcoma is a rare and aggressive malignancy that primarily affects the female genital tract in children. It arises from embryonal rhabdomyoblasts. The vagina is the most common site, but it can also occur, although rarely, in the cervix or uterine fundus. We report the case of a 2-year-old girl who presented with a rapidly growing mass in the vulvar region. A pelvic MRI revealed a grape-like mass occupying the vaginal lumen, suggestive of botryoid rhabdomyosarcoma. Biopsy of the mass confirmed the diagnosis.

Introduction

Rhabdomyosarcoma is the most common soft tissue sarcoma in children, accounting 5% of all childhood malignancies [1]. It arises from primitive mesenchymal cells of the skeletal muscle lineage. The most common tumor sites are the head and neck region, followed by the genitourinary tract [2]. There are 3 major histologic types of rhabdomyosarcomas: embryonal, alveolar, and pleomorphic [3]. Botryoid rhabdomyosarcoma is a polypoid variant of embryonal RMS. It originates from embryonal rhabdomyoblasts and accounts for approximately 3% of all RMS cases [4]. Management of this tumor has been improved by combining 3 therapeutic modalities: surgery, chemotherapy, and radiotherapy, depending on the extent of the disease. We report a case of vaginal botryoid rhabdomyosarcoma in a 2-year-old girl.

Case report

A 2-year-old girl with no significant medical history presented to the pediatric oncology service with a rapidly growing mass in the vulvar region protruding through the vaginal orifice for 1 month. On physical examination, the patient was conscious,

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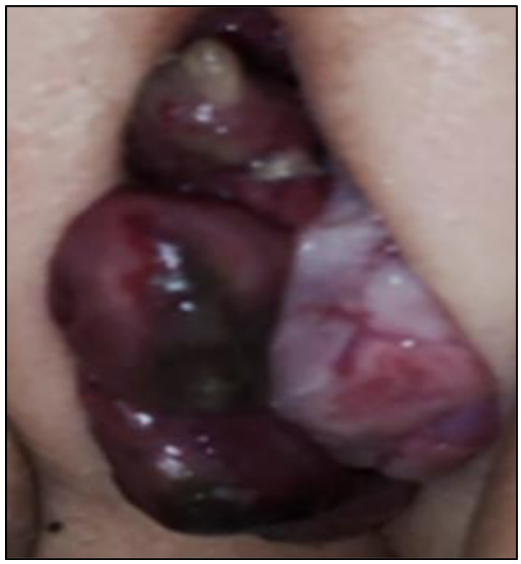

Fig. 1 – Pelvic examination revealed a red, grape-like mass the vulvar region, covering the entire vaginal introitus.

afebrile, and anicteric. Her weight and height were within the normal range for her age. Abdominal examination did not reveal any hepatosplenomegaly. Pelvic examination revealed a red, grape-like mass covering the entire vaginal introitus (Fig. 1) which bled on touch. Digital rectal examination identified a palpable mass pressing against the anterior wall of the rectum. No inguinal adenopathy was observed, and other systems were unremarkable.

Pelvic MRI was performed (Fig. 2) and revealed a large polypoid mass occupying the vaginal lumen and extending into the vulvar region. The mass measured 6.5 × 6.8 × 6.6 cm and appeared hypointense on T1-weighted sequences (Fig. 2A) and heterogeneously hyperintense on T2-weighted images (Fig. 2B and C). The mass also showed restricted diffusion (Fig. 2D) and significant enhancement after gadolinium administration (Fig. 2E and F). The uterus and ovaries had a normal appearance and there was no lymph node involvement.

The patient underwent a work-up for metastasis, including a thoracoabdominal CT scan and bone marrow biopsy, which did not reveal any metastatic lesions.

Based on the clinical presentation, the patient's age, and the pelvic MRI findings, the lesion was suggestive of the botryoid variant of embryonal rhabdomyosarcoma. A biopsy was performed under sedation. Histological and immunohistochemical studies confirmed the diagnosis (Figs. 3 and 4).

The patient underwent a work-up for metastasis, including a thoracoabdominal CT scan and bone marrow biopsy, which did not reveal any metastatic lesions.

The patient was classified as group C according to the European Pediatric Soft Tissue Sarcoma Study Group (EpSSG) RMS 2005 study. A multidisciplinary team recommended a treatment plan that included neoadjuvant chemotherapy, brachytherapy and surgery. Due to the unavailability of intravaginal brachytherapy for children in our country, the patient was referred for treatment abroad.

Discussion

Rhabdomyosarcoma is a high-grade malignant mesenchymal tumor that arises from embryonal muscle cells [5]. It predominantly affects children and adolescents, representing 5% of all childhood malignancies [1]. The median age at diagnosis is 5 years [6].

Histologically, RMS can be classified into 3 main types: embryonal, alveolar, and pleomorphic [3]. Embryonal rhabdomyosarcomas are further divided into 3 subtypes: botryoid, spindle cell, and anaplastic [7]. The botryoid variant is the most common variant of embryonal rhabdomyosarcoma and has a better prognosis. It commonly arises in the vagina, bladder, and biliary tract. It occurs almost exclusively in infants. The term botryoid refers to the characteristic grape-like appearance of the tumor [8].

The majority of RMS cases occur sporadically with no recognized predisposing factors. However, a small proportion of cases are associated with certain genetic conditions such as the Li-Fraumeni syndrome, which is caused by mutations in the TP53 gene [9].

Patients with botryoid ERMS typically present with a rapidly growing mass in the affected organ. In the case of vaginal involvement, symptoms may include a protruding mass, vaginal bleeding, and discharge. The mass is often described as polypoid and grape-like, as seen in our case. Other forms of presentation include urinary symptoms, especially when the tumor is located anteriorly, or tenesmus when there is posterior extension [10].

Despite its aggressive local behavior, metastasis in botryoid ERMS are relatively rare compared to other forms of rhabdomyosarcoma. When metastasis do occur, it usually involves the lymph nodes, lungs, cortical bones, liver, and bone marrow [12].

Ultrasonography is the first imaging investigation and typically shows a slightly hypoechoic, inhomogeneous, polypoid intravaginal mass with markedly increased internal flow.

Pelvic MRI is the preferred imaging modality [11], as it provides detailed information about tumor margin delineation, the extent of the tumor and its relationship with surrounding structures. The typical MRI appearance of botryoid RMS includes a polypoid mass that is hypointense on T1-weighted images and heterogeneously hyperintense on T2-weighted images. After gadolinium administration, the mass often shows significant enhancement, indicating its vascularity. MRI also aids in assessing the involvement of adjacent organs and the presence of lymph node metastases.

Biopsy and histological analysis are essential for confirming the diagnosis. Histological analysis reveals the presence of rhabdomyoblasts of varying differentiation dispersed within a loose, myxoid stroma [13]. The tumor cells are typically arranged in a cambium layer, just beneath the epithelial surface, which is a hallmark of the botryoid variant [14]. Immunohistochemical staining is also employed to detect specific markers, such as desmin, myogenin, and MyoD1, which help confirm the diagnosis by highlighting the muscle lineage of the tumor cells.

The treatment of botryoid ERMS involves a multidisciplinary approach, including surgical resection, chemotherapy,

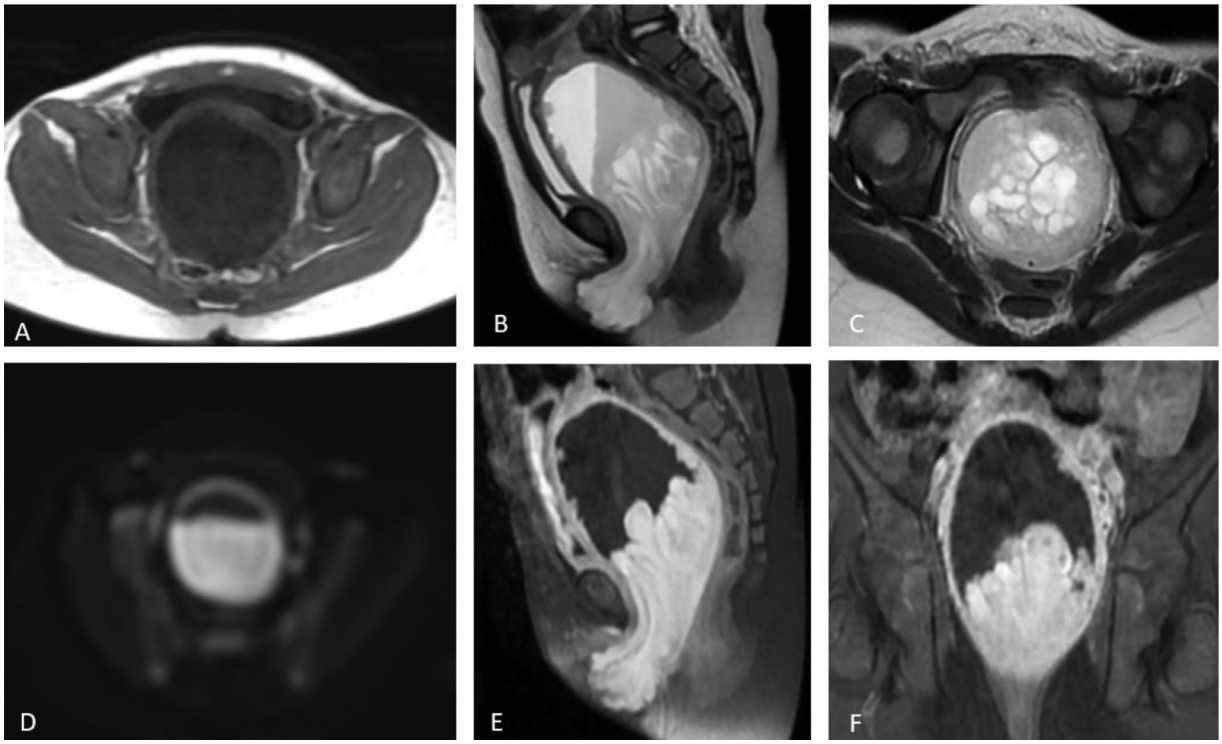

Fig. 2 – Pelvic MRI revealed a large polypoid mass occupying the vaginal lumen and extending into the vulvar region. It appeared hypointense on T1-weighted sequences (A) and heterogeneously hyperintense on T2-weighted images (B and C). The mass also showed restricted diffusion (D) and significant enhancement after gadolinium administration (E and F).

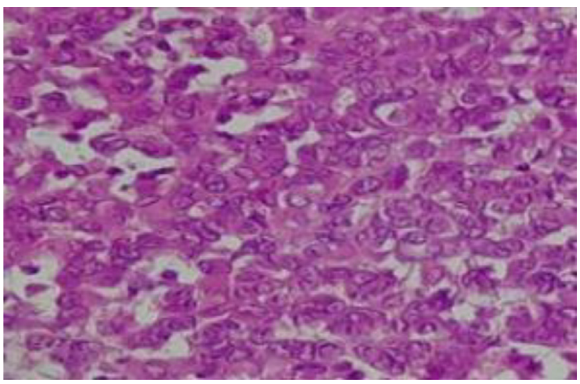

Fig. 3 – Histologic staining photomicrographs (H-E stain: original magnification x100) showing vaginal embryonal rhabdomyosarcoma, composed of various myogenic differentiated cells with myxoid stroma.

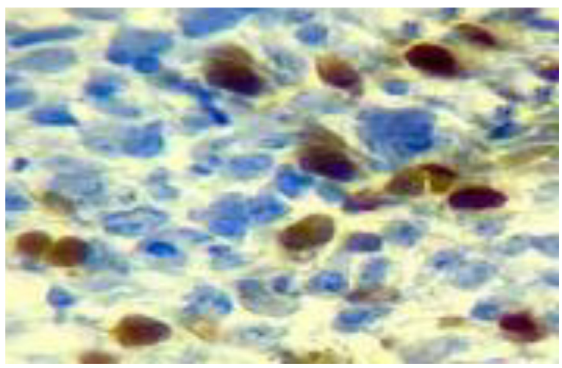

Fig. 4 – Immunohistochemistry (original magnification x400) shows positive myogenic antibody staining in tumor cells.

and radiotherapy. The treatment regimen is often based on guidelines from the European Paediatric Soft Tissue Sarcoma Study Group (EpSSG) RMS 2005 study [15], which provides evidence-based recommendations for the management of pediatric rhabdomyosarcoma. This comprehensive approach aims to maximize survival rates while minimizing long-term treatment-related morbidity.

The prognosis depends on the tumor's size, location, and extent of spread at the time of diagnosis. Overall, embryonal rhabdomyosarcoma has a more favorable prognosis compared to other subtypes. The botryoid variant, in particular, shows an even better prognosis [16].

Conclusion

The botryoid variant of embryonal rhabdomyosarcoma is a rapidly growing, rare malignancy that primarily affects children. This variant most commonly occurs in the genitourinary

tract. Early and accurate diagnosis, facilitated by advanced imaging techniques and histopathological analysis, is essential for effective management. A multidisciplinary treatment approach, incorporating surgical resection, chemotherapy, and radiotherapy, based on established protocols, significantly improves patient outcomes.

Patient consent

Written informed consent for the publication of this case report was obtained from the parents of the patient.

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