

Early Diagnosis of Embryonal Rhabdomyosarcoma: A Rare Case Confirmed by Vaginoscopy and Pathological Examination

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Case Report

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Abstract

Embryonal rhabdomyosarcoma (ERMS) is a rare, highly malignant neoplasm predominantly occurring in infants aged under 2 years. Its clinical manifestations are non-specific and easily misdiagnosed as benign lesions. This study reports a pediatric ERMS case confirmed via vaginoscopy and pathological examination. The patient presented with painless yellow watery vaginal discharge for 3 months, accompanied by intermittent gray-yellow tissue prolapsing from the vaginal introitus, which was initially misjudged as infectious vaginitis discharge. The diagnostic workflow of this case underscores the necessity of early identification of ERMS in children, as its atypical symptoms are frequently neglected. Combined vaginoscopy and immunohistochemistry enabled definitive pathological diagnosis, validating the clinical utility of such examinations for pediatric genitourinary malignancies. This case provides clinical reference for the diagnosis and standardized management of analogous pediatric cases, reminding clinicians to attach high vigilance to abnormal vaginal lesions in prepubertal girls. However, single-case limitation restricts the generalizability of the present findings, and the underlying pathogenic mechanisms of ERMS remain incompletely elucidated. Further investigations are required to elevate early diagnostic efficiency and therapeutic efficacy for improved long-term prognosis. This case report intends to inspire academic discussion and translational research to advance diagnostic and therapeutic strategies for ERMS.

Introduction

Rhabdomyosarcoma (RMS) ranks as the most prevalent soft tissue sarcoma among children and adolescents, responsible for over 50% of all pediatric soft tissue sarcoma cases. Based on the 2020 WHO Classification of Soft Tissue and Bone Tumours, RMS comprises four histological subtypes: embryonal RMS (ERMS, approximately 70% of paediatric cases), alveolar RMS (ARMS, approximately 20%), spindle cell/sclerosing RMS (SSRMS), and pleomorphic RMS (PRMS). PRMS predominantly occurs in adults, while SSRMS can arise in both children and adults. The botryoidal subtype represents a morphological variant of ERMS rather than an independent histological subtype [1]. ERMS predominantly affects infants and toddlers younger than 2 years, deriving from undifferentiated rhabdomyoblasts (primitive skeletal muscle progenitor cells). Clinically, ERMS manifests as a palpable soft tissue mass. Genital tract involvement in pediatric females frequently presents with abnormal vaginal discharge or protruding polypoid lesions[2]. Epidemiological evidence indicates that pediatric RMS remains an uncommon disease entity. Its vague clinical manifestations (local swelling, mild pain) frequently lead to missed diagnosis, highlighting the critical demand for heightened clinical vigilance to facilitate timely identification[3,4]. Affected prepubertal girls mostly complain of painless yellow vaginal discharge with gray-yellow polypoid lesions protruding from the vaginal introitus. Such manifestations mimic benign vulvovaginitis or vaginal foreign body irritation, creating diagnostic obstacles for clinicians[4]. Routine preliminary assessments including transabdominal B-ultrasound and vaginal smear tests fail to provide confirmatory diagnostic evidence. Vaginoscopy combined with pathological biopsy is thus mandatory to establish a definitive diagnosis[1]. Early detection of genital ERMS in young girls poses substantial

clinical challenges; clinicians should deepen their comprehension of this disease to elevate overall early diagnostic rates.

This case exhibits distinctive clinical value given its rarity and high risk of delayed diagnosis. We herein describe a botryoidal embryonal rhabdomyosarcoma lesion definitively identified via direct vaginoscopic visualization and subsequent pathological biopsy. Findings from this case provide practical clinical references for managing comparable pediatric patients, stressing the necessity of thorough evaluation for atypical vaginal lesions in children and the clinical priority of early diagnosis. This case report carries practical guiding implications for clinical practice and delivers novel insights for translational research on pediatric genitourinary sarcomas, meriting extensive academic discussion and further investigation.

Case Introduction

A 2-year-9-month-old female toddler was enrolled in the present study. Her caregivers reported a 3-month history of persistent yellow watery vaginal discharge without vaginal bleeding. Over the preceding month, gray-yellow soft tissue was intermittently seen prolapsing from the vaginal introitus during defecation, with spontaneous retraction afterward. The child received transabdominal ultrasonography and vaginal smear testing at a local primary hospital, and all preliminary examinations yielded unremarkable results. The patient was subsequently transferred to our department for definitive evaluation. Physical gynecological assessment demonstrated juvenile vulvar morphology, without cutaneous erythema or mucosal ulceration. Separation of the labia majora revealed an unremarkable vaginal introitus with no visible protruding lesions. Digital rectal palpation detected no paravaginal masses; digital compression on the vaginal orifice failed to induce tissue prolapse. Vaginoscopy was proposed to the guardians for further lesion visualization and targeted biopsy.

Diagnosis, Assessment, and Treatment Measures

After full explanation of the indications and potential perioperative risks of vaginoscopy, written informed consent was obtained from the patient's guardians. Standard preoperative preparation included 6 hours of fasting and 3 hours of water deprivation. Intravenous general anesthesia was induced with 1% propofol at an initial bolus dose of 2 mg/kg, followed by continuous maintenance infusion at 4–12 mg/kg/h. Vaginoscopy was performed using an Olympus OTV-S7 endoscope system (Japan) equipped with a 30° viewing lens and a 4.5-mm diagnostic outer sheath. Vaginal cavity distension was achieved with 0.9% normal saline under an insufflation pressure of 80 mmHg. Endoscopic visualization revealed multifocal clusters of gray-yellow polypoid cystic lesions distributed throughout the vaginal vault, extending to the anterior cervical lip (Fig. 1A). The diagnostic sheath was replaced with a 6.5-mm operative sheath; targeted resection of cervical polypoid lesions was completed via electrocautery and grasping forceps. Harvested specimens were fixed in neutral formalin and submitted for histopathological assessment. No intraoperative hemorrhage was noted at the biopsy site, and hymenal integrity was fully preserved without laceration or bleeding. The patient maintained stable vital signs under continuous sedation during the entire procedure. Spontaneous consciousness recovery occurred

roughly 30 minutes post-procedure, and 48-hour oral prophylactic antibiotics were prescribed after vaginoscopy.

Figure 1A. Endoscopic vaginoscopic manifestations of vaginal polypoid lesions. Figure 1B. Hematoxylin-eosin staining, ×200 magnification. Figure 1C. Positive MyoD1 immunohistochemical staining. Figure 1D. Ki-67 proliferation index (60% positive tumor cells)

Gross pathological observation identified two fragments of translucent cystic wall tissue (1.0–1.5 cm in maximum dimension) with gray-white to gray-yellow surface appearance. Histopathological microscopy demonstrated primitive neoplastic cells derived from cervical lesions, characterized by elongated spindle and oval cell morphology, with partial cytoplasmic eosinophilic staining. Integrating histological morphology and immunohistochemical profiles, a definitive diagnosis of botryoidal embryonal rhabdomyosarcoma was established. Immunohistochemical profiling revealed diffuse positive expression of Myoglobin, MyoD1, Myogenin, Actin, Desmin and Vimentin in tumor cells, whereas CD117, Fli-1, CD34 and TTF-1 showed complete negative staining. Ki-67 proliferation index reached 60% among neoplastic cells. The above biomarkers collectively validated the pathological diagnosis (Fig. 1B–D).

Discussion

Botryoidal embryonal rhabdomyosarcoma (ERMS) represents the predominant malignant vaginal neoplasm affecting prepubertal females. Despite its low overall incidence, the tumor's high biological aggressiveness and poor prognostic outcomes have aroused extensive clinical concern. Clinicians frequently encounter diagnostic obstacles when evaluating genital ERMS, as its heterogeneous, non-specific clinical manifestations overlap extensively with benign vaginal disorders. Typical clinical complaints encompass vaginal bleeding, protruding soft tissue masses and persistent abnormal vaginal discharge. Such overlapping symptoms commonly mimic benign vaginal lesions, inevitably resulting in missed diagnosis and delayed therapeutic intervention^[5, 6].

The novelty of the present case consists in the application of vaginoscopy for direct in vivo visualization of genital ERMS lesions; the final diagnosis was corroborated by endoscopic morphology and validated via immunohistochemical biomarkers. This finding underscores the synergistic diagnostic value of vaginoscopy and immunohistochemistry for pediatric genitourinary malignancies. Previous research has validated immunohistochemical profiling as an indispensable auxiliary diagnostic tool for ERMS, particularly for lesions with atypical cytological features^[7]. Notably, genetic molecular detection was not performed in this case. Existing genomic investigations of genital ERMS demonstrate a high prevalence of DICER1 pathogenic variants among cervical and vaginal ERMS patients, implying that germline genetic screening facilitates definitive diagnosis and familial tumor risk stratification^[8, 9]. Moreover, the perioperative workflow described herein verifies the safety and diagnostic efficacy of pediatric vaginoscopy for childhood gynecological malignancies, establishing a replicable clinical protocol for analogous cases. Standardized perioperative infection prophylaxis—including rigorous aseptic manipulation and timely postoperative antibiotic prophylaxis—was strictly executed in this patient,

confirming the essential role of infection prevention in postoperative pediatric care^[10]. Collectively, this case delivers actionable clinical guidance for frontline practitioners and reinforces the urgent demand for timely identification and precise pathological confirmation of genital ERMS. Distinguishing genital ERMS from benign vaginal pathologies poses persistent diagnostic difficulties for clinicians during initial assessment. Published cohort studies have documented substantial phenotypic overlap between ERMS and common benign vaginal lesions, which frequently leads to erroneous diagnosis and loss of optimal therapeutic windows^[11].

Vaginal discharge associated with genital ERMS is frequently misclassified as physiological secretion or simple infectious vaginitis, causing clinicians to neglect underlying malignant lesions. In addition, atypical histological features of ERMS may mimic benign mesenchymal vaginal proliferations on initial biopsy. Clinicians should therefore maintain a high index of suspicion for children presenting with unexplained persistent vaginal discharge to facilitate timely identification of occult malignant lesions^[12]. Comprehensive evaluation combining cross-sectional imaging and vaginoscopic biopsy is critical for suspected cases, as this multimodal diagnostic strategy elevates diagnostic specificity and lays a solid foundation for individualized therapeutic planning^[13]. Conventional diagnostic workflows for pediatric vaginal malignancies are hampered by non-specific clinical symptoms and ambiguous imaging manifestations, creating barriers to early lesion identification. Advances in vaginoscopic minimally invasive technology over recent decades have opened novel avenues for early screening and targeted interventional management of prepubertal genital ERMS. Vaginocopy enables real-time direct visualization of intra-vaginal lesions and performance of image-guided targeted biopsy, which significantly enhances diagnostic precision and minimizes false-negative diagnostic outcomes^[14]. Additionally, endoscopic minimally invasive resection achieves simultaneous hemostasis and partial lesion excision, effectively preserving pediatric reproductive anatomical structure and long-term quality of life^[14, 15].

The exact pathogenic cascade driving ERMS oncogenesis remains incompletely elucidated, yet accumulating preclinical evidence links tumor formation to dysregulated skeletal myogenic differentiation and somatic gene aberrations^[16, 17]. Immunohistochemical biomarker profiling yields high diagnostic sensitivity and specificity for ERMS, especially for atypical lesions with aberrant cytomorphology, serving as a definitive auxiliary diagnostic modality^[18, 19]. The present case achieved definitive diagnosis relying on combined vaginoscopic visualization and immunohistochemical biomarker detection. This dual diagnostic strategy verified favorable safety profiles and reliable diagnostic performance for childhood genitourinary sarcoma identification, underscoring the clinical imperative of timely and precise ERMS diagnosis. Further mechanistic research into ERMS tumorigenesis is anticipated to facilitate the development of individualized targeted therapeutics and optimize long-term survival outcomes for affected children.

Retrospective analysis of this patient's diagnostic and therapeutic workflow reinforces the critical clinical importance of heightened vigilance against genital ERMS among pediatric populations and the

paramount value of early definitive diagnosis. This case validates the superior clinical utility of integrating vaginoscopic exploration and histopathological verification for suspected vaginal ERMS. Consistent with previously published case series, the present patient presented with non-specific vaginal manifestations highly prone to misclassification as benign vulvovaginal disorders. Accordingly, clinicians must preserve a low diagnostic threshold for ERMS when evaluating prepubertal girls with unexplained persistent vaginal discharge. Timely vaginoscopy paired with standardized immunohistochemical testing can substantially elevate early diagnostic yield for genital ERMS and optimize long-term prognostic outcomes.

Several inherent limitations of this single-case report should be acknowledged despite its valuable clinical reference value. First, the sample size is restricted to one individual, without large-scale cohort data to validate broad clinical generalizability. Second, the complete molecular pathogenic cascade of ERMS remains unclarified; subsequent translational research is required to decipher core oncogenic pathways and formulate standardized targeted therapeutic regimens. Furthermore, ongoing investigations are warranted to establish optimized multimodal management protocols for genital ERMS, with the ultimate goal of prolonging overall survival and improving long-term life quality for pediatric patients. Collectively, this case report delivers practical clinical implications, establishes a baseline for subsequent translational research, and encourages extensive academic communication and collaborative exploration focusing on the diagnosis and treatment of pediatric genital ERMS.

Conclusion

This case report describes a 2-year-9-month-old prepubertal girl with botryoidal embryonal rhabdomyosarcoma (ERMS) who initially presented with 3 months of painless yellow watery vaginal discharge and intermittent gray-yellow tissue prolapsing from the vaginal introitus, a clinical picture that was easily misattributed to infectious vaginitis in primary care. Definitive diagnosis was successfully established via standardized vaginoscopic targeted biopsy combined with myogenic marker immunohistochemistry, with no intraoperative complications and full preservation of hymenal integrity. This case confirms that vaginoscopy is a safe, high-yield examination modality for evaluating atypical vaginal lesions in prepubertal children. Clinicians should maintain a high index of suspicion for unexplained persistent abnormal vaginal discharge in this patient population, to reduce the risk of delayed diagnosis of genital ERMS.

Abbreviations

ERMS Embryonal rhabdomyosarcoma

RMS Rhabdomyosarcoma

Declarations

Ethics approval and consent to participate

Not applicable

Consent for publication

All authors have read and approved the final manuscript. We have obtained written informed consent from the patient's legal guardian for the publication of relevant clinical details.

Availability of data and materials

Not applicable

Competing Interests

All authors declare no conflicts of interest

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Authors' contributions

Rongfang Zheng and Jiaxi Wang contributed equally to the conception and design of the study. Tiansheng Qin and Wei Fan supervised the research and critically reviewed the manuscript. Rongfang Zheng and Jiaxi Wang drafted the manuscript. Linjuan Qi collected data. All authors contributed to the revision of the manuscript and approved the final version for submission.

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Not applicable

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Figures

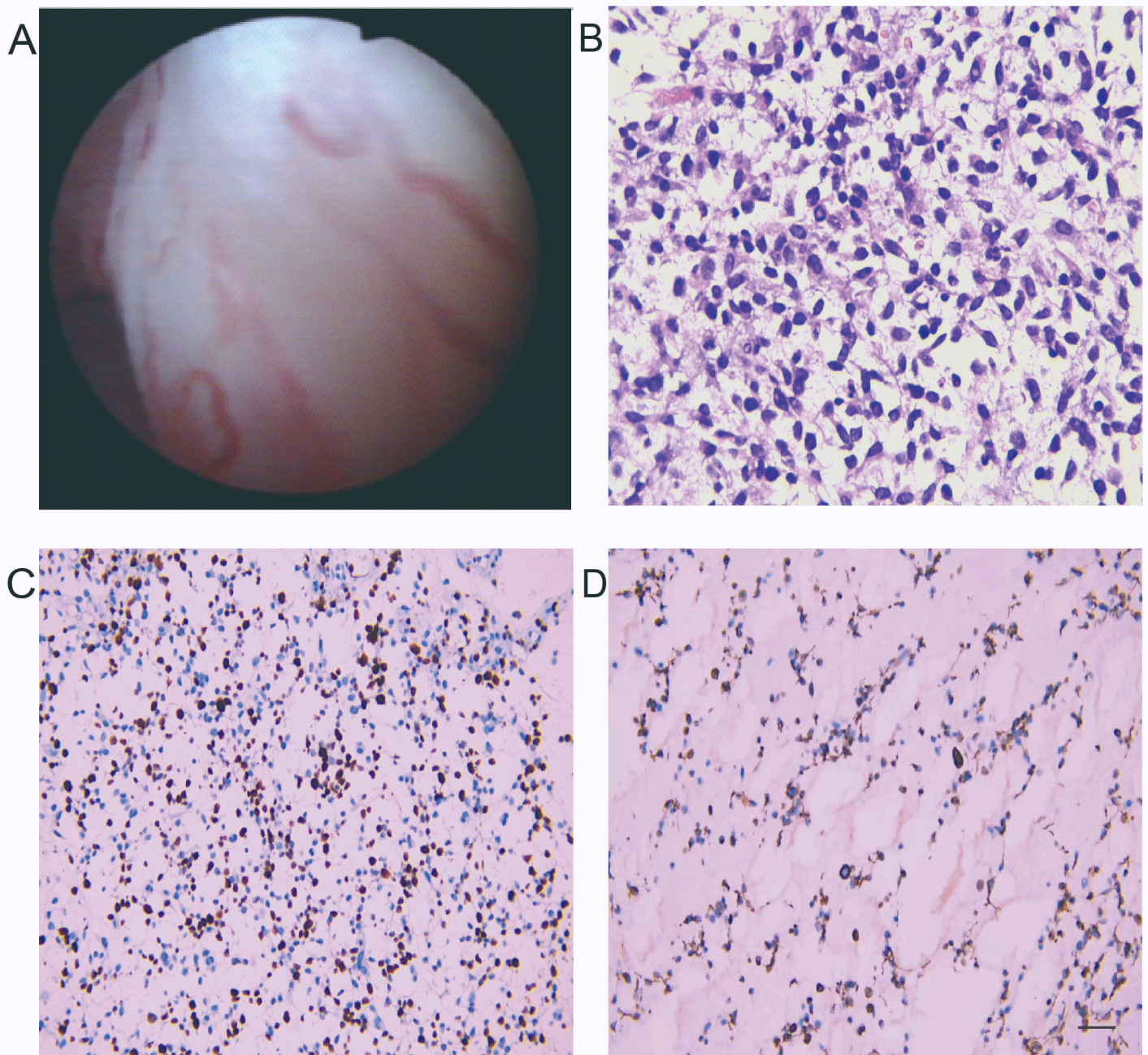


Figure 1

Figure 1A. Endoscopic vaginoscopic manifestations of vaginal polypoid lesions. Figure 1B. Hematoxylin-eosin staining, ×200 magnification. Figure 1C. Positive MyoD1 immunohistochemical staining. Figure 1D. Ki-67 proliferation index (60% positive tumor cells)

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