

Recurrent Vaginal Rhabdomyosarcoma in 2 Year Old: A Therapeutic Challenge

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Received: 02 Oct 2026

Accepted: 12 Oct 2026

Published: 14 Oct 2026

J Short Name: ACMCR

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Citation:

Meera Luthra, Recurrent Vaginal Rhabdomyosarcoma in 2 Year Old: A Therapeutic Challenge. Ann Clin Med Case Rep® 2026; V17(1): 1-3

1. Abstract

Vaginal rhabdomyosarcoma (RMS) is rare in children. Embryonal botryoid RMS generally has a favourable prognosis, but local recurrence can pose a significant therapeutic challenge.

2. Introduction

Rhabdomyosarcoma (RMS) is the most common soft tissue tumour in children, accounting for 4 to 6% of all malignancies in paediatric age group¹. Its diagnostic and management approach remains a matter of debate, particularly in very young patients where preservation of fertility is a major concern. Currently, local debulking surgery followed by adjuvant chemotherapy constitutes the primary line of treatment the goal is to minimize the use of radiation whenever possible. However, radical surgery and radiotherapy still have a role in extensive disease and in cases of relapse.

3. Case Report

A 2-year-old girl presented to the paediatric surgery outpatient department with a complaint of a mass protruding from the vagina for 15 days.

On examination, the child was afebrile, active, and appeared well. The abdomen was soft and non-tender, with no palpable mass. Cervical, axillary, and inguinal lymph nodes were not palpable. The external genitalia were typically female and a 1 cm × 1 cm mass could be seen protruding from the vagina (Figure 1). There was no associated pain, bleeding, discharge, or foul odour. Radiological evaluation was performed. Ultrasonography could not clearly define the origin of the vaginal mass but revealed right-sided hydronephrosis. MRI demonstrated a large, polypoidal, enhancing mass lesion distending the vagina with non-enhancing septae, extending from the external os up to the vaginal introitus. No extra-vaginal extension or infiltration was noted. Both ovaries were normal for age.

A DMSA scan, performed in view of the hydronephrosis,

showed a normally functioning left kidney and a poorly functioning/non-visualized right kidney.

Pan-endoscopy revealed multiple polyps arising from the anterior vaginal wall between the 2 o'clock and 4 o'clock positions. There was no involvement of the rectum or urinary tract.

Surgical excision of the mass was performed on 30/06/23 (Figure 2). Histopathological examination confirmed embryonal rhabdomyosarcoma, botryoid type.

The child was referred to the paediatric hemato-oncology team. After staging, she was started on combination chemotherapy with vincristine, actinomycin D, and cyclophosphamide (VAC regimen) for 26 weeks -13/02/24. PET CT (13/2/24) - Showed no evidence of metabolically active disease anywhere in the body.

3 days after PET CT Scan her parents noticed a mass coming out of vaginal orifice. She underwent excision of tumor soft tissue under GA on 27.02.2024 (Figure 2). Histopathology showed Embryonal Rhabdomyosarcoma. Case was discussed in Tumour Board on 5/3/24 and plan was made to give Chemotherapy and Radiation

She was lost to follow up for 6 months and on 18/9/24 again came with complaint of mass bulging out from vagina for last 10 days

PET CT on (20/9/2024) showed an ill-defined plaque like soft tissue with mild peripheral enhancement seen in the vaginal canal causing distension with mild FDG uptake (SUVmax3.6), not seen previously. No infiltration is seen. Thereafter, patient underwent excision of vaginal polyp under GA on 27.9.24. (fig2). The Histopathological features were compatible with a recurrent embryonal rhabdomyosarcoma. On 16/10/2024. Patient was planned for intravaginal brachytherapy but was lost to follow up. It has been 3 years since her primary diagnosis. Telephonic follow up with the family was established but the child has recurrent diseases and they are not willing for further treatment.



Figure 1: Mass Protuding Out of Vagina.



(30/06/2023)



(27/02/2024)



(27/09/2024)

Figure 2: Polypoidal Grape Like Structures Excised from Vagina.

4. Discussion

Embryonal rhabdomyosarcoma is associated with mutations in the RAS (rat sarcoma) signalling pathway.

Clinically, the tumour classically presents as a nodular, grape-like mass protruding from the vagina. Associated symptoms may include leucorrhoea and malodorous discharge.

Diagnostic criteria for the botryoid variant include²:

1. Polypoid appearance of the lesion
2. Origin beneath a mucous membrane

Initial evaluation typically involves ultrasonography, followed by contrast-enhanced MRI of the pelvis, which is preferred in the paediatric population. Pan-endoscopy is essential to determine the exact origin of the mass and assess involvement of adjacent structures such as the urinary tract and rectum.

After surgical excision, staging includes chest radiography, bone marrow biopsy (as lungs and bone marrow are common metastatic sites), and PET-CT to evaluate for distant metastases.

Combination chemotherapy using vincristine, actinomycin D, and cyclophosphamide (VAC regimen) remains the standard of care³.

The treatment outcomes for metastatic rhabdomyosarcoma have remained relatively unchanged over the past three decades, the 3-year progression-free survival rates of low-risk, intermediate-risk, and high-risk groups are 88, 55–76, and <30%, respectively⁴

5. Conclusion

Embryonal rhabdomyosarcoma of the vagina, although often considered a favorable subtype, can behave as an aggressive disease with a high rate of recurrence, particularly in cases of incomplete treatment or poor compliance. This case highlights the critical importance of strict adherence to multimodal therapy and close follow-up, as interruptions in treatment significantly worsen outcomes. Early detection of recurrence and timely institution of combined modalities including surgery, chemotherapy, and radiotherapy are essential for optimal disease control.

Furthermore, the prolonged and intensive nature of treatment places a substantial mental, physical, and financial burden on the patient's family. This underscores the need for comprehensive care that includes not only medical management but also psychosocial support and counseling to help families navigate the challenges associated with long-term cancer treatment.

The family is still in touch with us but they have taken a conscious decision to not do any further intervention as yet.

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