Adult Cervical Embryonal Rhabdomyosarcoma

M. Grant Liska, MD; Se Yeon Seo, MS; Yi Yeon Seo, MS; Jamie Marko, MD

Case Summary

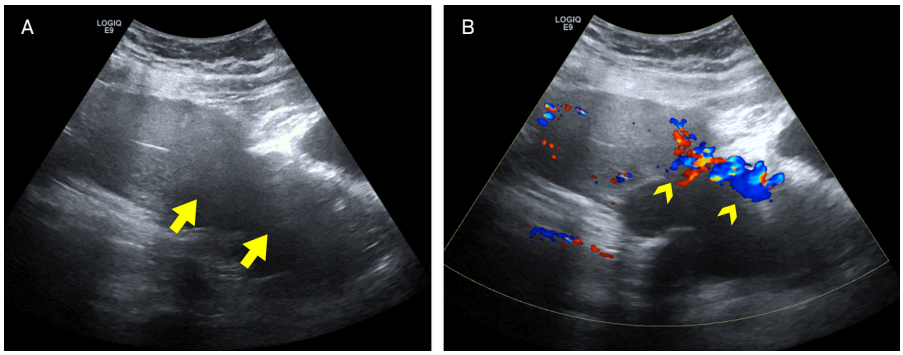
An adult patient presented with complaints of irregular vaginal bleeding with pelvic fullness and pain. Physical examination demonstrated a friable endovaginal soft-tissue mass, clinically presumed to be uterine prolapse.

The patient underwent a total abdominal hysterectomy and bilateral oophorectomy. Surgical pathology revealed cervical and uterine myoinvasion with no evidence of metastatic disease, consistent with stage IC according to the International Federation of Gynecology and Obstetrics (FIGO) Staging System for Adenosarcoma of Uterine Corpus, an unofficial staging system in this case. Following gross-total resection, the patient underwent four cycles of systemic chemotherapy, and the most recent postoperative imaging showed no evidence of disease recurrence.

Imaging Findings

Initial imaging with transabdominal US demonstrated an amorphous hypoechoic endocervical mass with significant vascularity (Figure 1). A follow-up MRI revealed a 9.0 cm lobular T1 hypointense, T2 hyperintense mass originating from the upper cervix/lower uterine segment

Figure 1. (A) Grayscale transabdominal US image in the sagittal plane reveals a poorly defined, hypoechoic mass (arrows) with posterior shadowing in the cervix. (B) Color Doppler image demonstrates significant vascularity (arrowheads).



border and protruding into the vaginal cavity (Figure 2). The mass demonstrated heterogeneous enhancement on T1 post-contrast images (Figure 2), as well as significant diffusion restriction (not shown). There was no convincing evidence of local invasion. On preoperative CT images, the mass appeared as an ill-defined hypoattenuating region causing cervical canal dilation and filling of the vaginal cavity (Figure 3).

Diagnosis

Cervical embryonal rhabdomyosarcoma (ERMS), stage IC.

Differential considerations include uterine prolapse, pedunculated cervical leiomyoma, endometrial polyp, cervical adenoma malignum, and other primary cervical malignancies.

Discussion

Rhabdomyosarcoma is a malignant mesenchymal tumor characterized by the presence of rhabdomyoblasts, immature skeletal muscle cells that fail to fully differentiate.[1] Among the various histological subtypes, ERMS is the most prevalent, accounting for roughly 50% of cases and affecting the genitourinary tract in 18-22% of cases.[2] ERMS typically affects infants, particularly in the vaginal area.[3] Cervical ERMS is a rare form of the disease, more commonly seen in adolescents during the 2nd decade of life.[4] Presentations of this entity in adulthood are even less common and are linked to somatic mutations in the DICER1 gene. Germline mutations in the DICER1 gene are frequently seen in pediatric patients with ERMS, which is associated with a number of other solid tumors.[5]

Affiliations: Department of Radiology, Walter Reed National Military Medical Center, Bethesda, Maryland (Grant Liska); Nova Southeastern University Dr. Kiran C. Patel College of Osteopathic Medicine, Clearwater, Florida (SY Seo, YY Seo); American College of Radiology Institute for Radiologic Pathology, Silver Spring, Maryland (Marko).
Disclosures: The authors have no conflicts of interest to disclose. None of the authors received outside funding for the production of this original manuscript, and no part of this article has been previously published elsewhere.

https://doi.org/10.37549/AR-D-25-0131

Figure 2. (A) Sagittal T2 sequence demonstrates a high water-content mass (bracket) protruding from the endocervical canal into the vaginal cavity. (B, C) Axial images show a T1 hypointense, T2 hyperintense mass with a thin rim of vaginal tissue (arrows). (D) T1 post-contrast subtraction images reveal heterogeneous contrast enhancement of the mass (bracket).

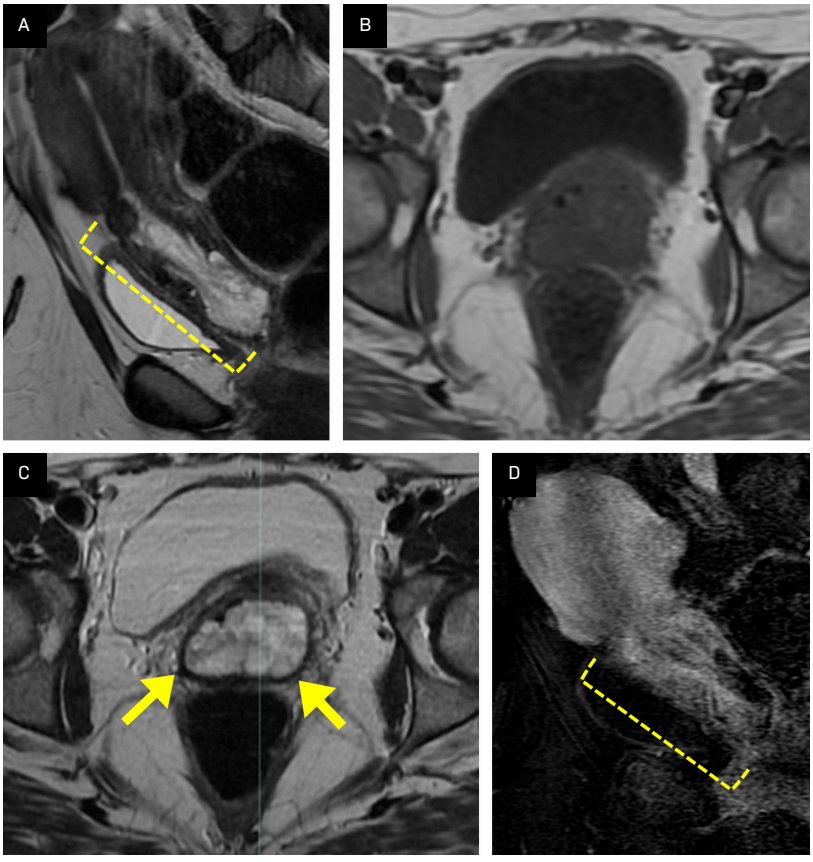
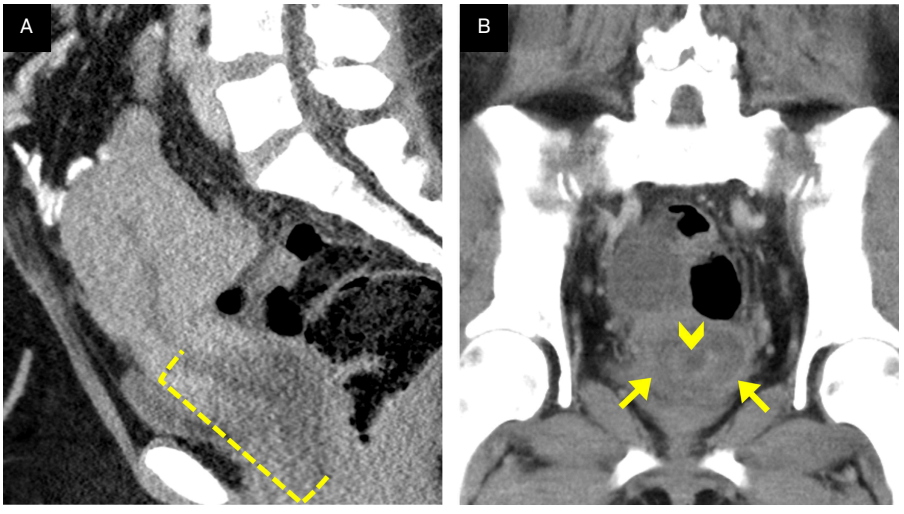


Figure 3. (A) Sagittal contrast-enhanced CT images demonstrate the low-density endocervical mass (bracket) with extension into the vaginal cavity. (B) Coronal images demonstrate the endocervical component of the mass (arrowhead) with surrounding dilated cervical tissue (arrows).



Cervical ERMS tumors in adults tend to be quite large at diagnosis, likely owing to their similar appearance to more common, benign entities such as uterine prolapse, endocervical fibroids, and cervical polyps on pelvic exam and US. Distinctive imaging features of this entity are helpful in making the correct diagnosis, with tumors having high water content (hypoattenuating on CT, T2 hyperintense on MRI) and often assuming a polypoid morphology likened to a bunch of grapes (sarcoma botryoides).[5,6] These features are not pathognomonic, though, and ultimately, tissue sampling is necessary for a definitive diagnosis.

There is no official classification of cervical ERMS; however, based on the largest study of cervical ERMS, the FIGO uterine adenosarcoma staging system was found to be superior to the uterine corpus sarcoma staging system for predicting survival.[5,7] Using this system, this patient's disease was classified as stage IC owing to surgical pathology's demonstration of greater than 50% myometrial/cervical invasion without extrauterine extension or metastatic disease.

Treatment guidelines are based on data from pediatric populations but have demonstrated similar efficacy in case reports of adult patients with cervical ERMS.[8] The prognosis for localized ERMS, including cervical cases, is generally favorable, particularly when early diagnosis and an aggressive surgical approach are achieved.[3-5,9,10] Following gross total resection, this patient underwent 4 cycles of vincristine, actinomycin, and cyclophosphamide therapy, with no imaging evidence of disease recurrence to date.

Conclusion

ERMS is a malignancy typically seen in children. Infrequently, this tumor can occur in the cervix of adolescents and, even more rarely, present as an adult

cervical cancer. Familiarity with this entity and its distinctive imaging features on MRI is critical to accurate, timely diagnosis in the adult population.

References

1) Liptak JM, Forrest LJ. Soft tissue sarcomas. In: Withrow SJ, Vail DM, Page RL, eds. *Withrow and MacEwen's small animal clinical oncology*. 5th ed. W.B. Saunders. 2013:356-380. doi:10.1016/B978-1-4377-2362-5.00021-9

2) Castle JT, Levy BE, Allison DB, Rodeberg DA, Rellinger EJ. Pediatric rhabdomyosarcomas of the genitourinary tract. *Cancers*. 2023;15(10):2864. doi:10.3390/cancers15102864

3) Kaserer K, Kainz C, Reinthaller A. Sarcoma botryoides (embryonales rhabdomyosarkom) als ungewöhnliche diagnose eines zervixpolypen [sarcoma botryoides (embryonal rhabdomyosarcoma) as an unusual diagnosis of cervix polyps]. *Pathologe*. 1995;16(4):287-291. doi:10.1007/s002920050104

4) Zeisler H, Mayerhofer K, Joura EA, et al. Embryonal rhabdomyosarcoma of the uterine cervix: case report and review of the literature. *Gynecol Oncol*. 1998;69(1):78-83. doi:10.1006/gyno.1998.4962

5) Devins KM, Young RH, Ghioni M, et al. Embryonal rhabdomyosarcoma of the uterine cervix: a clinicopathologic study of 94 cases emphasizing issues in differential diagnosis staging, and prognostic factors. *Am J Surg Pathol*. 2022;46(11):1477-1489. doi:10.1097/PAS.0000000000001933

6) Dehner LP, Jarzembowski JA, Hill DA. Embryonal rhabdomyosarcoma of the uterine cervix: a report of 14 cases and a discussion of its unusual clinicopathological associations. *Mod Pathol*. 2012;25(4):602-614. doi:10.1038/modpathol.2011.185

7) American Cancer Society. Rhabdomyosarcoma stages and risk groups. American Cancer Society website. Accessed January 26, 2026. https://www.cancer.org/cancer/types/rhabdomyosarcoma/detection-diagnosis-staging/staging.html

8) Ibrahim U, Saqib A, Mohammad F, et al. Embryonal rhabdomyosarcoma of the cervix: a rare disease at an uncommon age. *Cureus*. 9(11):e1864. doi:10.7759/cureus.1864

9) Ricker CA, Crawford K, Matlock K, et al. Defining an embryonal rhabdomyosarcoma endotype. *Cold Spring Harb Mol Case Stud*. 2020;6(2):a005066. doi:10.1101/mcs.a00506610.1097/PAS.0000000000001933

10) Smrkolj S, Rakar S, Malić S, Sinkovec J, Kobal B. Advanced embryonal rhabdomyosarcoma of the uterine cervix: a case report. *Eur J Gynaecol Oncol*. 2011;32(2):203-205. doi:10.12892/ejgo201102203