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Embryonal Rhabdomyosarcoma in the Pelvis of a 22-Year-Old Female: A Case Report

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ABSTRACT

Embryonal rhabdomyosarcoma is a rare, soft-tissue sarcoma responsible for less than 1% of malignancies in the adult population. We present a case of a 22-year-old female with a six-month history of abnormal uterine bleeding found to have an exophytic mass within the cervix. Magnetic resonance imaging revealed a large pelvic mass. Further workup included biopsies and surgical removal of the tumor, leading to the diagnosis of embryonal rhabdomyosarcoma. PET scan showed no FDG-avid malignancy, aiding in the decision-making process for adjuvant treatment to include vincristine, actinomycin D, cyclophosphamide, and mesna. This case is important to document due to the rarity of this disease. It also serves as a reminder to gynecologists to consider the addition of rhabdomyosarcoma in the differential diagnosis when evaluating young adult females presenting with abnormal uterine bleeding and a cervical mass lesion.

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embryonal rhabdomyosarcoma, pelvic mass, vaginal bleeding

INTRODUCTION

Rhabdomyosarcoma (RMS) is one of the most common soft tissue sarcomas (STSs) in pediatrics.[1] However, adult RMS is rare, representing only 3% of STSs and less than 1% of all malignancies.[2] Sixty to 70% of cases occur in patients less than ten years old, but diagnosis in adults yields less than a 1% incidence rate.[3,6] The reported embryonal RMS adult cases appear in the second and third decades of life and present with an exophytic mass and vaginal bleeding.[6] At the time of diagnosis, abdominopelvic RMS is usually a large mass due to its clinically silent characteristics until it compresses or invades vital organs. This kind of tumor in female adults is sometimes misdiagnosed as female malignancies due to the similarity in terms of clinical presentation. This case is important to document due to the rarity of this disease. This case also serves as a reminder to gynecologists to consider the addition of rhabdomyosarcoma in the differential diagnosis when evaluating young adult females presenting with abnormal uterine bleeding and a cervical mass lesion.

CASE

A 22-year-old nulligravid female was referred to the gynecologic oncology clinic for evaluation after a six-month history of heavy, abnormal uterine bleeding. Eight months prior, she had been admitted to an outside hospital for severe, symptomatic anemia with a hemoglobin and hematocrit of 3.8 g/dl and 17.0%, respectively. Her anemia was corrected with blood transfusions, and she was trialed on several oral contraceptives with no improvement in bleeding. The patient could not recall if she received a pap smear at age 21 at the outside facility. She was transferred to a new OB/GYN and switched to Aygestin 15mg daily for menstrual suppression. Initially, the patient noticed relative improvement, but bleeding and cramping associated with fatigue and tachycardia worsened after one month of treatment. She was urgently evaluated in the office with a pelvic examination revealing a concerning mass protruding from the cervix. A transvaginal ultrasound exhibited a uterine didelphys versus a bicornuate uterus with a large, vascular, complex mass extending from the fundus through the cervix. The patient was referred to

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gynecologic oncology.

A same-day appointment was made with the gynecologic oncologist. After similar physical exam findings in the office, the patient was scheduled for an exam under anesthesia (EUA) for a more thorough evaluation. During the EUA, an exophytic, necrotic, malodorous mass measuring approximately 6x6x6 centimeters was noted protruding from the cervix. Multiple biopsies were obtained at the time of the exam. Initial pathology from these biopsies revealed acute and chronically inflamed fibrin with dystrophic calcifications, necrotic and acutely inflamed granulation tissue, and cervical tissue with extensive ulceration with features suggestive of an underlying stromal process. Confirmatory diagnosis required further specimen collection.

Two weeks later, the patient underwent MRI to evaluate the mass further. It was found to contain cystic components and measured 2x7x6.8 cm with extension to the periphery of the cervical stroma, the myometrium, and the superior portion of the vagina. Though challenging to evaluate due to motion artifact, the uterus was described as distorted and was thought to be related to a congenital abnormality such as a septate uterus, bicornuate uterus, or possibly a desmoplastic response. A repeat biopsy was performed at the follow-up visit, and a pathology consultation from Mayo Clinic demonstrated a possible stromal process, though a deeper biopsy was requested for a definitive diagnosis. Further treatment options were discussed, and the decision was made to proceed with EUA, a possible radical trachelectomy, and a possible modified radical hysterectomy. During pre-anesthesia testing, the patient was found to be anemic with a hemoglobin of 4.3 g/dL and hematocrit of 14.4%. She was subsequently admitted, and her anemia was corrected prior to surgery.

On pelvic laparoscopy, the mass was found to be causing inversion of the uterus (Figure 1A) with dense adhesions to the pelvic sidewall, left adnexa, and sigmoid colon. After lysis of adhesions, reduction was attempted in hopes of performing a fertility-sparing trachelectomy. Due to the size of the mass, the extent of pelvic adhesions, and severe concern for malignancy, however, a modified radical hysterectomy was ultimately performed. After extraction, the full extent of the mass was assessed, measuring approximately 10x9x7cm (Figure 1A-B). The specimen was then sent for pathologic evaluation with consultation from Mayo Clinic.

Microscopic evaluation (Figures 1 and 2) of the tumor revealed malignant-appearing spindles and

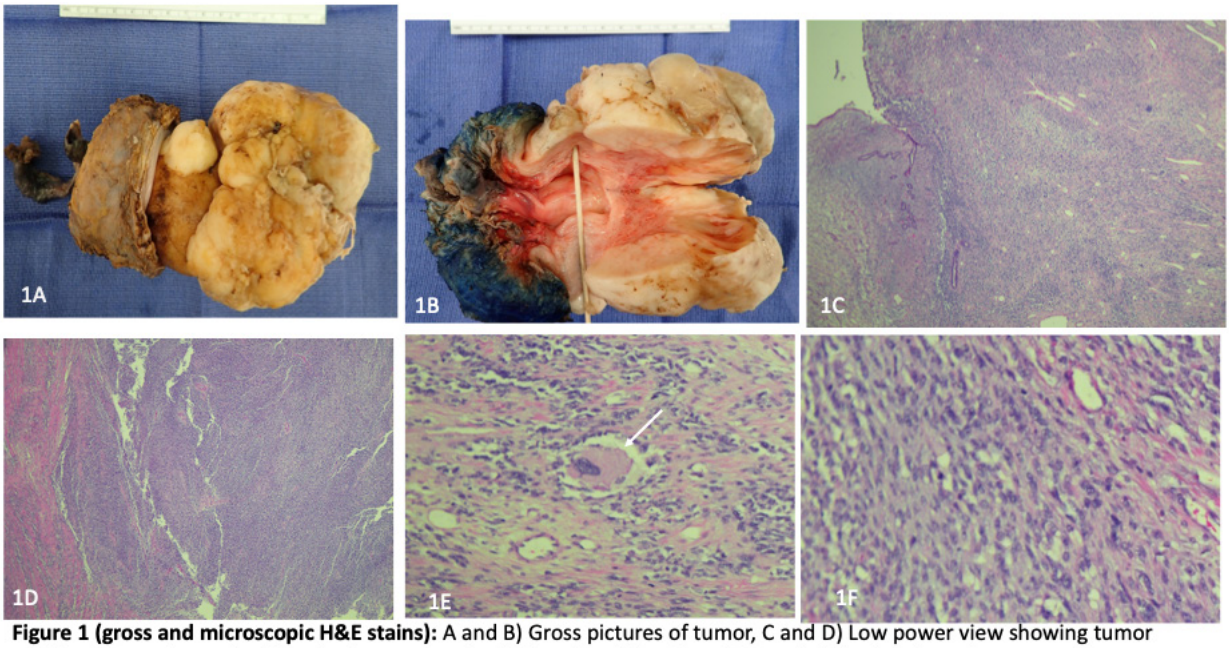

Figure 1 (gross and microscopic H&E stains): A and B) Gross pictures of tumor, C and D) Low power view showing tumor proliferation underneath of cervical mucosa and spindle cell proliferation of tumor infiltrating through myometrium, respectively, E and F) High power view showing epitheloid and rare rhabdoid features (arrow), and spindle cell proliferation,

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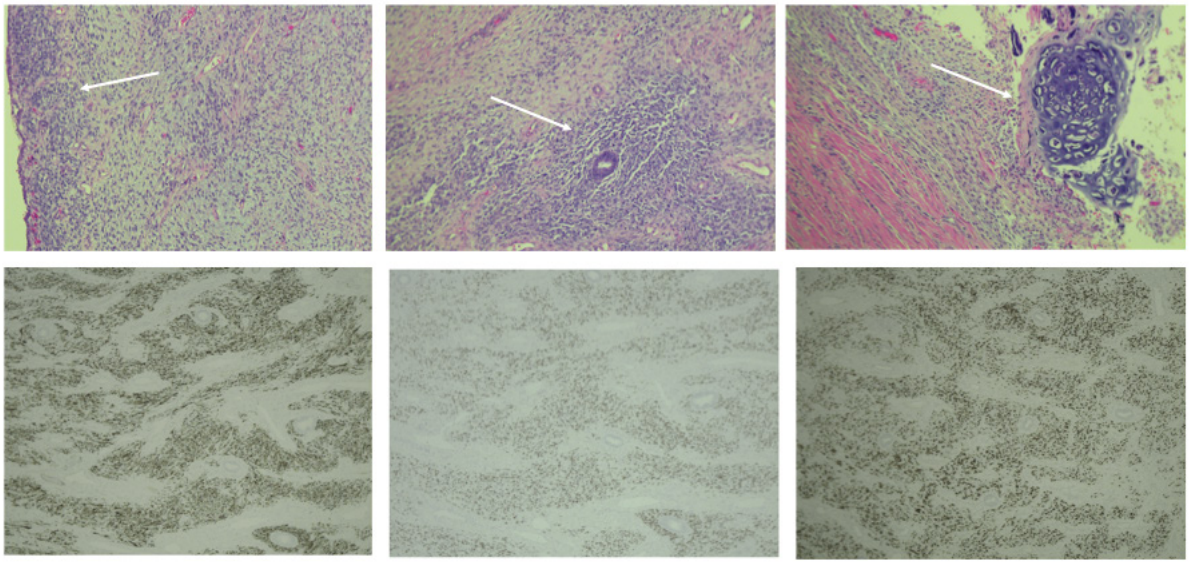

Figure 2 (microscopic H&E and immunohistochemical stains) : A and B) Cambian-type layer (arrow), condensation of tumor underneath of cervical mucosa and around benign endometrial glands in the background of myxoid stroma, respectively, C) heterologous component (cartilage), D, E and F) Immunohistochemical stain for desmin, MyoD1 and myogenin, respectively.

small round-cell tumor proliferation within the cervical and endometrial tissue with penetration into the myometrium. There were focal areas of cross-striation with eosinophilic cytoplasm and a focal area of cartilage present. Mitotic activity was abundant, and a high degree of cytologic atypia was identified. Characteristic features of cambium-type layers were also present. Immunohistochemical studies (Figure 3) showed strong immunoreactivity for muscle markers, including desmin, myogenin, and MyoD1. Immunostaining for CD10 and SMA demonstrated weak reactivity, and S-100 and HMB45 were negative. Immunomorphological findings along with clinical history supported a diagnosis of embryonal rhabdomyosarcoma with heterologous elements consisting of cartilage.

The patient underwent a PET scan for definitive staging that showed no evidence of FDG-avid malignancy. She was referred to the pediatric hematologist-oncologist for treatment consisting of 34 weeks of combination chemotherapy, including four cycles of vincristine, actinomycin, cyclophosphamide, and mesna, in addition to four cycles of vincristine and actinomycin D without radiotherapy.

DISCUSSION

Gynecologists commonly encounter vaginal bleeding complaints. At times, pelvic exams are deferred in younger patients to avoid trauma potentially associated with the exam. This patient was trialed on several oral contraceptives followed by Aygestin with continued menorrhagia resulting in hospitalization for blood transfusions. Patients with vaginal bleeding who are not responding to medical management need pelvic exams. As pelvic exams may not be well tolerated in the pediatric or adolescent population, ultrasound can be an appropriate starting point for evaluation prior to proceeding with an exam under anesthesia. Vaginal bleeding can have a broad differential that ranges from benign to malignant diagnoses.

Rhabdomyosarcoma (RMS) is an uncommon cancer that arises from soft tissue, typically in skeletal muscle, the bladder, the uterus, and the head and neck. RMS has been known to arise in all age groups, but it often affects children, with 60-70% of cases occurring in patients less than ten years old.[3,6] In adults, this diagnosis is rare, with less than a 1% incidence.[6] The reported embryonal RMS adult cases

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tend to appear in the second and third decades of life and present with an exophytic mass and vaginal bleeding.[6]

These tumors are derived from myogenic precursors and fall under the category of small round blue cell tumors. There are four histologic subtypes of RMS; the two most commonly seen subtypes are embryonal and alveolar.[3] Diagnosis of RMS utilizes imaging, biopsy, histology, and immunohistochemical testing to differentiate RMS from other small round blue cell tumors. The immunohistochemical markers specific for RMS (skeletal muscle differentiation) include desmin, myogenin, and MyoD1, to which our patient's tumor was strongly immunoreactive.[7] These three markers help distinguish RMS, but they do not help distinguish between the subtypes of RMS. For subtype differentiation, histology is utilized. Prognosis varies depending on the patient's age, tumor location and size, immunohistochemistry, lymph node involvement, and level of metastasis. Favorable sites include the orbit and eyelid, head and neck, genitourinary, and biliary tract. Unfavorable locations include the bladder, prostate, extremity, parameningeal, trunk, retroperitoneal, and pelvis.[8]

Treatment options in children and adults typically begin with surgical removal. The goal is to eradicate the cancer; however, that is not always possible, nor is surgery always an option. Chemotherapy and radiation therapies have increased embryonal RMS's 5-year, event-free survival to approximately 70%.[7] With our patient, we are utilizing PET scans to assure that her cancer has been completely removed and has not metastasized before finalizing her adjuvant treatment plan. Adjuvant chemotherapy for embryonal RMS is typically a regimen of vincristine, dactinomycin, and cyclophosphamide set forth by IRSG (2001). This particular combination of chemotherapy has been shown to optimize survival, minimize toxicity, and simplify the therapy plan for these patients.[5] Three-year survival rates of low, intermediate, and high-risk groups are 88%, 55-76%, and <30%, respectively.[3] Good prognoses in low-risk groups have led to adjustments in treatment in an attempt to reduce treatment-related complications.[3] While these treatment methods are suggested for embryonal RMS, they are not specific for the female genital tract.

CONCLUSION

While embryonal rhabdomyosarcomas are more common in pediatric populations, this disease should be added to the differential diagnosis for abnormal uterine bleeding in adult populations. Utilizing imaging, biopsy, histology and immunohistochemical staining is essential to make an accurate diagnosis and create an appropriate treatment plan for these patients. Prognosis is good for early-stage embryonal RMS; thus, prompt diagnosis is essential to achieve favorable outcomes. Cases of RMS should continue to be reported due to the rarity of the disease and a lack of studies detailing definitive treatment strategies directed towards the female genital tract.

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