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Imaging of Vaginal Yolk Sac Tumor: 2 Case Reports and Literature Review

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Abstract **Case Report**

Our article reports on two cases of pediatric vaginal yolk sac tumors (YSTs), underscoring the diagnostic challenges posed by their similarities to other vaginal tumors, particularly in differentiating from embryonal rhabdomyosarcoma based on clinical and radiological presentations. Emphasizing the critical role of MRI, histological examination, and alpha-fetoprotein levels in diagnosis, we highlight the complexities of treatment, which has evolved from radical surgeries to more conservative approaches with adjuvant chemotherapy, aiming to preserve future sexual and reproductive functions. Our findings advocate for individualized treatment plans and stress the importance of accurate diagnosis and the potential need for alternative therapeutic strategies in cases of atypical response to treatment.

Keywords: Vaginal yolk sac tumors (YSTs), Embryonal rhabdomyosarcoma, MRI, Radiology, Histology.

INTRODUCTION

The Endodermal Sinus Tumor (EST), though less common than Embryonal Rhabdomyosarcoma, presents a significant clinical challenge, particularly in pediatrics. It primarily affects young children, almost exclusively under the age of three. The clinical and radiological presentation of EST mirrors that of Embryonal Rhabdomyosarcoma, making accurate diagnosis challenging.

Due to the non-specific nature of imaging characteristics, the role of radiologists in identifying this tumor is limited. The definitive diagnosis of EST relies on histological analysis and elevated levels of Alpha-Fetoprotein (AFP). Notably, the vaginal subtype of EST is recognized for its aggressive nature. In this context, we report the cases of two patients with vaginal tumors to highlight their imaging characteristics, emphasize the difficulties in diagnosis, and delineate the distinguishing imaging features between Yolk Sac Tumors and other types of vaginal tumors.

These cases underscore the complexity and urgency in managing EST, emphasizing the need for heightened awareness and precise diagnostic approaches.

CASE REPORTS

The first case is a 6-month-old female infant who was admitted to our facility with a history of intermittent vaginal bleeding, her medical history was not significant and her family history was non-contributory. Laboratory investigations were largely within normal limits, except for an elevated alpha-fetoprotein (AFP) level. Pelvic ultrasonography indicated the presence of a vaginal mass, and follow-up MRI revealed a cervico-vaginal tumoral process with heterogeneous signal, featuring a necrotic center showing low signal intensity on T1 and high signal intensity on T2, and a peripheral wall exhibiting low signal intensity on T1 and high signal intensity on T2, with enhancement following contrast agent injection. A biopsy of the mass revealed a Germ Cell Tumor. With these findings and the elevated AFP levels, a diagnosis of vaginal yolk sac tumor was made. No metastasis was detected in the CT scan. After three cycles of chemotherapy, the patient underwent a control MRI which revealed a decrease in the size of the tumor.

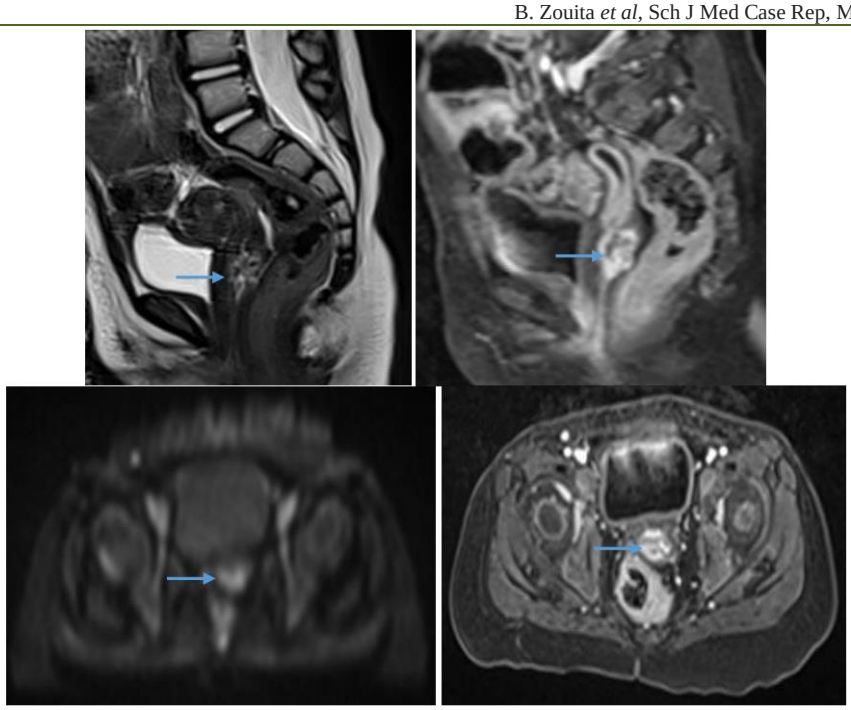

Fig 1: Pelvic MRI in DWI, T2 and T1 sequences after gadolinium injection revealing a Well-defined, largely necrotic cervico-isthmic uterine tumoral process related to a malignant germ cell tumor of the vagina

The second case is a 12-month-old female presented to our hospital with a complaint of vaginal spotting that has been ongoing for two months. She had no significant medical or family history. Routine blood tests were normal, except for a markedly elevated AFP level. Ultrasound examination of the pelvis identified a vaginal mass, which was further characterized by MRI as a large budding tumoral mass filling the vaginal cavity with breach of the musculature of the left lateral vaginal wall, infiltration of the adventitia, the perineum, and the left levator ani muscle. The tumor adheres without clear boundaries to the pelvic floor and the vesical trigone. The MRI appearance initially suggested a rhabdomyosarcoma of the urogenital sinus.

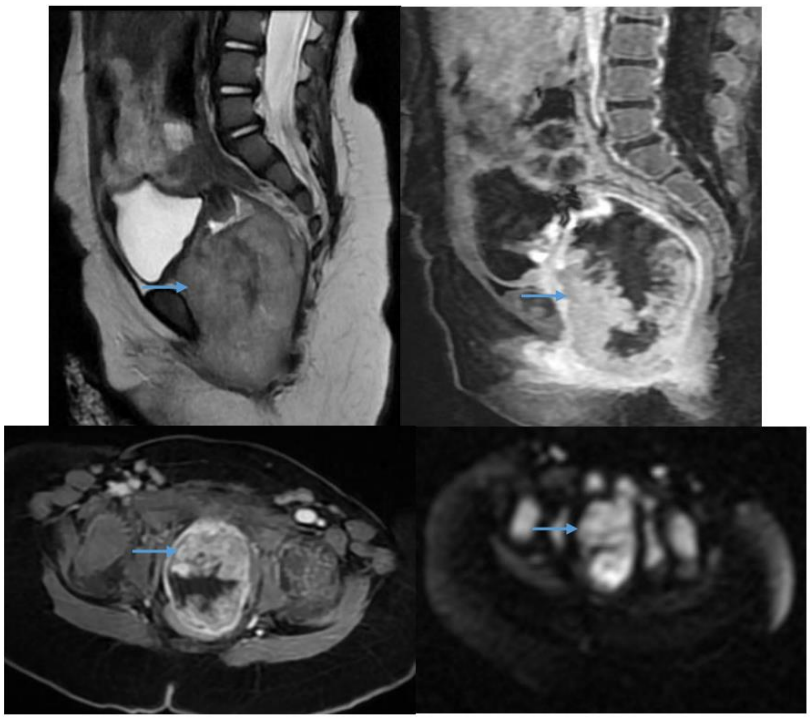

Fig 2: Pelvic MRI in DWI, T2 and T1 sequences after gadolinium injection revealing a budding tumor mass filling the vaginal cavity, consistent with a vaginal germ cell tumor

Biopsy of the mass showed a Germ Cell Tumor. The combination of histological features and high serum AFP supported the diagnosis of a yolk sac tumor of the vagina. The CT scan did not show any metastatic spread.

DISCUSSION

Our study's analysis of vaginal yolk sac tumors, reveals critical insights into their clinical presentations, diagnostic challenges, and therapeutic approaches. While RMS is the most common vaginal tumor in the first two years of life, the vagina remains a rare site for malignant Germ Cell Tumors (GCTs), with YST being the most prevalent histological subtype in girls under three years. Alpha-fetoprotein (AFP) has emerged as a reliable biomarker for diagnosing GCTs, assessing treatment response, and monitoring remission or disease progression.

In comparing our two case reports with the existing literature, several key observations emerge. Both of our cases align with the literature in terms of age of presentation and primary symptoms, which is consistent with the trend of these tumors occurring predominantly in young girls under three years old, with vaginal bleeding as a common initial symptom.

In terms of radiological diagnosis, MRI features of vaginal YST, characterized by solid heterogeneous appearances with necrotic areas, are similar to those of RMS. This similarity poses a significant diagnostic challenge, as other conditions like hemangiomas and hematometrocolpos can also mimic these malignancies.

The MRI findings in our first case, showing a necrotic center with distinct signal characteristics on T1 and T2, and the second case's presentation of a large budding mass with musculature and perineal infiltration, are in line with the typical radiological features described for these types of tumors.

Historically, the management of vaginal YST involved radical surgeries like vaginectomy, often resulting in severe impairment of sexual and reproductive functions. However, advancements in surgical techniques and adjuvant chemotherapy have facilitated more conservative approaches, preserving vaginal function and future fertility prospects.

Chemotherapy, has been effective in treating vaginal YSTs, but concerns about long-term toxicities such as hearing loss, secondary malignancies, and sterility remain.

Our review of pediatric vaginal tumors emphasizes the necessity of individualized treatment plans. In cases where vaginal preservation is feasible, upfront surgery may be considered, especially given the sensitivity of vaginal YST to chemotherapy. Recent trends indicate a shift towards chemotherapy-only approaches for vaginal YSTs in children, showing promising results with complete tumor response and AFP normalization.

Our study highlights the complexities in diagnosing and managing pediatric vaginal tumors, with a particular focus on the balance between effective treatment and minimizing long-term adverse effects.

CONCLUSION

In summary, it's crucial to thoroughly investigate vaginal bleeding in infants. Diagnostic accuracy is improved with ultrasound or MRI, while histological examination and AFP levels are essential for definitive diagnosis and follow-up.

Currently, the combination of conservative surgery and adjuvant chemotherapy is favored, potentially offering less toxicity in the long run due to fewer chemotherapy cycles.

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