CASE REPORT

Vaginal endodermal sinus tumor in an infant: a case report

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ABSTRACT

Background: Endodermal sinus tumor is a malignant germ cell neoplasm, most commonly of gonadal origin. Its occurrence in the vagina is extremely rare.
Case description: We report the case of a 7-month-old infant presenting with intermittent vaginal discharge and bleeding for 3 months. Initial ultrasound revealed a vascularized hypoechoic lesion located in the upper vaginal canal. Laboratory tests showed elevated serum alpha-fetoprotein levels (range: 1,415–2,500 ng/mL). Subsequent magnetic resonance imaging identified a solid tumor in the upper third of the vaginal canal, with mass effect on adjacent structures, avid heterogeneous contrast enhancement, diffusion restriction, and no evidence of regional infiltration. Histopathological analysis of an expelled vaginal blood clot confirmed the diagnosis of endodermal sinus tumor with Schiller-Duval bodies. The patient underwent six cycles of chemotherapy with bleomycin, etoposide, and cisplatin over 6 months, without clinical complications. Follow-up imaging 3 months after completion of treatment showed a reduction in tumor size and no evidence of intra- or extrapelvic metastases. Alpha-fetoprotein levels progressively decreased. A subsequent biopsy was negative for malignant neoplasia.
Conclusion: Vaginal endodermal sinus tumor represents an extremely rare extragonadal presentation, primarily observed in infancy. Imaging plays a crucial role in the diagnosis, follow-up, and therapeutic planning of this entity.

Keywords: Endodermal Sinus Tumor; Vaginal Discharge; Vaginal Neoplasms; Magnetic Resonance Imaging; Case Reports (Source: MeSH)

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Tumor de seno endodérmico vaginal en una lactante: reporte de un caso

RESUMEN

Antecedentes: El tumor de seno endodérmico es una neoplasia maligna de células germinales, habitualmente gonadal. Su localización vaginal es excepcional.
Descripción del caso: Se presenta el caso de una lactante de 7 meses de edad con flujo y sangrado vaginal intermitente de 3 meses de evolución. La ecografía inicial reveló una lesión hipoecogénica vascularizada ubicada en el canal vaginal superior. Los estudios de laboratorio revelaron niveles séricos elevados de alfa fetoproteína (rango: 1 415 – 2 500 ng/mL). Posteriormente, la resonancia magnética identificó una lesión tumoral sólida en el tercio superior del canal vaginal, con efecto de masa sobre estructuras circundantes, ávido realce heterogéneo al contraste, señal de restricción en la secuencia de difusión y ausencia de infiltración regional. El análisis anatomopatológico de un resto hemático vaginal expulsado confirmó el diagnóstico de un tumor de seno endodérmico con presencia de cuerpos de Schiller Duval. La paciente recibió 6 ciclos de quimioterapia con bleomicina, etopósido y cisplatino durante 6 meses, sin complicaciones clínicas. El último control por imágenes, 3 meses después de finalizar el tratamiento, reveló la reducción del tamaño tumoral y la ausencia de lesiones metastásicas intra y extrapélvicas. Los niveles de alfa feto proteína disminuyeron progresivamente. Un nuevo estudio de biopsia resultó negativo para neoplasia maligna.
Conclusión: El tumor de seno endodérmico vaginal constituye una presentación extragonadal muy rara, observado mayormente en la etapa infantil. El rol de las imágenes es clave para el diagnóstico, seguimiento y planificación terapéutica de esta entidad.

Palabras clave: Tumor del Seno Endodérmico; Excreción Vaginal; Neoplasias Vaginales; Imagen por Resonancia Magnética; Informe de caso (Fuente: DeCS)

INTRODUCTION

Endodermal sinus tumors, also known as yolk sac tumors due to their content of primitive endodermal cells, are malignant neoplasms derived from germ cells (1). Because of their germ cell origin, they usually arise in the gonads of both sexes; however, less frequent extragonadal locations have also been reported, including the vagina, uterus, vulva, mediastinum, liver, prostate, and diaphragm, among others (2).

Vaginal yolk sac tumors are sporadic, accounting for 3–8% of all extragonadal tumors, and they primarily affect girls under 3 years of age (3). The clinical presentation is usually asymptomatic at the beginning and subsequently manifests with painless vaginal bleeding or abnormal vaginal discharge (2).

In girls, the limited access to the vaginal cavity makes imaging studies essential for determining the location and extent of the tumor. Ultrasound is the initial study used to evaluate pelvic structures, such as the uterus and ovaries. At the same time, magnetic resonance imaging (MRI) is considered the indispensable tool for adequately characterizing lesions, their location, and possible infiltration of adjacent organs (4). Likewise, the assessment of alpha-fetoprotein (AFP) levels is beneficial for both diagnosis and monitoring treatment response (5).

We present the case of an infant treated at a national pediatric referral center in Lima, Peru, who presented with intermittent vaginal discharge and bleeding and was ultimately diagnosed with a vaginal endodermal sinus tumor.

CASE DESCRIPTION

The case of a 7-month-old female patient is presented. She was born at 39 weeks of gestation by eutocic delivery, with a birth weight of 4.8 kg and length of 53 cm. No records of prenatal care or follow-up of growth and development were available. The patient's mother attended the outpatient clinic of a national pediatric referral center in Lima, reporting that for the past 3 months the infant had presented with intermittent vaginal discharge of "creamy" appearance, with progressively appearing blood streaks that became evident during straining.

At the center, vaginoscopy, laboratory tests including alpha-fetoprotein (AFP) measurement, and imaging studies by transabdominal pelvic Doppler ultrasound and magnetic resonance imaging (MRI) were indicated. Transabdominal pelvic ultrasound revealed a hypoechoic, ovoid structure in the upper vaginal canal, with marked vascularization on Doppler mode.

Hematological tests showed a hemoglobin level of 11.7 g/dL, a hematocrit of 34.5%, a total leukocyte count of 9,930/µL, and a platelet count of 373,000/µL. The percentages of neutrophils, band cells, eosinophils, and lymphocytes were 19%, 0%, 0.1%, and 6.9%, respectively. Blood biochemical tests revealed glucose levels of 103 mg/dL, urea levels of 11.3 mg/dL, and creatinine levels of 0.3 mg/dL. Serum tumor marker tests showed elevated AFP levels in all measurements, ranging from 1,415 to 2,500 ng/mL. Urine culture was negative. Cytopathological examination of a vaginal sample revealed superficial squamous cells without significant alterations, a chronic inflammatory infiltrate, and a few atypical spindle-shaped cells.

MRI confirmed the presence of an ovoid tumor in the upper third of the vaginal canal, measuring 35 × 32 × 24 mm (cranio-caudal × anteroposterior × transverse). The mass had lobulated borders, an intermediate T2 signal, and a low T1 signal, peripheral microcysts, avid heterogeneous contrast enhancement, and a high diffusion restriction signal. The lesion displaced the uterus and ligaments cranially, as well as the posterior bladder wall, without evidence of infiltration of these structures or the rectal wall, and no nodal involvement was observed in the pelvic or extrapelvic cavity (figura 1).

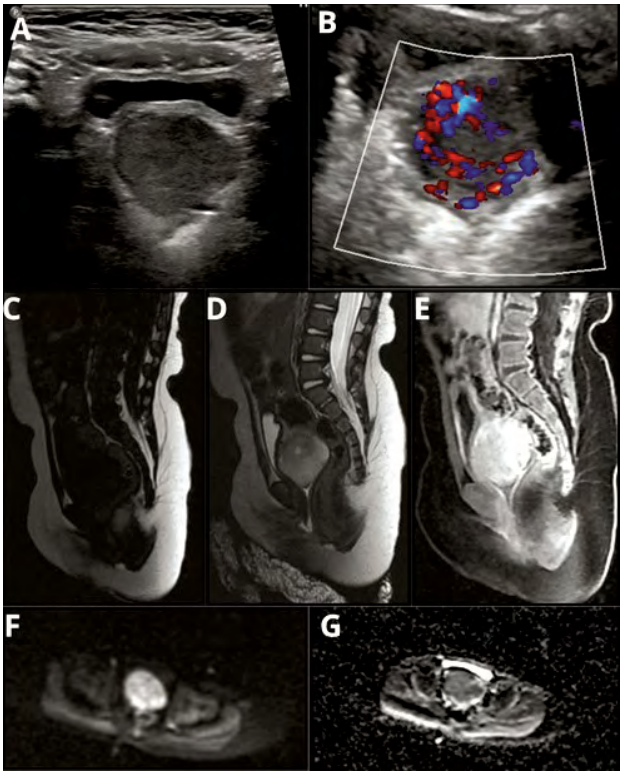

Figure 1. Imaging of the vaginal yolk sac tumor. **A-B**) Ultrasound images showing a hypoechoic mass with marked vascular flow on Doppler study. **C-G**) MRI images demonstrating an ovoid tumor lesion with intermediate T2 signal and low T1 signal, with avid contrast enhancement and diffusion restriction (**C**: T1 TSE, **D**: T2 FRFSE sagittal, **E**: T1+C, **F**: DWI, **G**: ADC).

Following the spontaneous expulsion of blood residues through the vaginal canal, the sample was sent for histological analysis in the Department of Pathology. The study revealed cellular atypia characterized by primitive nuclei, clear cytoplasm, and atypical mitoses, along with multiple cystic structures of varying sizes and the presence of Schiller–

Duval bodies, a pathognomonic finding of yolk sac tumors. Immunohistochemical studies showed positivity for Glypican-3 and Sall-4, focal positivity for AFP, and negativity for Desmin (figura 2).

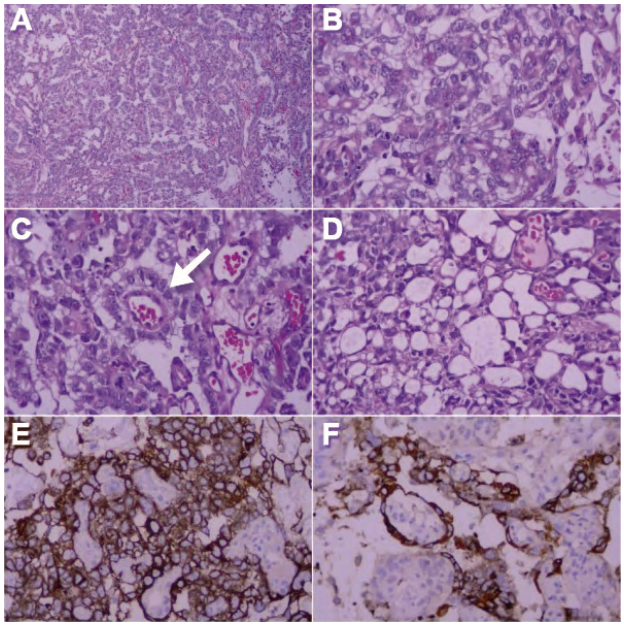

Figure 2. Histological sections of vaginal blood residue. **A**) Reticular growth pattern composed of a network of irregular, anastomosed channels. **B**) At higher magnification, primitive nuclei with prominent nucleoli, clear cytoplasm, and atypical mitoses are observed. **C**) The arrow indicates Schiller–Duval bodies, characterized by a central blood vessel surrounded by loose connective tissue, which is in turn encircled by epithelium with a primitive appearance. **D**) Cystic structures of varying sizes. **E**) Neoplastic cells with strong positivity for Glypican-3. **F**) Neoplastic cells with focal cytoplasmic positivity for AFP.

The patient underwent chemotherapy with six cycles of bleomycin, etoposide, and cisplatin over six months, demonstrating good oral tolerance and remaining afebrile, with hematological parameters within normal ranges, and without any relevant clinical complications during or after treatment.

The most recent MRI follow-up, performed three months after the last chemotherapy session, revealed a significant reduction in tumor size and no evidence of intra- or extrapelvic metastatic lesions. AFP levels progressively decreased from initial values of 2,500 ng/mL to 10.3 ng/mL, 8.56 ng/mL, and 6.3 ng/mL at 3, 6, and 8 months, respectively. A repeat biopsy of the tumor at six months post-treatment was negative for malignancy.

DISCUSSION

The current World Health Organization classification of pediatric tumors describes germ cell tumors independently of the primary organ involved (6). These are categorized as non-invasive germ cell tumors, germinoma family tumors, and non-germinomatous germ cell tumors. The latter group includes endodermal sinus or yolk sac tumors, in both prepubertal and postpubertal presentations. Several theories have been proposed to explain the extragonadal presentation of yolk sac tumors, including: 1) aberrant or ectopic migration of germ cells during embryogenesis, 2) reverse migration of germ cells, 3) abnormal differentiation of somatic cells, 4) derivation from pluripotent stem cells included in a somatic tumor, 5) origin from residual fetal tissue following an incomplete abortion (for primary endometrial yolk sac tumors), and 6) metastasis from an occult primary gonadal tumor (7). In infants with vaginal germ cell tumors, the theory of aberrant or ectopic migration is the most widely accepted (8).

The clinical presentation of unexplained vaginal bleeding in pediatric patients encompasses a broad etiological spectrum, ranging from idiopathic causes to tumors. Among the most common differential diagnoses in pediatric patients with intravaginal tumors is botryoid sarcoma, an embryonal rhabdomyosarcoma that presents with foul-smelling vaginal bleeding. This tumor is the most common sarcoma of childhood, accounting for up to 4.5% of pediatric cancers (9). In contrast, intravaginal localization of yolk sac tumors is extremely rare, presenting as a sessile, friable, whitish-gray lesion, predominantly attached to the posterior vaginal wall (10). In this context, histopathological and immunohistochemical studies are crucial for establishing a definitive diagnosis and guiding therapeutic management.

Initial imaging typically begins with a transabdominal pelvic ultrasound, which can identify nodular lesions with significant vascularization when viewed in Doppler mode. Yolk sac tumors share similarities with botryoid sarcoma; however, Sun *et al.* (9) described notable differences in the shape and extension of these vaginal tumors. While yolk sac tumors tend to be more localized and have a rounded morphology, botryoid sarcoma exhibits a "grape-like" appearance, filling the entire vaginal canal. On MRI, both display high T2 signal, high diffusion restriction, and heterogeneous contrast enhancement. In the present case, a tumor confined to the upper third of the vaginal canal was observed, without evidence of infiltration into surrounding structures, consistent with the findings described in the literature (11,12).

According to the Children's Oncology Group (13), extragonadal germ cell tumors are staged as follows: Stage I – complete resection of the mass at any location, with negative margins and tumor markers that may be positive or negative; Stage II – microscopic residual disease after surgery, negative lymph nodes, and tumor markers that may be positive or negative; Stage III – macroscopic residual disease or biopsy only, with retroperitoneal nodes that may be negative or positive and tumor markers that may be positive or negative; Stage IV – distant metastases, including liver involvement, regardless of surgical response and tumor marker status.

Current treatment of extragonadal yolk sac tumors in childhood combines surgery and chemotherapy with 4–6 cycles of cisplatin, etoposide, and bleomycin. This regimen has remained the standard recommendation for over two decades. With this approach, overall survival exceeds 90% in stages I and II, and 80% in stages III and IV (14).

Serum AFP levels in yolk sac tumors correlate with tumor size and are the most useful marker for diagnosis, follow-up, and detection of recurrence in infant vaginal tumors (3). Following treatment, normalization of AFP levels indicates a favorable response, whereas persistent elevation or slow decline may suggest residual disease or treatment resistance (15). Other tumor markers, such as β-HCG, carcinoembryonic antigen, and CA 19-9, may be elevated in the presence of concomitant tumor components (16). In this case, serum AFP levels decreased progressively after chemotherapy, correlating quantitatively with the significant reduction in tumor size observed on serial MRI follow-up.

The main limitations of this report include the inability to generalize the findings because it is based on a single case; limited retrospective clinical information, which prevented access to complete data on prenatal care, postnatal growth, and development, and genetic studies; and a short follow-up period of only 8 months after chemotherapy, which hindered the assessment of late complications and long-term treatment effectiveness.

The present case represents a vaginal yolk sac tumor, a rare extragonadal location. Imaging studies are crucial for defining tumor localization, extent, and infiltration into adjacent structures, as well as for evaluating lesion behavior; however, definitive diagnosis requires histopathological confirmation.

This case report was approved by the Institutional Research Ethics Committee of the center where the patient was treated.

Author contributions

Boris Borja-Zapata: Conceptualization, Investigation, Methodology, Project administration, Supervision, Validation, Visualization, and Writing – review & editing.

Carla Cruzado-Villanueva: Investigation, Supervision, and Methodology.

Georgette Borja-Urbano: Investigation, Supervision, and Methodology.

Conflicts of interest

The authors declare no relevant financial or non-financial conflicts of interest.

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Ethical aspects

This case report was approved by the Institutional Research Ethics Committee of the center where the patient was treated.

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