Malignant Yolk Sac Tumor of the Ovary Presenting as Adnexal Torsion in a Child: A rare Case Report with Review of Literature

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Abstract

Introduction: *Malignant yolk sac tumor is a rare ovarian neoplasm with peak incidence in young women. Its occurrence in children less than 10 years old is extremely uncommon. In this manuscript we report a rare case of malignant yolk sac tumor presenting with adnexal torsion, emphasizing the role of multimodality imaging in management.*

Case Presentation: *A 9-year-old female child was brought with the complaints of acute right lower quadrant pain, nausea and vomiting. On examination, the abdomen was tender with guarding. Laboratory investigations revealed leukocytosis, raised inflammatory markers and markedly elevated alpha-fetoprotein (AFP) levels. Ultrasonography revealed a mixed echogenic tumor in the right adnexa with features of adnexal torsion. Cross sectional imaging confirmed the presence of a heterogeneously enhancing tumor in the right adnexa with pelvic and paraaortic lymphadenopathy. Subsequently, the child underwent emergency laparotomy for adnexal detorsion with tumor excision. Postoperative histopathological examination confirmed malignant yolk sac tumor.*

Conclusions: *This case underscores the rare presentation of ovarian malignant yolk sac tumor as adnexal torsion in a child and the role of multimodality imaging in its management.*

Keywords: *Malignant Yolk Sac Tumor, Ovarian Germ Cell Tumor, Adnexal Torsion, Pediatric Ovarian Tumors, Multimodality Imaging*

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Abbreviations

1. AFP - Alpha-Fetoprotein
2. LDH - Lactate Dehydrogenase
3. HCG - Human Chorionic Gonadotropin
4. CA-125 - Cancer Antigen 125
5. CA-19.9 - Cancer Antigen 19.9
6. CEA - Carcinoembryonic Antigen
7. CECT - Contrast Enhanced Computed Tomography
8. MRI - Magnetic Resonance Imaging
9. US - Ultrasonography
10. RLQ - Right Lower Quadrant
11. BEP - Bleomycin, Etoposide, and Cisplatin (Chemotherapy regimen)
12. OGCT - Ovarian Germ Cell Tumors
13. GCT - Germ Cell Tumors
14. CT - Computed Tomography
15. DWI - Diffusion Weighted Imaging
16. ADC - Apparent Diffusion Coefficient
17. PID - Pelvic Inflammatory Disease
18. UTI - Urinary Tract Infection
19. ESR - Erythrocyte Sedimentation Rate
20. PAS - Periodic Acid-Schiff
21. SALL4 - Spalt-Like Transcription Factor 4

Introduction

Ovarian germ cell tumors arise from the germ cells of the ovary and account for ~ 15-20% of all ovarian neoplasms (1). Malignant germ cell tumors of the ovary are rare with an incidence of ~ 1-2% (2). Yolk sac tumors, also known as endodermal sinus tumors, are highly aggressive with a predilection for gonadal sites (3). The gonads, particularly the ovaries, are the primary sites for these tumors. However, extragonadal sites like broad ligament, retroperitoneum, mediastinum and sacrococcygeal region have also been reported (3-5) . These tumors are usually seen in young women and adolescent girls. Occurrence in children less than 10 years of age is extremely uncommon with a few sporadic case reports. Unlike epithelial tumors, malignant yolk sac tumors are locally aggressive and possess high metastatic potential. Due to their aggressive nature, early detection and management are critical for improving survival outcomes.

This highlights the rare presentation of malignant yolk sac tumor complicated by adnexal torsion, emphasizing the role of multimodality imaging in timely management.

Case Presentation

A 9-year-old girl child was brought to the emergency department with acute onset, severe right lower abdominal pain, nausea and few episodes of non-bilious vomiting. No history of preceding trauma, fever, or changes in urinary or bowel habits could be elicited. She had no prior abdominal surgeries or known health issues.

On physical examination, the patient appeared visibly distressed and was clutching her abdomen. She was afebrile with stable vital signs. Abdominal examination revealed marked tenderness in the right lower quadrant with guarding. Laboratory investigations showed a leucocytosis (white blood cell count of 18,600/µL) and elevated inflammatory markers (ESR 32 mm/hr). Notably, alpha-fetoprotein (AFP) level was markedly elevated at 32,134 ng/mL, strongly suggestive of a malignant germ cell tumor. Lactate Dehydrogenase (LDH) was also elevated at 635 IU/L. Levels of other tumor markers were within normal limits (Human Chorionic Gonadotropin (HCG)- 0.74 ng/ml; CA-125- 14.6 U/ml; CA-19.9 <0.8 U/ml; Carcinoembryonic Antigen (CEA)- 1.52 ng/ml).

Imaging Findings

Pelvic ultrasound revealed a large, heterogeneous, solid-cystic mass measuring 8 x 7.5 cm in the right adnexa. The mass exhibited mixed echogenicity, with areas of necrosis (Figure 1). Colour Doppler imaging demonstrated no vascularity within the lesion, suspicious of adnexal torsion. Free fluid was noted in the pelvis, raising further concern of a possible adnexal torsion. Few enlarged para aortic lymph nodes also noted.

Contrast enhanced computed tomography (CECT) confirmed the presence of a lobulated heterogeneously enhancing solid pelvic mass measuring approximately 8.0 x 8.0 cm in the right adnexa (Figure 2). It was seen compressing the uterus, displacing it towards the left, and causing a significant mass effect on the urinary bladder and the right ureter, resulting in moderate right-sided hydroureteronephrosis. Minimal ascites was present. Multiple enlarged heterogeneously enhancing para-aortic and pelvic lymph nodes were identified, largest measuring up

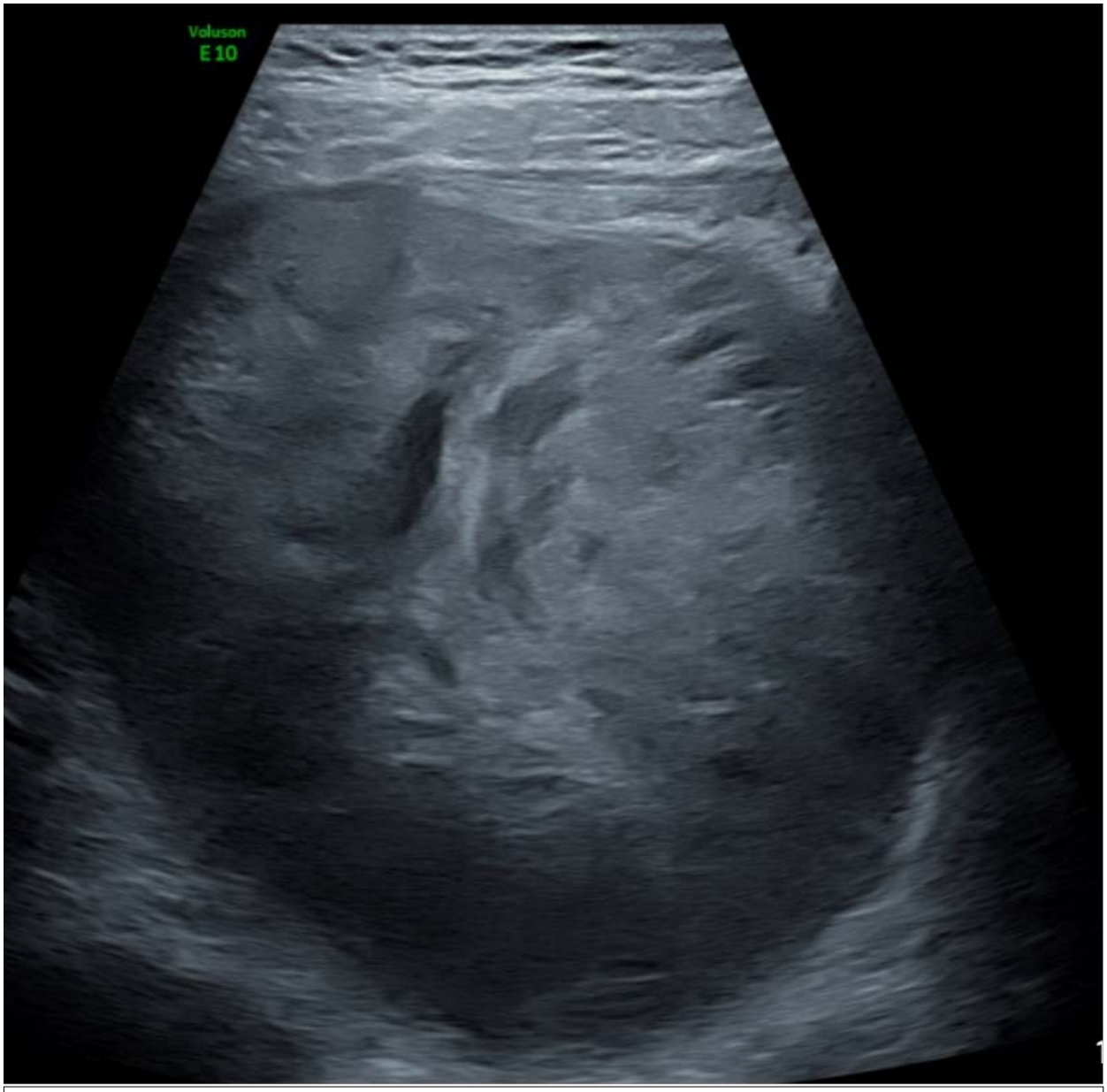

Figure 1: *Grey-scale ultrasonogramm showing a solid heterogeneous in the pelvis, predominantly involving the right adnexa.*

to 2.5 cm in diameter, consistent with nodal metastasis. No evidence of distant organ involvement was observed.

Magnetic resonance imaging (MRI) was subsequently performed for further characterization. It revealed a lobulated solid lesion in the right hemipelvis, appearing isointense on T1 and heterogeneously hyperintense on T2 with areas of restricted diffusion (Figure 3). Right ovary could not be visualized separately from the lesion. Uterus was displaced to the left. The left ovary was visualized distinctly and measured 1.5 x 1.0 cm. Minimal hemoperitoneum was noted. It was also compressing the urinary bladder and the right ureter, causing moderate hydroureteronephrosis. Additionally, multiple enlarged heterogeneous pelvic and paraaortic lymph nodes were noted.

Considering the radiological features, malignant ovarian germ cell tumor with multiple metastatic lymph nodes was considered. In view of acute presentation, absent vascularity on colour Doppler and presence of intraperitoneal hemorrhagic fluid, possibility of associated adnexal torsion was also considered.

Management

Considering the possibility of adnexal torsion, emergency laparotomy was performed. Intraoperatively, a solid right adnexal

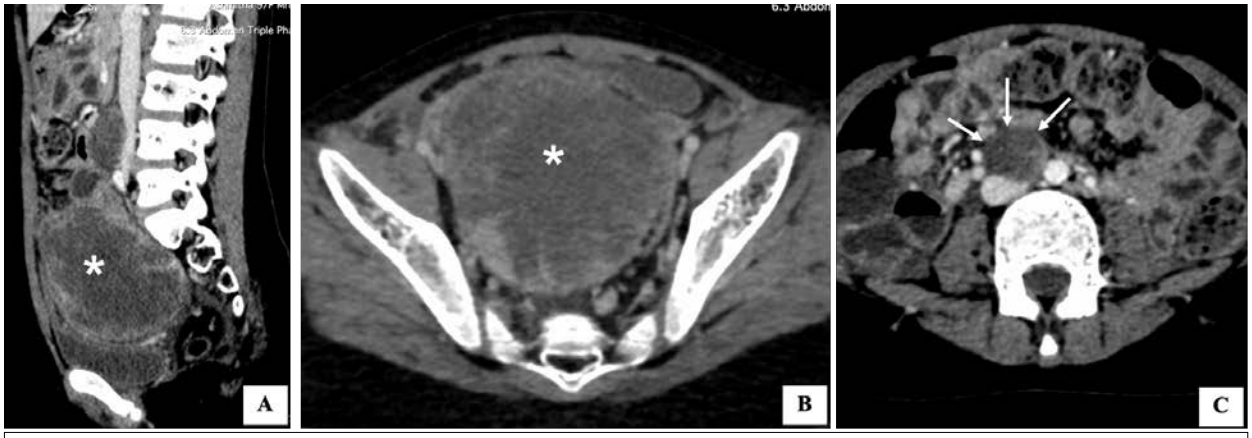

Figure 2: *Sagittal reformatted (A) and axial (B and C) contrast enhanced CT images show a heterogeneous lesion in the pelvis (asterisk) with enhancing solid areas, predominantly in the periphery. An enlarged heterogeneously enhancing paraaortic lymph node is also noted (white arrows).*

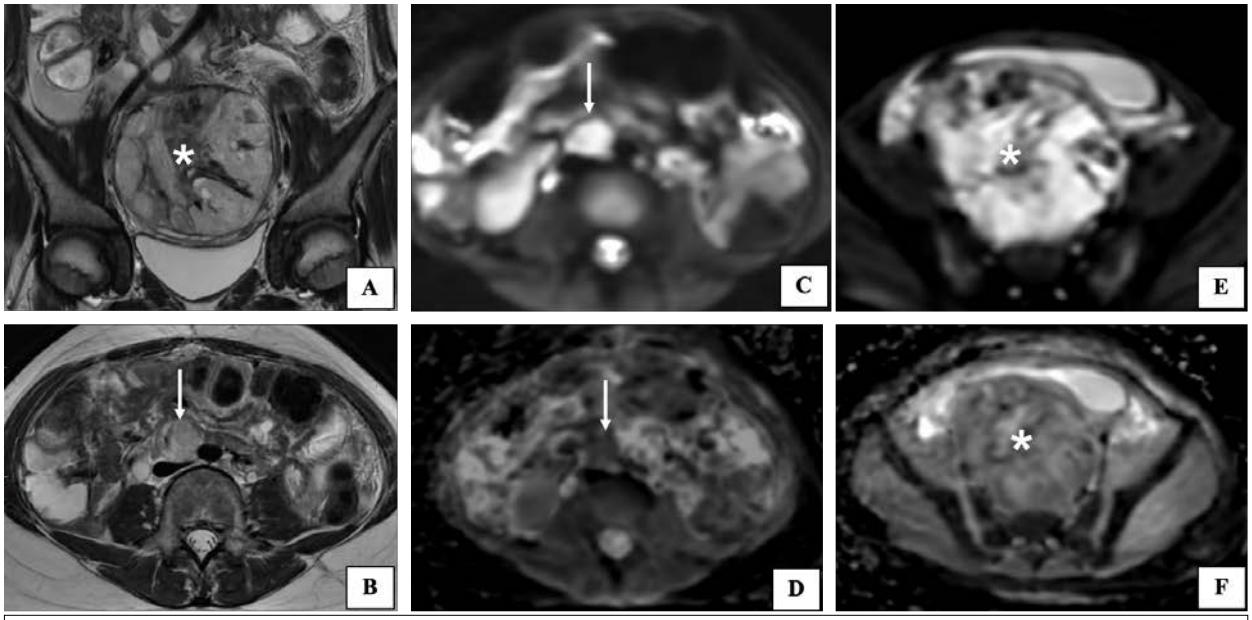

Figure 3: *MRI Coronal T2 (A), axial T2 (B), axial DWI (C and E) and axial ADC (D and F) images show a heterogeneous solid lesion (asterisk) in the pelvis appearing hyperintense on T2 with internal hypointense fibrous septae and demonstrating restricted diffusion. An enlarged necrotic paraaortic lymph node (white arrow) is also noted with diffusion restriction.*

mass with necrotic lymphadenopathy was observed, complicated by twisted vascular pedicles. Right salpingo-oopherectomy with lymph node dissection was performed.

Histopathological Examination

Histopathological examination (Figure 4) revealed a tumor composed of cells arranged in microcystic, festoon, and glandular patterns. The tumor cells exhibited nuclear pleomorphism and atypia. Hyaline globules were observed, along with many areas showing Schiller-Duval body formation. Extensive areas of hemorrhage and necrosis were noted. Dense inflammatory infiltrate composed of neutrophils was present, and brisk mitotic activity was observed. The ovarian capsule was ruptured. The fallopian tubes were also infiltrated by tumor cells. The tumor cells showed Periodic Acid Schiff (PAS)- positive hyaline globules and strong immunoreactivity to Alpha-FetoProtein (AFP) and Sal-like protein 4 (SALL4). These findings were diagnostic of malignant yolk sac tumor. Para aortic and Pelvic lymph nodes showed evidence of metastatic involvement.

Discussion

One of the most common causes of Right lower quadrant (RLQ) pain in children is acute appendicitis, which typically presents with periumbilical pain migrating to the RLQ,

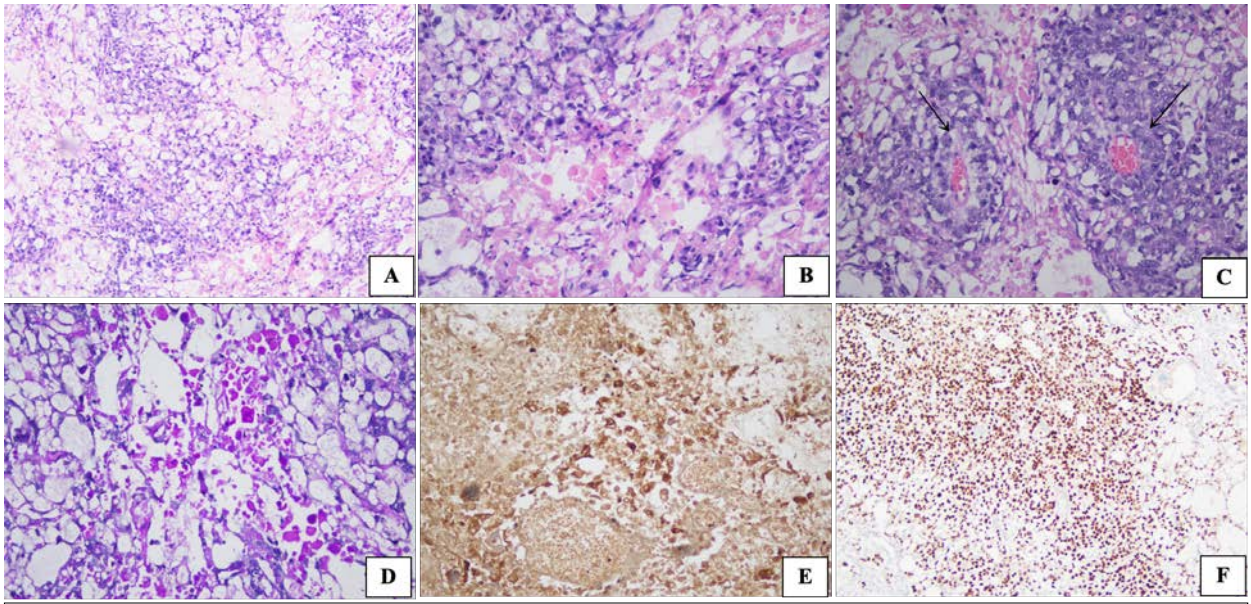

Figure 4: *Microscopic images representing the histomorphology and immunophenotyping of the tumor. Histopathological images with hematoxylin and eosin (H&E) stain in 10x (A) and 20x (B and C) magnification show tumor cells arranged in microcystic and glandular configuration, demonstrating an eosinophilic cytoplasm, nuclear pleomorphism and cellular atypia. Multiple Schiller-Duval bodies (black arrows in C) characterized by a central vessel with surrounding sheets of tumor cells are noted. Periodic Acid Schiff (PAS) stain (D) highlights the presence of hyaline globules. The tumor cells show positive immunoreactivity to AFP (E) and SALL4 (F).*

accompanied by fever, anorexia, and nausea. (6). Ultrasonography is often the first-line imaging modality, with sensitivity ranging from 75-90% (7). Another consideration is mesenteric adenitis, which mimics appendicitis but often follows a viral upper respiratory infection. It is more common in younger children and is usually self-limiting (8). Other important causes include intussusception, which presents with intermittent abdominal pain and bloody stools ("red currant jelly stool"). It is most common in infants and toddlers and requires emergent ultrasound or air enema for diagnosis and treatment.

In female children, especially adolescents, gynecologic causes must be considered. Ovarian torsion is a surgical emergency and typically presents with sudden onset of severe RLQ pain, often associated with nausea and vomiting. Ultrasound with Doppler is the diagnostic imaging modality of choice, though surgical exploration may be required in equivocal cases (9).

Another important consideration is ovarian cyst rupture, which may cause acute RLQ pain and free fluid visible on imaging. Additionally, Mittelschmerz, or mid-cycle ovulation pain, may present as self-limited RLQ pain in pubescent girls (10). For sexually active adolescents, pelvic inflammatory disease (PID) and ectopic pregnancy are critical to rule out. A urine or serum pregnancy test is essential in this population to exclude ectopic pregnancy, even if the patient denies sexual activity (11). Urinary tract infections (UTIs) and pyelonephritis are common in girls and may present with RLQ pain, dysuria, fever, or flank pain. A urine analysis and urine culture are critical in evaluating these conditions. Kidney stones can also present with RLQ pain radiating to the groin, though they are less common in children (12).

Radiological imaging plays a critical role in the evaluation of adnexal lesions in female children, guiding both diagnosis and management. Ultrasound (US) is the first-line imaging modality due to its non-invasive nature, lack of ionizing radiation, and high sensitivity in characterizing adnexal masses. Gray-scale ultrasound can identify simple cysts, complex cysts, or solid masses, while Doppler imaging assesses vascular flow to detect conditions such as ovarian torsion, which presents with reduced or absent blood flow to the affected ovary (13). Functional ovarian cysts, the most common adnexal lesions in children, typically appear as simple, thin-walled, anechoic cysts, whereas neoplastic lesions may have mixed echogenicity or solid components. Color Doppler studies integrated with ultrasound can enhance diagnostic accuracy in emergencies like ovarian torsion, as normal vascular flow

on Doppler can help rule out torsion with high sensitivity.
For complex or indeterminate masses, magnetic resonance imaging (MRI) is preferred for further evaluation due to its superior soft-tissue resolution and ability to characterize the lesion's origin and composition (e.g., fat, fluid, or solid). MRI is particularly helpful in distinguishing benign from malignant lesions, such as differentiating mature teratomas from malignant germ cell tumors (14). While computed tomography (CT) is not routinely used in pediatric adnexal lesions due to radiation exposure, it may be employed in cases where malignancy is suspected or for staging purposes, particularly in advanced tumors. CT is also useful in identifying metastatic spread or assessing adjacent organ involvement (13). In summary, a tailored approach combining ultrasound, MRI, and occasionally CT allows clinicians to effectively evaluate adnexal lesions in children while minimizing risks.

Ovarian germ cell tumors (OGCTs) are the most common ovarian neoplasms in children, accounting for 60-80% of all pediatric ovarian neoplasms (15). Unlike in adults, the majority of pediatric ovarian germ cell tumors are malignant (16,17). Germ cell tumors include teratoma (mature and immature), dysgerminoma, yolk sac tumor, embryonal carcinoma and mixed germ cell tumors. These tumors arise from primitive germ cells and include both benign and malignant types. Benign tumors, such as mature teratomas (dermoid cysts) typically consist of mature tissues like skin, hair, and fat. These tumors are often asymptomatic unless complications like torsion or rupture occur. Malignant OGCTs include dysgerminomas, yolk sac tumors, immature teratomas, and mixed germ cell tumors. Dysgerminomas, the most common malignant subtype, are solid and radiosensitive, while yolk sac tumors are aggressive and characterized by elevated alpha-fetoprotein (AFP) levels. Immature teratomas contain immature neural or mesenchymal tissue with malignant potential, and mixed germ cell tumors feature elements of multiple subtype (18).
Serum tumor markers play a pivotal role in the diagnosis, monitoring, and follow-up of ovarian tumors, particularly in pediatric and adolescent patients. Among ovarian germ cell tumors (OGCTs), alpha-fetoprotein (AFP) is the most widely used marker and is elevated in yolk sac tumors (endodermal sinus tumors). AFP levels are both diagnostic and prognostic, helping to monitor treatment response and detect recurrences. Beta-human chorionic gonadotropin (β-hCG) is another crucial marker, often elevated in choriocarcinomas and some mixed germ cell tumors. Dysgerminomas, the most common malignant OGCT, are associated with elevated lactate dehydrogenase (LDH) levels, reflecting their high metabolic activity.
For epithelial ovarian tumors, which are rare in children, markers such as CA-125 may be elevated, though it is more commonly used in adults. Inhibin and estradiol may be elevated in sex cord-stromal tumors like granulosa cell tumors, which are rare but hormonally active, potentially causing precocious puberty.

The utility of tumor markers extends beyond diagnosis to guiding management. Preoperative levels of AFP, β-hCG, and LDH can provide insights into the likelihood of malignancy and the tumor subtype, assisting in surgical planning. Postoperative monitoring of these markers is essential to evaluate treatment response and identify early recurrences. Despite their utility, serum tumor markers must be interpreted alongside clinical findings and imaging, as false positives can occur in certain physiological or pathological conditions (19).

The treatment of germ cell tumors (GCTs) in children involves a multidisciplinary approach that focuses on achieving optimal oncological outcomes while preserving fertility and minimizing treatment-related toxicities. The primary modality for both benign and malignant GCTs is surgical resection. For benign ovarian GCTs, such as mature teratomas, fertility-sparing surgery is the cornerstone of treatment. The goal is to remove the tumor while preserving the unaffected ovary and uterus. Malignant germ cell tumors, such as dysgerminomas, yolk sac tumors, and immature teratomas, require more extensive surgical excision, often accompanied by lymph node evaluation and staging procedures. However, fertility preservation remains a critical consideration during these surgeries (20).

In cases of malignant GCTs, adjuvant chemotherapy is a crucial component of treatment, particularly for advanced-stage or recurrent disease. Platinum-based chemotherapy regimens, most commonly consisting of bleomycin, etoposide, and cisplatin (BEP), are highly effective and have signifi-

cantly improved survival rates in pediatric patients. For localized dysgerminomas, surgery alone may be sufficient, as these tumors are highly radiosensitive. However, chemotherapy is preferred for advanced-stage disease or for tumors like yolk sac tumors that exhibit aggressive behavior (21). Radiotherapy is used sparingly in children to avoid long-term complications, such as infertility or secondary malignancies, and is generally reserved for specific cases of dysgerminomas.

Treatment response is monitored using serum tumor markers such as alpha-fetoprotein (AFP), beta-human chorionic gonadotropin (β-hCG), and lactate dehydrogenase (LDH). Declining marker levels post-treatment indicate effective tumor control, while persistent elevation suggests residual disease or recurrence. Long-term follow-up is essential to monitor for late effects of chemotherapy, including nephrotoxicity, ototoxicity, and secondary malignancies, as well as to assess fertility outcomes. With the advent of modern multimodal treatments, the prognosis for pediatric GCTs is excellent, with 5-year survival rates exceeding 90% for early-stage disease and around 70-80% for advanced disease (22).

Conclusions

This case report highlights the role of imaging in preoperative evaluation, diagnosis and management of ovarian tumors in children. Multimodality imaging involving ultrasonography, computed tomography and magnetic resonance imaging is indispensable in characterizing pediatric ovarian tumors and evaluation of associated complications like tumor rupture, torsion and metastasis.

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Ethical considerations
All procedures performed in this study involving a human participant were in accordance with the ethical standards of the institutional ethical committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

Consent for publication
Informed consent was obtained from the patient's parents to publish the case and use their radiological images involved in the preparation of this manuscript.

Data availability
Data sharing is not applicable to this research article as no new data were created or analysed in this study.

Authors' contributions
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KN: study of concept and design, literature research, manuscript editing
PS: study of concept and design, literature research, manuscript editing
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Declarations

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