Synovial Sarcoma of the Ovary in an Adolescent: A Rare Case Report

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ABSTRACT

Background: *Synovial sarcoma (SS) is a rare and aggressive malignant mesenchymal neoplasm, predominantly affecting adolescents and young adults. Although SS has been documented across virtually all anatomical sites, primary ovarian involvement represents an exceptionally rare occurrence, with only a handful of cases reported in the literature, predominantly in postmenopausal women. The diagnosis is further complicated at atypical sites due to significant histological overlap with other mesenchymal and ovarian neoplasms, rendering accurate diagnosis particularly challenging.*

Case Presentation: *A 14-year-old girl presented with a two-month history of progressive abdominal enlargement. Imaging revealed a large mixed solid-cystic adnexal mass measuring 16.0 × 25.1 × 30.6 cm with associated ascites and bilateral hydropneumothorax. She underwent right salpingo-oophorectomy with adhesiolysis and omentectomy. Initial histopathological examination raised a differential diagnosis of teratoma with sarcomatoid components versus embryonal rhabdomyosarcoma. A subsequent comprehensive IHC panel demonstrated nuclear TLE1 positivity, cytoplasmic BCL2 positivity, focal CD99, and focal EMA expression with negative SMA, leading to a final diagnosis of primary ovarian SS. The patient developed disease recurrence and died prior to initiation of chemotherapy.*

Conclusion: *This case underscores the diagnostic complexity of primary ovarian SS in a paediatric patient and highlights the indispensable role of IHC analysis, particularly TLE1 and BCL2, in securing an accurate diagnosis. Clinicians should maintain a high index of suspicion for SS in young patients presenting with high-grade pelvic masses.*

Key Words: *Immunohistochemistry, Ovarian sarcoma, Synovial sarcoma, TLE1*

INTRODUCTION

Synovial sarcoma (SS) is a rare and aggressive malignant mesenchymal neoplasm that poses significant diagnostic and therapeutic challenges. SS accounts for approximately 5–10% of all soft tissue sarcomas, with an estimated 800–1,000 new cases diagnosed annually in both the United States and Europe.[1] Despite its name, SS is a mesenchymal tumour with partial epithelial differentiation. The reference to synovial is considered a misnomer, as the cell of origin remains uncertain and tumour cells do not originate from the synovium.[2] This malignancy predominantly affects younger populations, and in children, SS is the most common non-rhabdomyosarcoma soft tissue sarcoma, accounting for 26–36% of such cancers.[1] Histologically, SS is classified into three subtypes, monophasic, biphasic, and poorly differentiated, each of which may present with considerable morphological overlap with other mesenchymal tumours, making accurate diagnosis particularly demanding.[3] Recent advances in immunohistochemistry have improved diagnostic accuracy. TLE1, considered the best available immunohistochemical marker to date, demonstrates up to 90% sensitivity, though only moderate specificity, as it may be positive in 15–30% of malignant peripheral nerve sheath tumours and in nearly all mesotheliomas.[4]

Although SS most commonly arises in the extremities near large joints, it has been documented in virtually every anatomical region of the body, including rare visceral sites. Gynaecological primary SS cases are rare but have been reported in the uterus, fallopian tubes, vagina, and vulva. However, ovarian involvement as a primary SS has been reported only a handful of times in the literature.[5] Cases previously described in the literature affected postmenopausal women, and all demonstrated biphasic-type morphology diagnosed based on morphology and immunohistochemistry alone, without identifying the underlying translocation. The rarity of this tumour at an ovarian site, combined with its histological mimicry of more common ovarian neoplasms, renders diagnosis exceptionally challenging.[5,6] The clinical picture is further complicated in the paediatric and adolescent population. Synovial sarcoma accounts for approximately 5–10% of all soft tissue sarcomas, and while 30% of SS cases occur in patients younger than 20 years, delay in diagnosis and treatment is common, as the tumour frequently remains unrecognised for a prolonged period. The management of SS in young patients remains challenging, as wide border resection, radiation, and adjuvant chemotherapy constitute the standard treatment modalities, though the evidence regarding chemotherapy effectiveness in adult patients remains limited and debated.[7] The present case of a 13-year-old girl with primary ovarian SS thus represents a uniquely rare and clinically significant occurrence.

CASE PRESENTATION

A 14-year-old with no prior medical history of hypertension, diabetes, allergies, or asthma presented with a two-month history of progressive abdominal enlargement. She had attained menarche at age 13, with regular menstrual cycles of 28–30 days duration, lasting 5–7 days with normal flow. She was initially evaluated at a regional hospital, where she was hospitalised for five days. Abdominal ultrasonography revealed a solid-cystic mass in the abdominal cavity of unclear origin, suspected to represent a dermoid ovarian cyst,

accompanied by mild bilateral hydronephrosis. She was subsequently referred to Dr. Soetomo General Academic Hospital, Surabaya, with a working diagnosis of suspected intra-abdominal mass with possible dermoid cyst and bilateral hydronephrosis.

On physical examination, the abdomen was distended with a palpable cystic mass reaching the level of the xiphoid process. Vital signs were stable. Body weight was 42 kg, height 141 cm, and BMI 21.1 kg/m². Three days later, the patient returned with worsening dyspnoea and abdominal pain and was admitted for further evaluation and optimisation. Chest radiography demonstrated bilateral pneumonia, bilateral pleural effusion, with no cardiac abnormalities. Bilateral chest tube insertion was performed, resulting in symptomatic improvement. Contrast-enhanced triple-phase CT scan of the abdomen demonstrated a large cystic lesion with minimal solid components measuring approximately 16.0 × 25.1 × 30.6 cm, extending from the pelvic cavity to the abdominal cavity, originating from the adnexa, consistent with a malignant ovarian mass. Ascites and bilateral hydropneumothorax were also noted. Gynaecological ultrasonography confirmed a large cystic mass with solid components exceeding the probe dimensions, with papillary projections, septa, and ascites, originating from the adnexa. Tumour markers examination showed LDH 609 U/L, AFP 1.04 ng/mL, CEA 0.18 ng/mL, and CA-125 229.2 U/mL.

The patient underwent exploratory surgery consisting of right salpingo-oophorectomy, vesicoureteral catheterisation, sharp adhesiolysis, omentectomy, abdominal drain placement, and bilateral double-J ureteric stent insertion. Intraoperative findings revealed a mixed solid-cystic mass measuring approximately 30 cm, densely adherent to the omentum, bilateral abdominal walls, sigmoid colon, and rectum. Sharp adhesiolysis was performed, confirming the origin of the mass from the right ovary. The left tube and ovary, uterus, and omentum appeared grossly normal. A residual mass measuring approximately 10 × 5 × 2 cm was identified, adherent to the posterior uterine wall, pouch of Douglas, and rectum, suggesting infiltration. Intraoperative frozen section yielded findings of a germ cell tumour with haemorrhagic infarction.

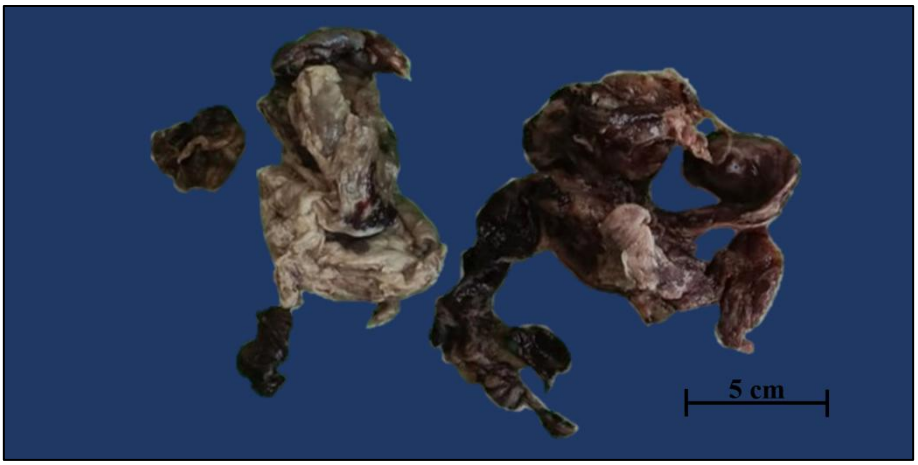

Figure 1. Gross pathological specimen of the right ovarian cyst.

Histopathological examination of the right ovarian cyst specimen with H&E staining demonstrated features raising the differential diagnosis of teratoma with sarcomatoid components versus sarcoma, with suspicion of embryonal rhabdomyosarcoma. Omentum sections showed no evidence of malignancy or tumour infiltration.

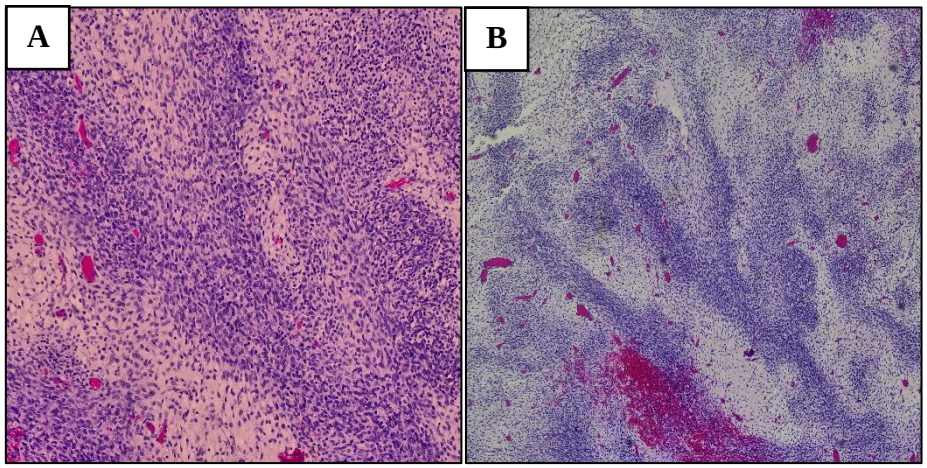

Figure 2 (A, B). Histopathological examination using H&E staining H&E staining showing densely cellular spindle/small round cell tumour.

A post-operative multidisciplinary tumour board review concluded the provisional diagnosis of suspected ovarian carcinoma versus sarcoma, and recommended immunohistochemical (IHC) panel analysis with Desmin and Myogenin to further characterise the tumour type, alongside referral to Paediatric Haematology-Oncology for chemotherapy planning. Immunohistochemical analysis demonstrated the following staining profile. Desmin focal cytoplasmic positivity, Myogenin focal nuclear positivity, SMA negative, TLE-1 positive in tumour cell nuclei, BCL-2 positive in tumour cell cytoplasm, CD99 focal membranous positivity, and EMA focal cytoplasmic positivity. The overall IHC profile was concluded to be most consistent with SS of the ovary.

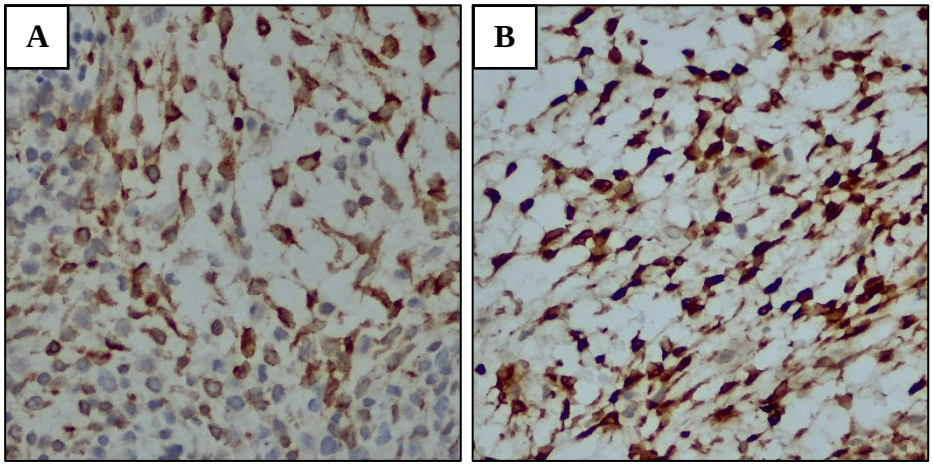

Figure 3 (A, B). Histopathological examination with BCL2 staining showed diffuse cytoplasmic positivity in tumour cells.

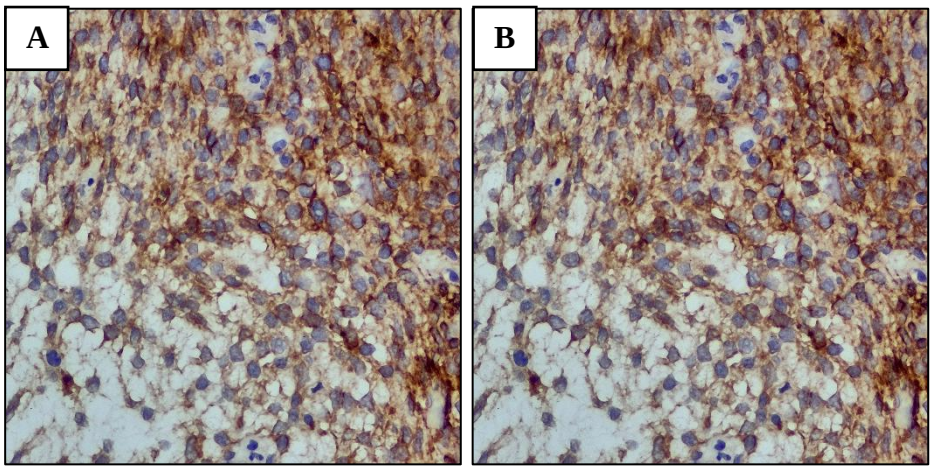

Figure 4 (A, B). Histopathological examination with TLE1 staining showed diffuse nuclear positivity in tumour cells.

The patient then returned to the Paediatric Haematology-Oncology Clinic with recurrent abdominal distension. Her weight had decreased from 42 kg to 35 kg), with blood pressure 95/60 mmHg and heart rate 120 bpm. Pelvic ultrasonography confirmed mixed solid mass exceeding probe dimensions, indicative of disease recurrence. Regrettably, the patient passed away 5 days later, prior to initiation of any chemotherapy regimen.

DISCUSSION

The histopathological diagnosis of SS represents one of the most demanding challenges in surgical pathology, particularly when the tumour arises at an unusual anatomical site such as the ovary. Synovial sarcoma is a heterogeneous tumour of uncertain origin, divided into two major subtypes, monophasic and biphasic. The monophasic subtype may present as either the spindle cell type, which is the most common, or the rarer epithelial subtype, the biphasic subtype contains both fibroblast-like spindle-shaped cells and epithelial cells, while poorly differentiated, ossifying, and myxoid variants have also been described.[8] In the present case, H&E staining of the right ovarian cyst specimen (Figure 2A & 2B) demonstrated a densely cellular neoplasm composed predominantly of small spindle and round cells arranged in fascicular to sheet-like patterns, with interspersed haemorrhagic areas, consistent with a high-grade malignant mesenchymal tumour. Monophasic fibrous SS is typically composed of monotonous solid sheets of small spindle cells with indistinct cytoplasmic membranes and small hyperchromatic nuclei without nucleoli, and may feature staghorn-shaped blood vessels similar to those seen in haemangiopericytoma.[9] The initial morphological impression from the frozen section was interpreted as a germ cell tumour with haemorrhagic infarction, and permanent sections yielded a differential diagnosis including teratoma with sarcomatoid components and embryonal rhabdomyosarcoma, a finding that underscores the well-recognised diagnostic pitfalls associated with SS at unconventional sites. Primary SS in the pelvis is extremely rare with scattered case reports, and the tumour often poses a diagnostic challenge on small biopsy and/or with aberrant immunohistochemical

marker expression.[10] The H&E morphology alone, while suggestive, was therefore insufficient to render a definitive diagnosis, necessitating a comprehensive IHC panel.

The IHC analysis in this case yielded a profile strongly supportive of SS, characterised by positive nuclear staining for TLE1, cytoplasmic positivity for BCL2, focal membranous positivity for CD99, and focal expression of EMA, with negative SMA, collectively consistent with the established IHC fingerprint of this malignancy. Regarding TLE1, a systematic review and meta-analysis by Baranov et al. (2020) demonstrated that the mean sensitivity and specificity of TLE1 in detecting SS were 94% and 81% respectively, with a negative predictive value of 96%, establishing it as one of the most reliable single IHC markers available in this context.[4] As confirmed by the nuclear TLE1 positivity observed in Figures 4, TLE1 is regarded as the most sensitive and specific marker for SS among currently available IHC stains, and its intense diffuse nuclear expression in the present case provided critical supportive evidence.[6,11] Equally important, BCL2 positivity. demonstrated as cytoplasmic staining in Figure 3, further corroborated the diagnosis. BCL2 was positive in 97.3% of SS in a large IHC validation study, reinforcing its role as a reliable constituent of the diagnostic panel alongside TLE1, EMA, MIC2, and CK7.[12]

TLE1 expression was significantly correlated with the pathognomonic translocation t(X;18) in SS, suggesting that in settings where molecular testing is unavailable, TLE1 combined with BCL2 positivity and CD34 negativity may constitute a cost-effective and diagnostically valid immunohistochemical approach.[13] In this case report, while FISH confirmation of the SS18-SSX translocation was not performed, a limitation acknowledged by the authors, the combined H&E morphology, clinical context, and the convergent IHC profile of TLE1 positivity, BCL2 expression, focal EMA, CD99, and negative SMA collectively led the pathology team to conclude the diagnosis as most consistent with ovarian SS. This case further reinforces the critical importance of a thorough and systematic IHC evaluation in atypical-site sarcomas, particularly when conventional morphological assessment alone fails to yield a definitive diagnosis.

CONCLUSION

This case report describes an exceptionally rare presentation of primary SS of the ovary in a 13-year-old girl, highlighting the profound diagnostic challenge this tumour poses when arising at an atypical anatomical site. The diagnosis was ultimately established through a comprehensive IHC panel, in which nuclear TLE1 positivity and cytoplasmic BCL2 expression proved decisive in distinguishing ovarian SS from its histological mimics. The rapid clinical deterioration and fatal outcome prior to chemotherapy initiation underscore the aggressive biological behaviour of this malignancy and the critical consequences of diagnostic delay. Clinicians and pathologists should therefore maintain a high index of suspicion for SS in young patients presenting with high-grade pelvic masses, and timely access to IHC analysis remains paramount in securing an accurate and prompt diagnosis.

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