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Case Report

A rare case of embryonal sarcoma in the liver of a young adult: diagnostic and therapeutic perspectives ☆

Mena Louis, DO[a,*], *Joseph Conway, MD*[b,1], *Maurice Asouzu, MD*[a,1], *Louise Jones, PhD*[c], *Hardeep Singh, PhD*[c], *Nelson A. Royall, MD, FACS*[d,1]

[a] *Northeast Georgia Medical Center, General Surgery department, 743 Spring St NE, Gainesville, GA 30501, USA*
[b] *Northeast Georgia Medical Center, Gainesville Regional Pathology Associates, 743 Spring St NE, Gainesville, GA 30501, USA*
[c] *Northeast Georgia Medical Center Graduate Medical Education Research Department, 743 Spring St NE, Gainesville, GA 30501, USA*
[d] *Northeast Georgia Medical Center, Northeast Georgia Physician Group, Hepatobiliary and Pancreatic surgery, 743 Spring St NE, Gainesville, GA 30501, USA*

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ABSTRACT

Embryonal sarcoma of the liver (ESL) is a rare and aggressive neoplasm primarily affecting children, with its occurrence in adults being exceptionally rare. This case report details the presentation, diagnosis, and management of ESL in a 20-year-old patient, highlighting the challenges and strategic approaches required in managing such atypical presentations. The patient presented with progressive right upper quadrant abdominal pain and significant weight loss, with imaging revealing a large mixed-density mass in the right lobe of the liver. Despite the nonspecific clinical symptoms and normal tumor markers, advanced imaging techniques including MRI and CT scans played a pivotal role in the diagnostic process. The mass exhibited characteristics that led to a differential diagnosis of a possible benign condition; however, the decision for surgical resection was made based on the tumor's rapid growth and potential malignancy suggested by imaging. Histopathological examination postsurgery confirmed the diagnosis of ESL. This case illustrates the importance of considering ESL in the differential diagnosis of rapidly enlarging liver masses in adults, despite its rarity in this age group. The effective management of this case through surgical intervention without prior biopsy, due to the risk of tumor seeding, followed by adjuvant chemotherapy, reflects the critical need for a multidisciplinary approach. The outcomes from this case

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* Corresponding author.
E-mail address: drmenalouis@yahoo.com (M. Louis).
[1] Co author.

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contribute to the existing knowledge base, providing insights into the complexities of diagnosing and treating adult cases of ESL and affirming the adaptability of pediatric protocols to adult patients.

Introduction

Embryonal sarcoma of the liver (ESL), first distinguished as a unique entity from other hepatic sarcomas by Stocker and Ishak in 1978, remains a rare but aggressive neoplasm that predominantly affects children aged 6 to 10 years [1]. Despite its established presence in pediatric oncology, ESL is seldom seen in adults, making each adult case a critical opportunity for expanding current understanding and enhancing diagnostic and therapeutic strategies [2].

This malignancy presents considerable challenges due to its nonspecific clinical and radiological manifestations, often mimicking less aggressive hepatic conditions and thereby complicating timely and accurate diagnosis [3]. Typical presentations can range from asymptomatic masses discovered incidentally to rapid abdominal enlargement accompanied by pain, weight loss, and varying degrees of systemic symptoms [4].

This report details the diagnostic and management journey of an unusual adult case of ESL, typically a pediatric malignancy. By elucidating the comprehensive diagnostic process and management strategies from initial symptom presentation to postoperative care, this case offers valuable insights into ESL's clinical spectrum [5]. It highlights the importance of a thorough differential diagnosis and provides a practical narrative that benefits not only radiologists, but the entire multidisciplinary team involved in oncologic care, offering strategies to effectively manage similar cases in the future.

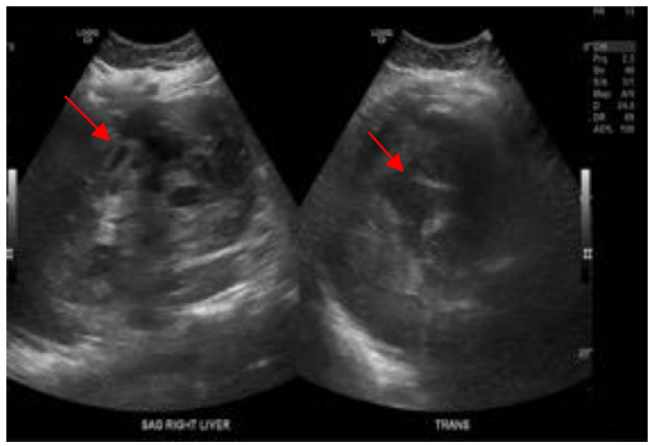

Fig. 2 – RUQ ultrasound showing complex solid-cystic mass within the right lobe of the liver (red arrows). No evidence of internal blood flow sonographically.

Case presentation

A 20-year-old patient with no significant past medical history presented to the emergency department reporting a 3-month history of progressive, dull, achy pain in the right upper quadrant (RUQ) of the abdomen, which radiated to the right flank. Accompanying symptoms included postprandial nausea, fever, chills, diarrhea, hematochezia, or melena. Over

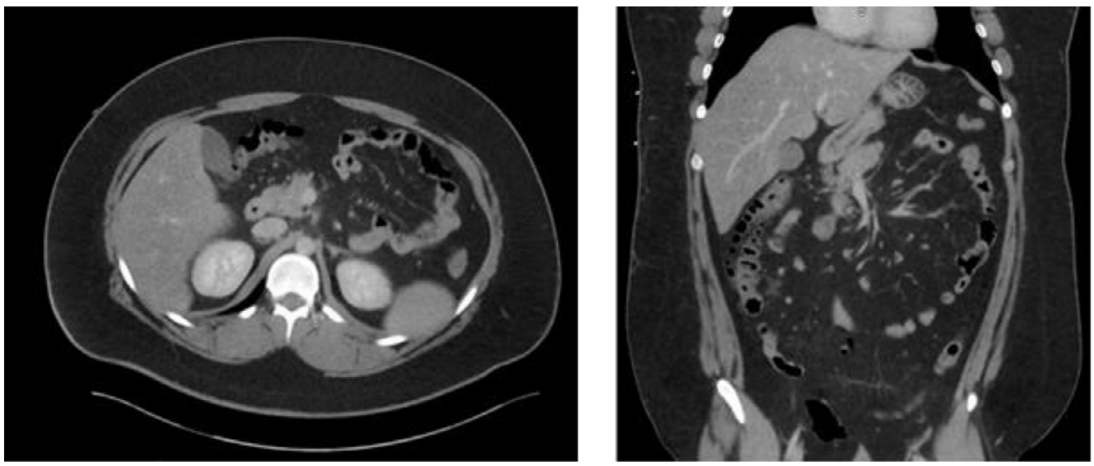

Fig. 1 – Axial and Coronal CT Obtained 22 months before presentation, with no evidence of liver masses.

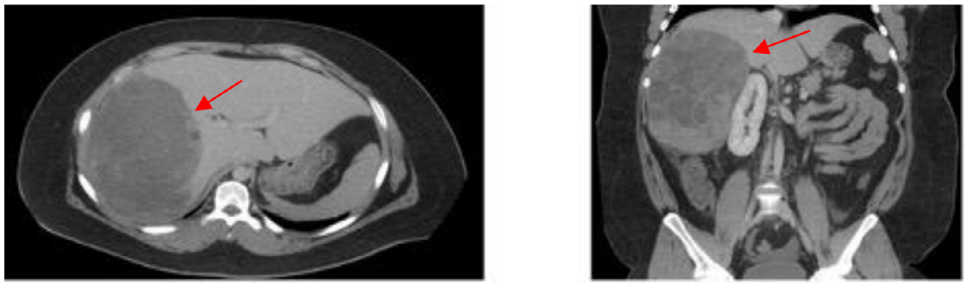

Fig. 3 – Axial left and Coronal right CT revealing 20.4 x 18.7 x 13.2 cm right hepatic mass (red arrows).

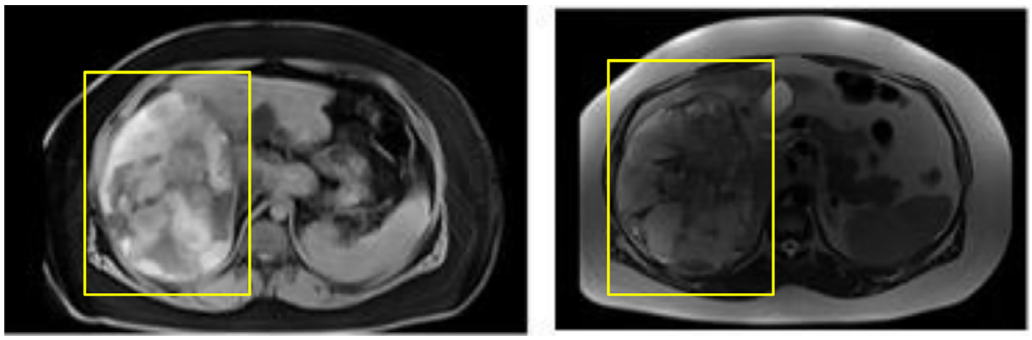

Fig. 4 – MRI T1 on the left and T2 on the right showing large hepatic mass with complex internal fluid collections and some solid components (yellow rectangles).

this period, the patient noted a weight loss of approximately 27 pounds.

Upon examination, the patient, who was obese with a body mass index (BMI) of 41.4, exhibited a tender, palpable mass in the RUQ. Initial laboratory tests and tumor markers did not show significant abnormalities, aligning with the often-elusive biomarker profile of ESL.

Based on the patient's past medical records, a computerized tomography (CT) scan with IV contrast of his abdomen and pelvis done approximately 22 months before presentation was completely normal with no evidence of any intra-abdominal masses as seen in Fig. 1.

An ultrasound was initially performed and revealed a solid cystic tumor in the right lobe of the liver (Fig. 2). Subsequent imaging with a CT scan of the abdomen and pelvis with IV contrast confirmed the presence of a large mixed-density mass involving the right lobe of the liver (Fig. 3). This mass measured 20.4 cm in vertical length and approximately 18.7 × 13.2 cm transversely, extending from the diaphragm and displacing the left lobe.

MRI further characterized the mass as large and complex, primarily involving the right lobe with internal heterogeneity and encapsulation (Figs. 4 and 5). Given the lesion's characteristics and the lack of enhancement in parts of the mass, a preliminary differential diagnosis considered was biliary cystadenoma.

This complex presentation was extensively discussed at a multidisciplinary tumor board, which opted against biopsy due to the risk of seeding and decided on surgical resection

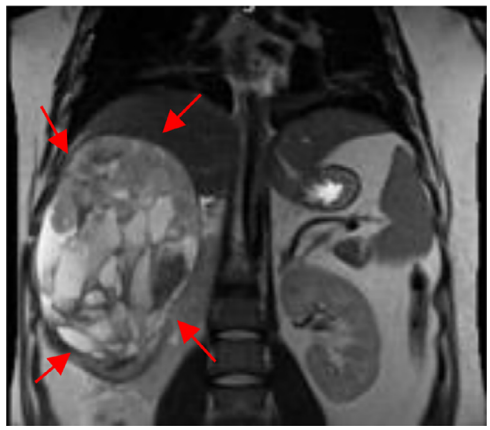

Fig. 5 – T2 Coronal MRI showing hepatic mass with complex internal components (red arrows).

based on the imaging findings and clinical presentation. The patient underwent surgery, where the mass was resected and sent for pathology (Fig. 6), which ultimately confirmed the unexpected diagnosis of embryonal sarcoma.

Histopathological studies (Figs. 7–9) revealed a pleomorphic population of rounded, ovoid, polygonal, and large bizarre multinucleate cells having amphophilic cytoplasm and atypical vesicular nuclei with readily identified mitotic figures. A subset of the cells contains characteristic eosinophilic hyaline

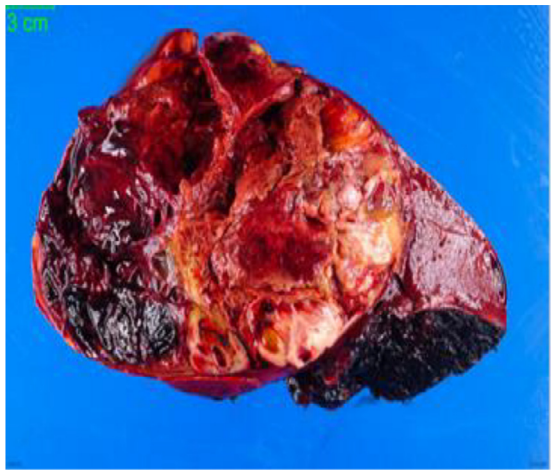

Fig. 6 – Gross picture of the resected mass showing areas of hemorrhage and necrosis.

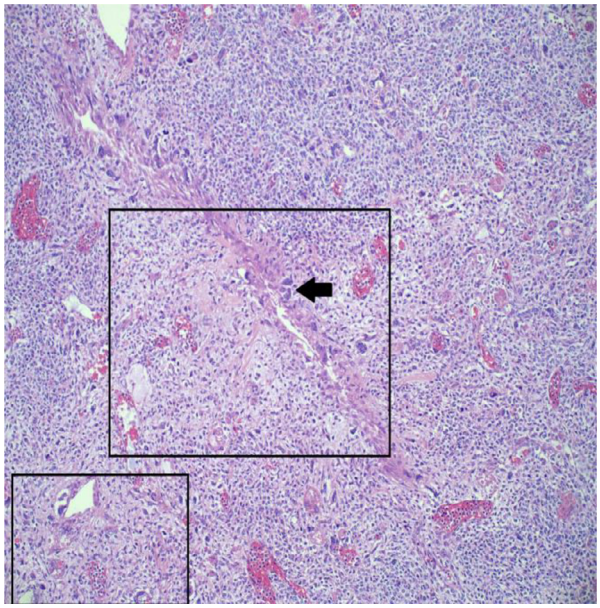

Fig. 7 – ESLP (embryonal sarcoma low power). H&E 100X. The solid black arrow points to a small cluster of giant cells. Rectangles highlight hypocellular myxoid areas that are surrounded by more cellular tumors.

lysosomal droplets. As often seen in lesions of this type, there is multifocal positivity for desmin and Pan-keratin and there is also multifocal positivity for alpha-1-antitrypsin. These pathological findings were suggestive of Embryonal sarcoma of the liver, which was an unexpected finding.

Discussion

Embryonal sarcoma of the liver (ESL), first identified as a distinct entity in 1978 by Stocker and Ishak, is predominantly a pediatric malignancy, with most cases occurring in children between the ages of 6 and 10 years [1]. The diagnosis of ESL in adults, as illustrated by this case, remains exceedingly rare, presenting unique diagnostic and therapeutic challenges due to the disease's aggressive nature and the often ambiguous clinical and radiological manifestations [6].

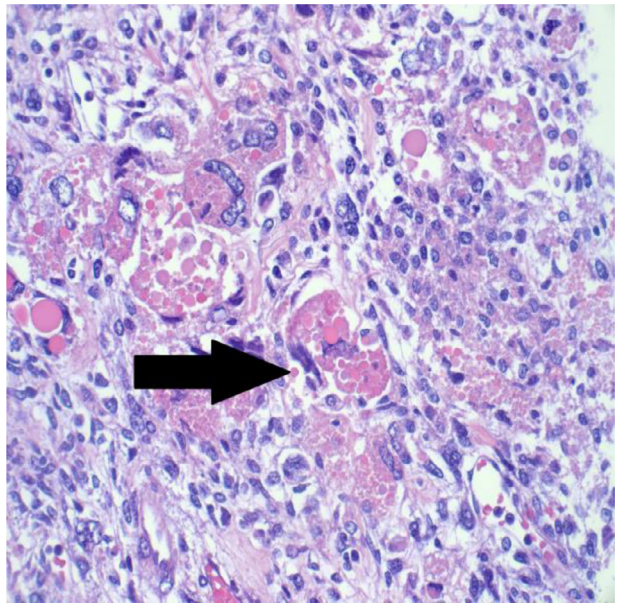

Fig. 8 – Eosinophilic globules. H&E 400X. The solid black arrow points to a single eosinophilic granule within a tumor cell containing multiple eosinophilic granules with multiple similar cells in the background.

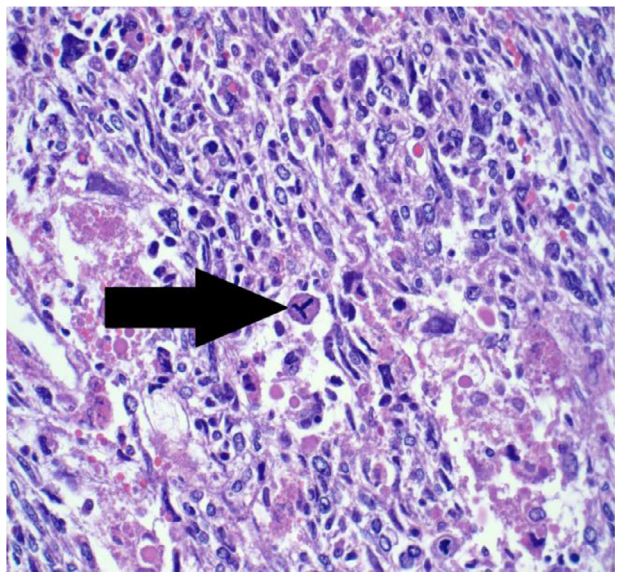

Fig. 9 – Tripolar mitosis. H&E 400X. The solid black arrow points at abnormal tripolar mitosis.

The initial presentation of ESL can be deceptively nonspecific, mimicking more common hepatic pathologies, which often lead to diagnostic delays [7]. In this case, the patient's symptoms of abdominal pain and weight loss were initially suggestive of more common etiologies such as a benign hepatic cyst or biliary disease [2]. This is compounded by the lack of specific tumor markers for ESL, as reflected in the normal levels of alpha-fetoprotein, CEA, and CA 19-9 observed in our patient [8]. The utilization of advanced imaging techniques played a critical role in the diagnostic process, with MRI providing detailed characterization of the lesion that was not possible with ultrasound or CT alone [1,9]. However, even with sophisticated imaging, the definitive diagnosis was only possible through surgical resection and subsequent histopathological examination, highlighting the essential role of histopathology in confirming ESL [10,11].

The management of ESL typically involves a combination of surgical resection and chemotherapy, with the goal of achieving negative margins and addressing any potential metastatic disease [8]. In this case, the decision to proceed directly to surgery without a prior biopsy was based on the multidisciplinary team's assessment of the imaging characteristics suggestive of malignancy and the potential risks associated with biopsy, such as tumor seeding [9,12]. This approach highlights the importance of a tailored strategy in managing complex liver tumors, where the risks and benefits of each diagnostic and therapeutic step must be carefully weighed [5,13].

This case contributes to the limited but growing body of literature on adult ESL and highlights several areas for further research [4]. Developing more sensitive diagnostic tools, including biomarkers and imaging modalities that can differentiate ESL from other liver masses at an earlier stage, remains a critical need. Additionally, exploring the genetic and molecular underpinnings of ESL may provide insights into targeted therapies that could improve outcomes for these patients.

Conclusion

This case report details the diagnosis and management of embryonal sarcoma of the liver (ESL) in an adult—a presentation rare enough to pose substantial diagnostic and therapeutic challenges. Through this patient's journey, it becomes evident that ESL, while typically a pediatric malignancy, can occur in adults with presentations that may obscure timely diagnosis. Key to managing such cases is a thorough diagnostic workup leveraging advanced imaging techniques and a decisive surgical approach aimed at complete resection. This case emphasizes the critical nature of considering ESL in differential diagnoses for rapidly growing liver masses in adults, demonstrating that pediatric treatment protocols can be effectively adapted for adult patients to achieve favorable outcomes.

Patient consent

We confirm that we have obtained written, informed consent from the patient for the publication of this case report. The patient has been thoroughly informed about the details that will be published and understands the implications of the publication. The written consent is stored securely and is available for review by the editorial team upon request.

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