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Unique manifestation of primary renal rhabdomyosarcoma in patient with autosomal polycystic kidneys: Case report and review of literature

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ABSTRACT

Primary embryonal rhabdomyosarcomas of the kidney are extremely rare, especially in adults. The presented case, a 32-year-old female with a background of autosomal polycystic kidney disease, was initially referred with a left hemorrhagic renal cyst. Despite angioembolization, she eventually underwent radical nephrectomy which revealed the diagnosis of embryonal rhabdomyosarcoma. The diagnosis and presentation of this case is unique as she presented with hemorrhagic renal cyst. Adult renal rhabdomyosarcoma has a poor prognosis, as shown by other reported cases. Unfortunately, She passed away 88 days post operatively due to disease progression.

1. Introduction

Rarely reported in the literature, renal rhabdomyosarcomas develop from skeletal muscle progenitor cells and are a subtype of renal mesenchymal tumors.[1] In adults, primary rhabdomyosarcomas occur quite seldom. It is difficult to offer concrete suggestions for diagnosis and treatment based on the limited number of instances reported in the literature. However, it is evident that, like other primary sarcomas, embryonal rhabdomyosarcoma (ERMS) is aggressive and presents late. Since primary ERMS is so uncommon in adults, there is little published data to help with diagnosis and treatment.

2. Case presentation

The patient is a 32-year-old woman who has a family history of polycystic kidney disease. She was referred from a local hospital with a hemorrhagic left renal cyst, which received one packed red blood cell. At presentation, vital signs were stable. The abdomen was distended, and the left kidney was palpable, reaching the midline of the abdomen. However, the abdomen was soft without signs of peritonitis. Laboratory workup showed a hemoglobin level of 9.6 g/dl and a creatinine level: 64 μmol/l. Enhanced Computed Tomography (CT) of the abdomen demonstrated features of polycystic kidneys, an enlarged left kidney with a hemorrhagic component without associated extravasation, the images was discussed with intervention radiologist confirmed no signs of active bleeding. After making sure the stability of the patient, she was discharged home and to be seen, followed as an outpatient. She was seen in the clinic 10 days later and was still symptomatic with abdominal pain. On examination, the abdomen was distended more.

As a result, the decision was made to admit her electively for angioembolization. Repeated enhanced CT showed a left complex renal cyst with thick septation with enhancement with contrast extravasation suggesting active bleeding (Fig. 1A–B). Angiogram was warranted, which showed no evidence of extravasation and, in addition, there were significant tortuous arteries in the lower pole that were embolized (Fig. 2). Eventually, increasing in the abdominal distention with a dropping of hemoglobin even after angioembolization necessitated surgical intervention to evacuate the hematoma as the patient was symptomatic. Intraoperatively, there was a capsulated mass around 30 cm crossing the midline with bleeding around it. Thereafter, radical nephrectomy was done without clear spillage of the tumor complicated by bowel injury which was repaired primarily intraoperative by general surgery team. Post operatively, the patient developed chyle leak which was managed conservatively. Unexpectedly, histopathology revealed the diagnosis of embryonal rhabdomyosarcoma (Fig. 3A–D). Her case was discussed in the tumor board and was planned for chemotherapy. She had multiple peritoneal metastatic deposit, liver and bone metastasis. She received first cycle of Dactinomycin, Vincristine,

Abbreviations: ERMS, Embryonal Rhabdomyosarcoma; CT, Computed Tomography.

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Cyclophosphamide which was complicated by sepsis, pulmonary embolism and profound neutropenia requiring intensive care unit admission. Unfortunately, she passed away 88 days post-operatively due to disease progression.

3. Discussion

Clinical and radiological detection of primary renal sarcoma in individuals with RMS is notoriously challenging. Most sarcomas seem identical on imaging tests to renal cell carcinoma.[1] According to Grignon et al., the diagnostic criteria for renal sarcomas consist of three parts. To rule out metastasis, sarcoma must first be excluded as a differential diagnosis. Second, the tumor must be sampled appropriately to rule out an epithelial component and therefore rule out the possibility of a sarcomatoid RCC. Histological evidence now rules out the possibility of subsequent renal invasion by a retroperitoneal sarcoma.[2]

The presented case had autosomal polycystic kidney disease and was referred to with a hemorrhagic renal cyst. On examination, the abdomen was distended and palpable left kidney, and the patient's hemoglobin level of presentation was 9.6 g/dl and the patient's creatinine level: 64 μmol/l. Fang et al. documented a case with comparable symptoms in their literature analysis, including nausea, vomiting, and discomfort in the flanks.[3] Fanous et al. described a patient with an unusual presentation: a 37-year-old female with a 6-month history of increased fatigue, weight loss, right flank discomfort, and hematuria.[1] A 37-year-old woman with primary renal ERMS was reported by Fanous et al., in 2012. Her symptoms of weight loss, right flank discomfort, lethargy, and excessive urination had been going on for six months prior to her presentation. The right kidney was replaced by a massive, heterogeneous mass measuring 16 cm (cm) on CT of the abdomen. Pathology results from a patient who had a right radical nephrectomy revealed myogenin-positive and MyoD-positive tissue. Patient was disease-free for 4 months after surgery without any adjuvant therapy.[1] A case report of a 33-year-old woman with extensive hematuria and a primary kidney tumor diagnosed as the botryoid type of RMS was reported in 2017 by Allameh et al.[4] The described subject died during treatment due to illness progression and postoperative complications that resulted in sepsis. Other reported cases show that the prognosis for adult renal RMS is generally poor.

On histology, ERMS resembles embryonal skeletal muscles in varying stages of differentiation. It is arranged in sheets of hypocellular areas with loose myxoid matrix and hypercellular areas with differentiating rhabdoid cells. Perivascular condensation is usually present within the hypocellular areas. Tumor cells may be spindled, stellate, or small and round with scant eosinophilic cytoplasm and marked nuclear atypia. Multipolar mitotic figures are standard. The botryoid variant is a hypercellular area underneath an epithelial surface, the so-called cambium layer. A hypocellular polyploid nodule is also present. ERMS shows skeletal muscle differentiation by immunohistochemistry. Tumor cells must be positive for desmin. Myogenin and MyoD1 are also positive but usually less diffuse than alveolar rhabdomyosarcoma. There is no specific translocation found for ERMS to date. Chromosomal aneuploidies are common, and gain in chromosome 8 is found in about 90% of cases. Other chromosomal gains are 2, 11, 12, 13, and 20. Somatic activation of RAS family genes and germline inactivation of TP53 has been documented.[5]

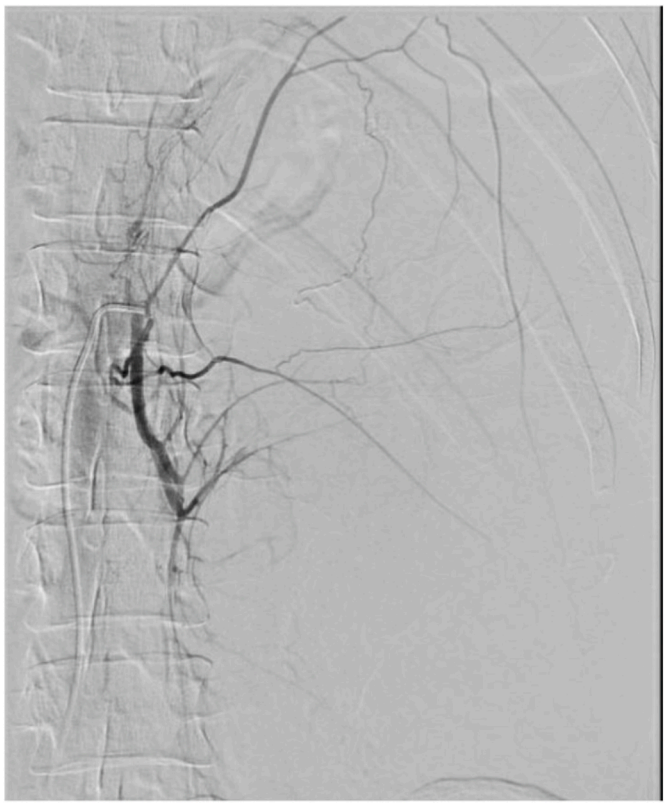

Fig. 2. Angiogram showed no extravasation, torties blood supply.

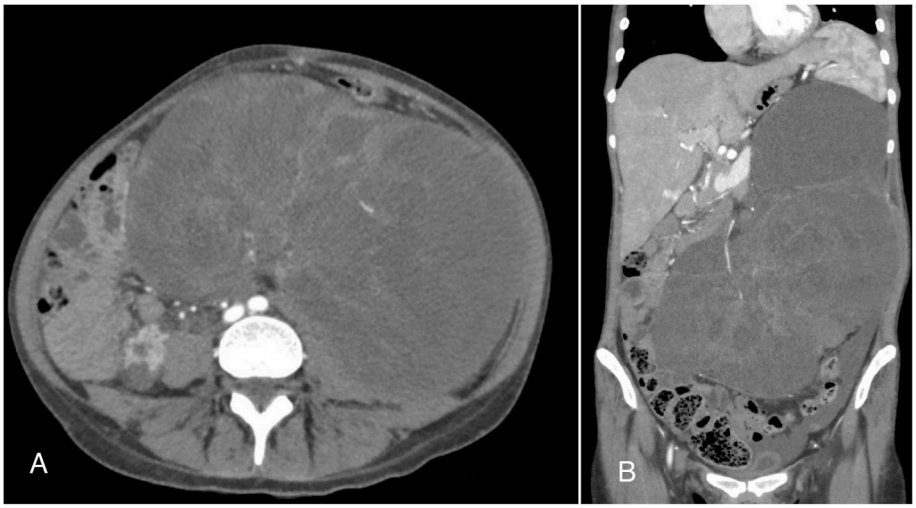

Fig. 1. A–B: Left complex renal cyst with thick enhancing septation.

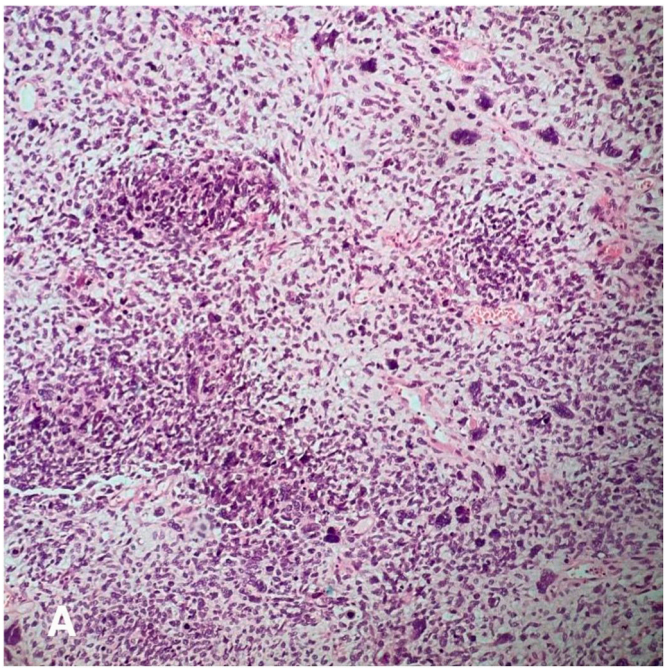

Fig. 3a. Neoplastic cells in embryonal rhabdomyosarcoma show moderate cellularity and are arranged in hypocellular and hypercellular areas with loose stromal backgrounds (200×).

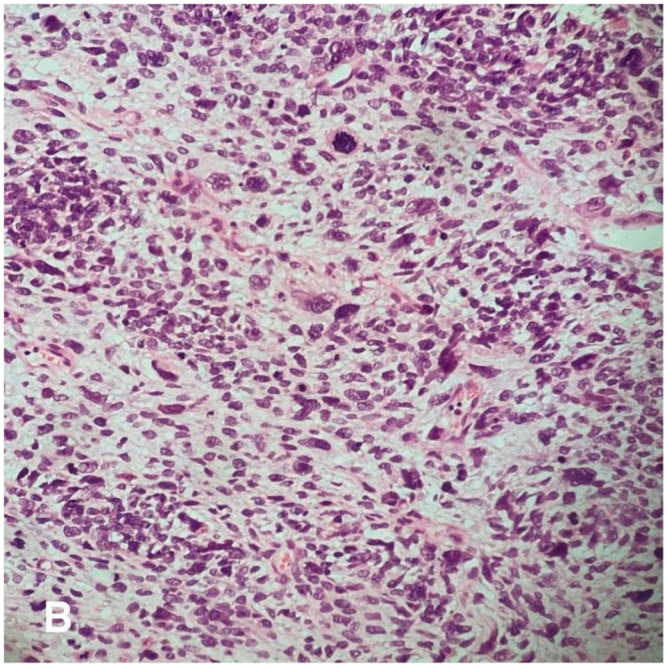

Fig. 3b. Tumor cells are spindled with abundant atypical mitotic figures (400×).

4. Conclusion

Primary renal embryonal rhabdomyosarcoma in autosomal polycystic disease is a rare entity with a poor prognosis in adults. This patient presented with a hemorrhagic cyst, and unfortunately, histopathology revealed an aggressive type of tumor, embryonal rhabdomyosarcoma.

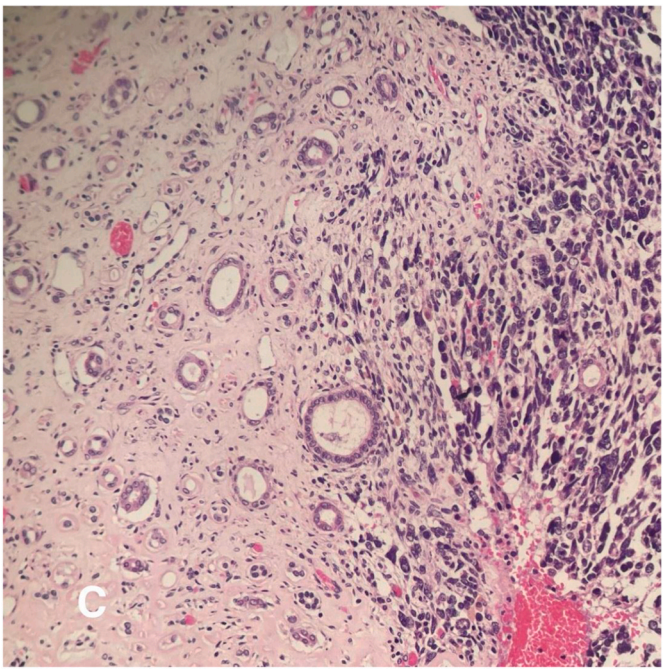

Fig. 3c. Focus of tumor with adjacent uninvolved renal tubules shows high grade anaplastic features.

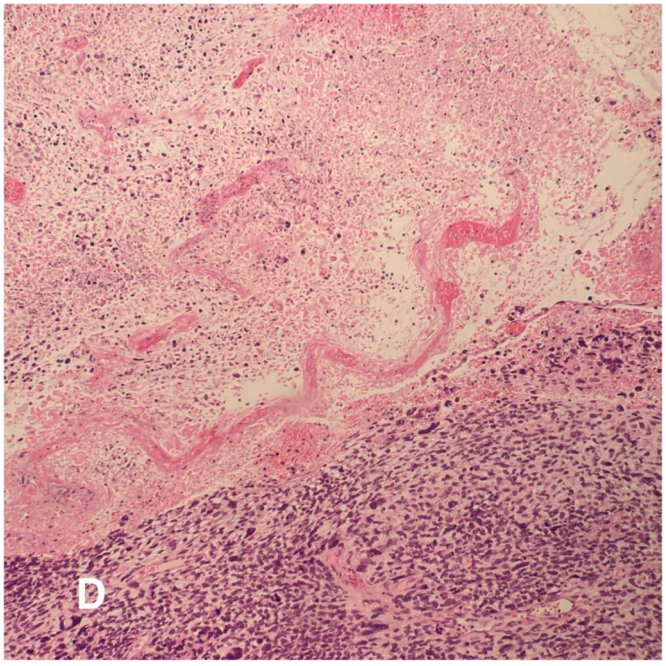

Fig. 3d. Tumor cell necrosis is extensively present.

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Conceptualization, Methodology, Supervision.

Declaration of competing interest

None.

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