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Pediatric hepatic rhabdoid tumor: A rare cause of abdominal mass in children

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ABSTRACT

Pediatric hepatic rhabdoid tumors are rare tumors of the liver, with few cases reported in the literature. These aggressive tumors can be difficult to differentiate from hepatoblastomas on imaging alone, and surgical biopsy combined with special immunohistochemical stains can assist in differentiating these 2 tumor types. We present a case of hepatic rhabdoid tumor in a 7-month-old female infant, which was originally thought to be a hepatoblastoma; however, using BAF47 staining for INI-1 we were able to diagnose a rhabdoid tumor and affect the patient's medical oncologic therapy. Earlier detection and a better understanding of the imaging features of hepatic rhabdoid tumor may aid in improved patient management and treatment planning.

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Introduction

Primary, malignant hepatic neoplasms in pediatric patients are rare, accounting for only 1%-2% of pediatric cancers. Hepatoblastoma is the most common primary malignant hepatic neoplasm, followed by hepatocellular carcinoma, undifferentiated sarcoma, angiosarcoma, and embryonal rhabdomyosarcoma [1]. Malignant rhabdoid tumors (MRTs) are rare, aggressive lesions most commonly found in the kidney but also arising in other soft tissues and the central nervous system. Rhabdoid tumors may be seen in the liver commonly presenting with abdominal distension and hepatomegaly. It may be difficult to distinguish rhabdoid tumors from hepatoblastoma, although the poorer prognosis and differing treatment approaches make early differentiation important.

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As opposed to hepatic MRT, hepatoblastoma typically has an older age at diagnosis (16 months vs 8 months), less frequently undergoes spontaneous rupture with systemic symptoms, and often has an elevated alpha fetal protein (AFP) (90% of cases) [2]. There is also strong evidence suggesting that the prevalence of hepatoblastoma is inversely proportional to birth weight [1]. We present a case of pediatric hepatic rhabdoid tumor, which resembled hepatoblastoma on imaging and required surgical biopsy and immunohistochemical staining for final diagnosis.

Case report

A 7-month-old female infant was transferred from an outside hospital for management of fever in the setting of suspected leukemia. She had a 1-week history of fever with nonbilious, nonbloody emesis, 0.5 lb weight loss, and progressive abdominal distension. At the outside hospital, she was initially diagnosed with a viral infection and prescribed alternating doses of acetaminophen and ibuprofen. The fever appeared to resolve; however, her family noticed cessation of solid food intake, increased irritability, and a slight regression in motor milestones. She was taken back to the emergency department where laboratory results were significant for the following: white blood cells count 29.89 (4-10 × 10^9/L), hemoglobin 5.8 (12-15 g/dL), platelet count 578 (150-400 × 10^9/L), and absolute neutrophil 15,540 (1500-8000/mm^3). The physicians at the outside hospital were concerned about possible leukemia, and she was transferred to our institution for further evaluation.

Additional laboratory results on admission demonstrated reticulocytes 12.88% (0.5%-1.5%), gamma-glutamyl transferase 99 (6-50 U/L), aspartate/alanine aminotransferase 348 and 296 respectively (5-30 U/L), lactatde dehydrogenase 872 (5-150 U/L), and mildly elevated AFP. An abdominal radiograph demonstrated marked hepatomegaly without visible calcification, and abdominal ultrasound revealed a heterogeneous solid and cystic mass in the right hepatic lobe with some demonstrable internal blood flow (Figs. 1-3). The differential diagnosis included hepatoblastoma, mesenchymal hamartoma, and less likely hemangioendothelioma or neuroblastoma. Elevated reticulocytes in combination with anemia made a primary marrow process less likely. Magnetic resonance imaging (MRI) of the abdomen showed a solid and cystic mass with peripheral enhancement in the superior cystic component, including a few enhancing septations, and heterogeneously enhancing solid components. There were multiple fluid-fluid levels noted within the mass on T2-weighted images, which were concerning for hemorrhage. Computed tomography failed to demonstrate calcifications or macroscopic fat within the mass, which favored hepatoblastoma. No metastases were noted on any of the imaging obtained. A surgical biopsy of the mass was performed and pathology showed an undifferentiated small round cell tumor or extrarenal malignant rhabdoid tumor. On immunohistochemical staining it was cytokeratin positive, INI-1 deficient, weakly CD99 positive, and negative for desmin (confirms myogenic tissue origin), CD45 (immune cells), HepPar-1 (sensitive for hepatocellular carcinoma), beta-catenin (fibroblasts), and myogenin. Following biopsy, the patient had a declining hematocrit with concern for hemorrhage into the mass, which resulted in a hepatic arteriogram and polyvinyl alcohol embolization performed by interventional radiology.

Following embolization the patient continued to have fever and electrolyte imbalance likely due to tumor necrosis. Chemotherapy composed of vincristine, doxorubicin, and

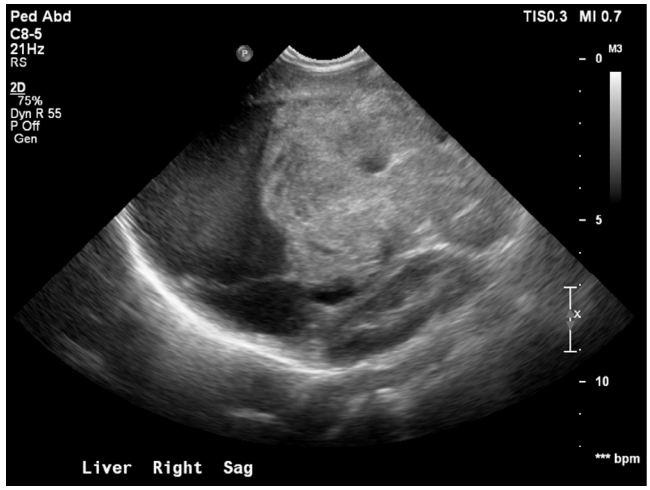

Fig. 1 – Sagittal grayscale image through the right hepatic lobe showing a heterogeneous solid and cystic mass. The contents of the cyst are not completely anechoic due to the presence of hemorrhage into the cyst at the time of diagnosis. Mass effect is noted on the adjacent right kidney

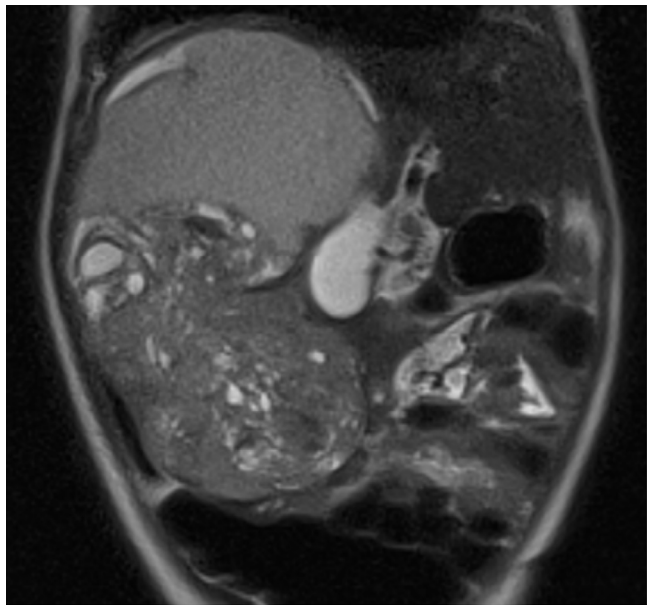

Fig. 2 – Coronal T2-weighted magnetic resonance sequence through the right hepatic lobe. Compared to the normal left lobe, the lesion is relatively T2 hyperintense. A large cystic component superior demonstrates intermediate T2 signal, compatible with the presence of hemorrhage into the cyst. The interior portion of the lesion demonstrates heterogeneous T2 signal with both cystic and solid components

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