

Embryonal rhabdomyosarcoma of the biliary tree: A rare cause of obstructive jaundice in children which can mimic choledochal cysts

Dhara J Kinariwala, Andrew Y Wang¹, Patrick D Melmer, William P McCullough

Division of Pediatric Imaging, Department of Radiology and Medical Imaging, ¹Division of Gastroenterology and Hepatology, Department of Medicine, University of Virginia Health System, Charlottesville, Virginia, USA

Correspondence: Dr. William P McCullough, Department of Radiology and Medical Imaging, University of Virginia Health System, Charlottesville, Virginia - 22908-0170, USA. E-mail: wpm2k@virginia.edu

Abstract

Jaundice in children is more often due to hepatic disease than obstruction. Differential considerations for obstructive jaundice in children include choledocholithiasis, choledochal cysts and rare neoplasms. Rhabdomyosarcoma, the most common soft tissue sarcoma in pediatric patients, typically involves the head and neck, genitourinary system and extremities. Embryonal rhabdomyosarcoma of the biliary tree is a rare entity. We present a 3-year-old boy with abrupt onset obstructive jaundice. Although initial imaging suggested a dilated biliary system with fusiform common bile duct, sludge, and possible cholelithiasis, endoscopic retrograde cholangiopancreatogram (ERCP) diagnosed a common bile duct embryonal rhabdomyosarcoma and further imaging showed involvement of the cystic duct. This case illustrates the importance of considering malignant etiologies in cases of obstructive jaundice, particularly when imaging is not classic for common causes.

Key words: Biliary choledochal cyst; obstructive jaundice; pediatric; rhabdomyosarcoma

Introduction

Obstructive jaundice in pediatric patients beyond the neonatal period can be caused by choledocholiths, choledochal cysts, strictures in the settling of chronic cholangitis and rarely neoplasms such as biliary rhabdomyosarcoma. Rhabdomyosarcoma, a malignancy of skeletal muscle, is the most common soft tissue tumor in the pediatric population, accounting for 1% of malignancies among children ages 0–14 years.^[1] It is rarely found in the biliary tract, where it typically presents with signs and symptoms of obstructive jaundice. The low incidence of biliary rhabdomyosarcoma

makes diagnosis challenging and imaging evaluation particularly important. We present a case of a child with embryonal rhabdomyosarcoma involving both the common bile duct and cystic duct for which initial imaging suggested a choledochal cyst or obstruction due to sludge given a history of hemolytic anemia.

Case History

A 3-year-old boy was transferred from an outside hospital for management of obstructive jaundice. He had a remote

This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 3.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as the author is credited and the new creations are licensed under the identical terms.

For reprints contact: reprints@medknow.com

Cite this article as: Kinariwala DJ, Wang AY, Melmer PD, McCullough WP. Embryonal rhabdomyosarcoma of the biliary tree: A rare cause of obstructive jaundice in children which can mimic choledochal cysts. Indian J Radiol Imaging 2017;27:306-9.

Access this article online

Quick Response Code:



Website:
www.ijri.org

DOI:
10.4103/ijri.IJRI_460_16

history of presumed postinfectious immune-mediated hemolytic anemia as an infant that was reportedly in remission. He presented to the outside hospital with abrupt onset of jaundice, vomiting and fatigue, thought to be related to a pigmented gallstone given his past history. He was constipated and his stools had recently become pale. Imaging obtain from the outside institution included a right upper quadrant ultrasound and contrast-enhanced magnetic resonance cholangiopancreatography (MRCP) showing dilation of extra- and central intrahepatic biliary ducts with debris in the gallbladder, sludge filled common bile and cystic ducts mural thickening involving the cystic and common bile ducts, and a possible cystic duct choledocholith. Upon arrival to our hospital, he was jaundiced with mild, non-tender hepatomegaly. His hemoglobin was normal making a hemolytic process unlikely. His total bilirubin was 9.1 mg/dL and mostly conjugated; transaminases were mildly elevated; alkaline phosphatase and GGT were markedly elevated; lipase was normal and white blood cell count was mildly elevated. A repeat right upper quadrant ultrasound showed findings similar to the outside exam including a fusiform configuration of the common duct. Given this finding, the possibility of a type 1 choledochal cyst was entertained and endoscopic retrograde cholangiopancreatography (ERCP) was planned to clear and if necessary stent the common bile duct.

At ERCP the major papilla was grossly enlarged and bulbously protruded into the duodenal wall. Cannulation of the common bile duct was difficult but achieved and a biliary sphincterotomy was made, immediately after which a grayish translucent mass with faint vasculature was seen protruding from the ampulla [Figure 1]. The common bile duct was then cannulated and a cholangiogram was performed demonstrating a large mass within the dilated common bile duct, dilated cystic, and common hepatic ducts and normal intrahepatic ducts [Figure 2]. The mass was biopsied, which later demonstrated an embryonal rhabdomyosarcoma of the botryoid type on pathologic evaluation [Figure 3]. Biliary stents were then placed, stenting both the cystic duct and common hepatic duct.

Further imaging was performed to better evaluate local extent and facilitate staging. A repeat ultrasound revealed arterial waveforms within intraluminal masses in both the common bile duct and cystic duct [Figure 4]. The masses had been mistaken for echogenic debris and tumefactive sludge on preceding sonograms. An oncology protocol contrast-enhanced magnetic resonance imaging/MRCP (MRI/MRCP) of the abdomen showed asymmetric mural thickening with enhancement and reduced diffusion involving the common bile duct and the cystic duct with an intervening ductal segment with a normal-appearing wall. A mural nodule within the cystic duct had similar imaging features. A peripherally-enhancing T2 hyperintense mass filled the dilated common bile duct [Figure 5]. Positron emission tomography-computed tomography (PET-CT)

showed increased metabolic activity of the thickened ductal wall segments with maximum SUVs of 6.7 [Figure 6]. There was no evidence of distant metastasis on either study. Chest CT showed no evidence of thoracic metastasis.

The patient was discharged with a plan for neoadjuvant chemotherapy including vincristine, adriamycin and cyclophosphamide based on Children's Oncology Group protocol. Surgical intervention would be considered after chemotherapy. Genetic work-up for Li-Fraumeni syndrome (for germline TP53 mutations) was also recommended given the association of soft tissue sarcoma in children with that disease.

Discussion

The differential for obstructive jaundice in children includes choledocholithiasis, choledochal cysts, strictures and rare neoplasms. Imaging plays an important role in defining the etiology with ultrasound typically the first imaging modality. Initial imaging in this case demonstrated a dilated, fusiform common duct filled with apparent debris and resultant obstruction of the biliary tree. These findings excluded stricture and simple choledocholithiasis leaving an imaging differential of choledochal cyst filled with sludge and primary biliary tree neoplasm or primary liver tumor with drop metastases to the biliary tree. A primary liver tumor was excluded given the normal appearance of the liver. Initial ultrasounds failed to show vascularity within the dilated common duct. The initial outside MRI

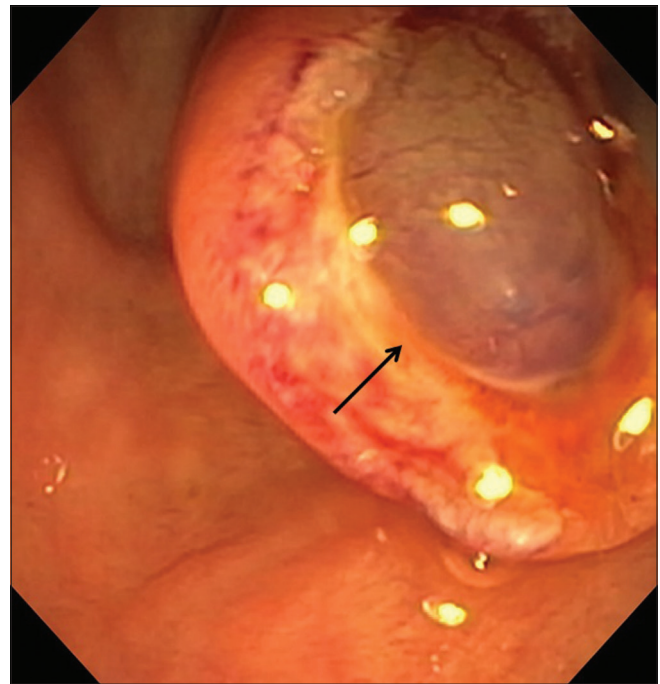


Figure 1: White light endoscopy image. Following biliary sphincterotomy of the bulbous major papilla, a translucent tumor with a faint vascular pattern (black arrow) emerged from the exposed common bile duct. Cold biopsies were obtained from the mass (not shown)

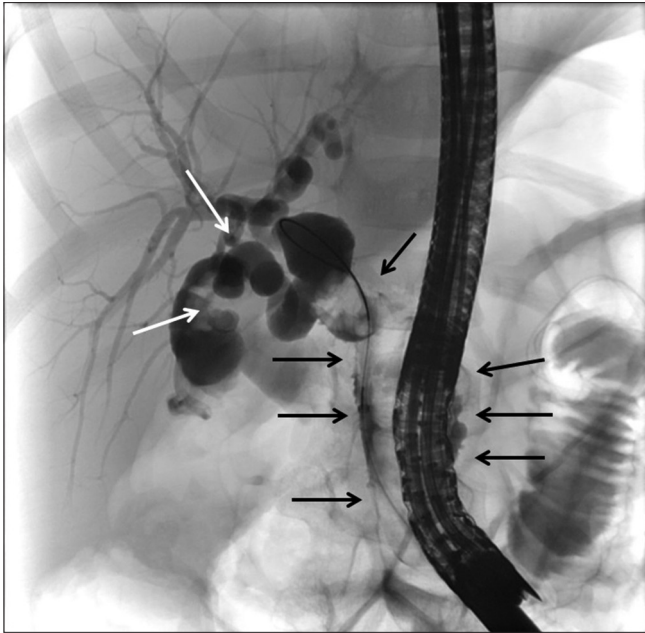


Figure 2: Endoscopic retrograde cholangiogram. The common bile duct (partially obscured by endoscopy scope) is dilated with a large filling defect (black arrows). The cystic duct is dilated and tortuous with a filling defect in the mid portion (white arrows). The central hepatic ducts are prominent

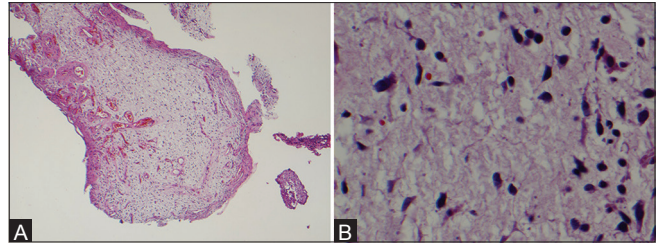


Figure 3 (A and B): Photomicrographs. On low power (A) a polypoid fragment shows myxoid stroma and spindle-cell proliferation. On high power (B) atypical hyperchromatic spindle cells with eosinophilic cytoplasmic projections suggestive of skeletal muscle differentiation

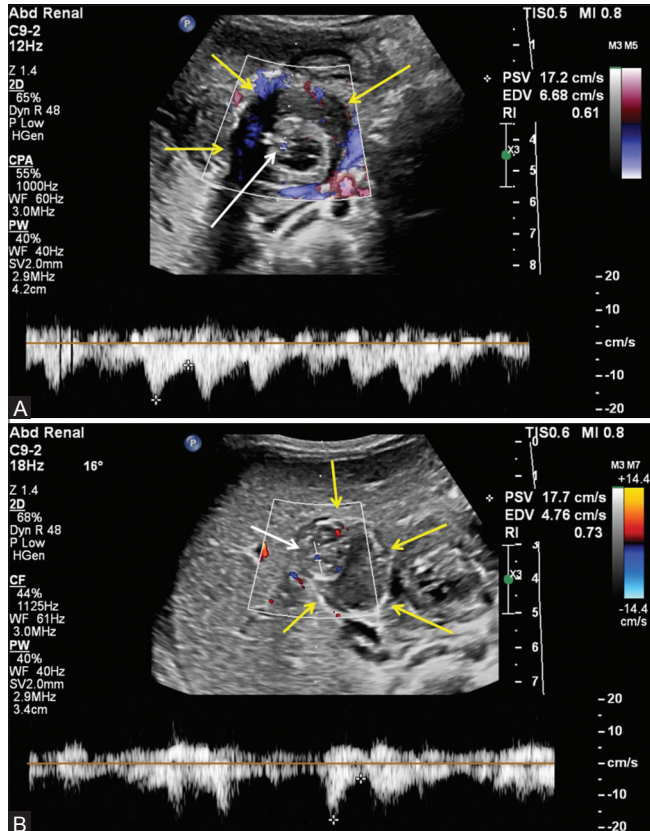


Figure 4 (A and B): US images. Transverse image of the common bile duct (A) (yellow arrows) showing an arterial waveform within the peripheral aspect of the intraluminal mass (white arrow). Transverse image of the cystic duct (B) (yellow arrows) showing an arterial waveform within the cystic duct mural nodule (white arrow)

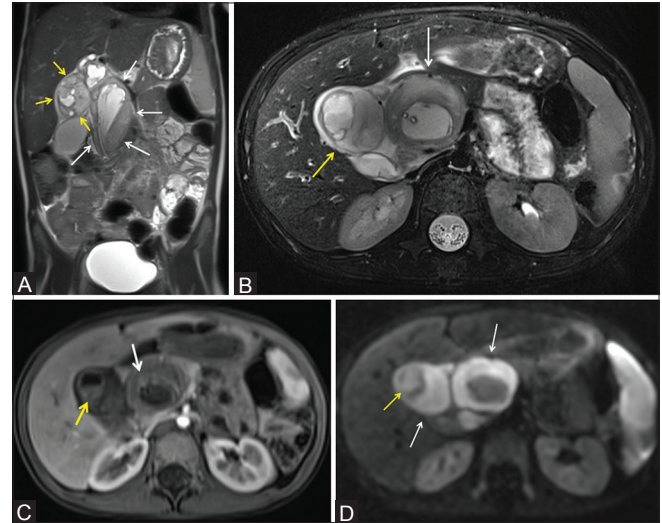


Figure 5 (A-D): MRI images. Coronal T2 (A) with fusiform configuration and asymmetric mural thickening of the common bile duct (white arrows). The adjacent cystic duct demonstrates asymmetric mural thickening with a mural nodule (yellow arrows). Axial T2 fat saturation (B) with asymmetric mural thickening of the common bile duct (white arrow) and adjacent cystic duct (yellow arrow). Two stents are partially imaged. Axial T1 post contrast fat saturation (C) with peripheral enhancement of the common bile duct mass (white arrow) and enhancement of the cystic duct mural nodule (yellow arrow). Axial b800 diffusion weighted (D) showing restricted diffusion of the duct walls (white arrows) and mural nodule (yellow arrow)

failed to demonstrate enhancement of the intraductal material. ERCP was performed to clear debris from the presumptive choledochal cyst at which time the mass was discovered. This case demonstrates the importance of careful Doppler sonography technique. Repeat ultrasound after the ERCP clearly demonstrated arterial waveforms within the intraductal mass. Repeat contrast-enhanced MRI clearly showed an enhancing intraductal mass. The correct diagnosis could have been made by imaging prior to ERCP.

Rhabdomyosarcoma is the most common soft tissue tumor in the pediatric population. Rhabdomyosarcoma of the biliary tract is a rare cause of obstructive jaundice in the pediatric patient. Of all cases of rhabdomyosarcoma in the Intergroup Rhabdomyosarcoma Studies I-IV between 1972 and 1998, only 0.5% of cases (25 of 4291) involved the intrahepatic or extrahepatic biliary tree.^[2,3]

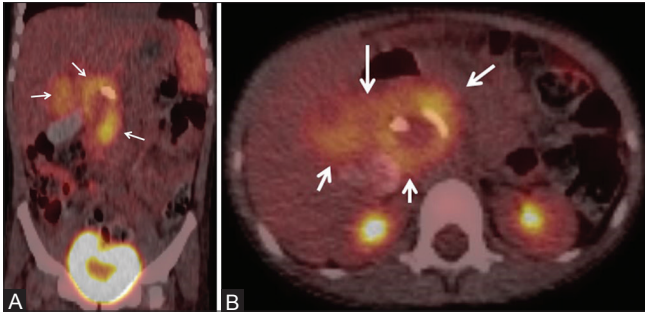


Figure 6 (A and B): PET CT images. Coronal (A) and axial (B) fused PET CT images demonstrate increased metabolic activity of the common bile duct and cystic duct walls (white arrows)

Biliary rhabdomyosarcoma typically presents with obstructive jaundice caused by the tumor, which can be accompanied by abdominal distention and possibly hepatomegaly. Elevated liver enzymes and conjugated bilirubin are frequently seen. Infrequent features include abdominal pain, nausea, vomiting and fever.

Pathologic subtypes of rhabdomyosarcoma include alveolar pleomorphic, and embryonal, which is further subdivided into botryoid and spindle-cell variants. Embryonal is three times more common than alveolar, and the botryoid subtype has the best prognosis.^[3,4] The Intergroup Rhabdomyosarcoma Study Group (IRSG) has developed a modified tumor-node-metastasis staging system that separates patients into low, medium and high-risk groups which are used to guide therapy.^[3] The system considers site of tumor as well as regional nodal and distant metastases. In the absence of distant metastases, as seen in our case, biliary tract rhabdomyosarcoma is considered stage 1, placing our patient in the IRSG low risk group.

Computed tomography (CT) and MRI are typically used to assess tumor extent and characteristics at diagnosis, while ultrasound may be useful to evaluate response to presurgical chemotherapy.^[5] Studies led by the Children's Oncology Group aim to evaluate the use of PET CT to accurately differentiate post-therapy residual benign tumor from viable tumor, which CT and MRI may not be able to assess.^[6]

Ultrasound, typically the initial imaging study in children with obstructive jaundice, shows dilation of the intra- and extrahepatic bile ducts with an intraductal mass. On MRI, these tumors follow the signal of skeletal muscle on T1, are hyperintense on T2 and enhance heterogeneously. Large lesions may demonstrate hemorrhage and necrosis.^[5] The imaging appearance of a biliary tree rhabdomyosarcoma with central tumor necrosis can mimic the appearance a choledochal cyst as was seen with this patient. Choledochal cysts are exceedingly unusual in the cystic duct, with only individual case reports described in the literature,^[7]

suggesting a more complex pathology in our patient. Indeed, previous case reports have remarked that biliary rhabdomyosarcoma is readily misdiagnosed as a choledochal cyst on imaging.^[2]

The surgical management of biliary rhabdomyosarcoma has historically been determined by tumor location and extent, ranging from aggressive resection necessitating choledochojejunostomy or pancreaticoduodenectomy to surgery for staging purposes only via exploratory laparotomy or laparoscopy. Presently, resection of residual tumor following neoadjuvant chemotherapy is advocated and has been associated with good outcomes.^[8]

Biliary rhabdomyosarcoma presents as obstructive jaundice and in tumors with central necrosis, can mimic a choledochal cyst. Diagnostic imaging plays a critical role in the diagnoses and management of this malignancy. Rhabdomyosarcoma should be included in the differential diagnosis for children with obstructive jaundice.

Acknowledgements

Robin D. LeGallo, MD provided the pathology images and descriptions. Jeffrey W. Gander, MD and Sara K. Rasmussen, MD, PhD, provided valuable surgical perspective for this case.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Ali S, Russo MA, Margraf L. Biliary rhabdomyosarcoma mimicking choledochal cyst. *J Gastrointest Liver Dis* 2009;18:95-7.
2. Kumar V, Chaudhary S, Kumar M, Gangopadhyay AN. Rhabdomyosarcoma of biliary tract-a diagnostic dilemma. *Indian J Surg Oncol* 2012;3:314-6.
3. Raney RB, Maurer HM, Anderson JR, Andrassy RJ, Donaldson SS, Qualman SJ, *et al.* The intergroup rhabdomyosarcoma study group (IRSG): Major lessons from the IRS-I through IRS-IV studies as background for the current IRS-V treatment protocols. *Sarcoma* 2001;5:9-15.
4. Parham DM. Pathologic classification of rhabdomyosarcomas and correlations with molecular studies. *Mod Pathol* 2001;14:506-14.
5. Roebuck DJ, Yang WT, Lam WW, Stanley P. Hepatobiliary rhabdomyosarcoma in children: Diagnostic radiology. *Pediatr Radiol* 1998;28:101-8.
6. Malempati S, Hawkins DS. Rhabdomyosarcoma: Review of the children's oncology group (COG) soft-tissue sarcoma committee experience and rationale for current COG studies. *Pediatr Blood Cancer* 2012;59:5-10.
7. Upreti L, Puri S, Verma A, Sethi S. Choledochal cyst of the cystic duct: Report of imaging findings in three cases and review of literature. *Indian J Radiol Imaging* 2015;25:315-20.
8. Pollono DG, Tomarchio S, Berghoff R, Drut R, Urrutia A, Cédola J. Rhabdomyosarcoma of extrahepatic biliary tree: Initial treatment with chemotherapy and conservative surgery. *Med Pediatr Oncol* 1998;30:290-3.