Application of ERCP and Spyglass Technique in Diagnosis of Pediatric Botryoid Rhabdomyosarcoma of the Biliary Duct

Keywords: Biliary rhabdomyosarcoma; ERCP; Spyglass technique

Introduction

Rhabdomyosarcomas account for 40% of all soft tissue neoplasms in the 1-4 years age group and can be difficult to diagnose and treat depending on the location [1]. Biliary involvement is rare and remains a significant diagnostic challenge. New diagnostic modalities are now available to aid in diagnosis. The Spyglass endoscope can be inserted directly into the bile duct, allowing for direct visualization of the bile duct with a more targeted biopsy. We present a case of biliary rhabdomyosarcoma in a 3-year-old female diagnosed via ERCP using novel Spyglass technology. This case demonstrates how a biopsy obtained during cholangiscopy aids in establishing the specific tumor diagnosis and guiding the subsequent treatment course.

Case Report

A 3-year-old female with no prior medical history, presented with a 1-week history of intermittent constipation, fatigue, acholic stools, pruritus, jaundice and severe, non-radiating epigastric pain. Outpatient work up at her pediatrician's office revealed a cholestatic pattern of liver injury with an aspartate aminotransferase (AST) 102 U/L, alanine aminotransferase (ALT) 148 U/L, total bilirubin (Tbili) 1.3 mg/dl and alkaline phosphatase (ALP) 817 U/L. On admission to the emergency department, liver function tests were as follows: AST of 219 U/L, ALT 148 U/L, Tbili 1.3 mg/dl, ALP 731 U/L and gamma-glutamyltransferase (GGT) 570 U/L.

Abdominal ultrasound revealed a soft tissue mass obstructing the common bile duct (CBD), causing intrahepatic and extrahepatic biliary ductal dilatation (Figure 1). Magnetic resonance cholangiopancreatography (MRCP) re-demonstrated the mass, without associated restricted diffusion or definite enhancement. Thus, ERCP with the Spyglass direct visualization system was utilized to obtain a diagnostic tissue sample and to better delineate the extent of the mass within the biliary tree and to obtain a diagnostic tissue sample. ERCP revealed a severe stenosis in the common bile duct (CBD) and Spyglass cholangioscopy showed a multifocal large polypoid mass which was biopsied (Figure 2). Histopathological analysis of the biliary duct biopsy revealed botryoid variant embryonal rhabdomyosarcoma (Figure 3). The diagnosis was confirmed by immunohistochemistry (IHC) with tumor cells positive for desmin and myogenin. Subsequent metastatic workup including bone marrow biopsies and positron emission tomography, were negative. The patient was taken for exploratory laparotomy. The tumor involved the common hepatic duct, with extensive involvement of the right anterior and right posterior hepatic ducts. The left lobe of the liver had a segment 4 duct and a segment 2/3 duct that were emptying separately at the biliary confluence. Therefore, a right hepatectomy (segments 5, 6, 7 and 8) with en bloc resection of the extrahepatic biliary tree (proximal left bile ducts, common hepatic duct, gall bladder and common bile duct) was performed. The left lobe bile ducts were resected as high as possible with grossly negative margins and were then reconstructed to a roux-en-Y hepaticojejunostomy.

Figure 1: Ultrasound showing the CBD intraluminal soft tissue density, thickness of 1 cm.

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of cases (25 of 4291) involved the intrahepatic or extrahepatic rhabdomyosarcoma occurring within the first two decades of life, only conducted between 1972 and 1997, found that of all reported cases of a bunch of grapes. The Intergroup Rhabdomyosarcoma Studies I-IV, the ductal lumen with the characteristic gross appearance resembling leads to protrusion of tumor masses, still covered by the epithelium, into the en-y jejunal limb. The tumor was extensive and was filling the entirety of the biliary tree. Surgically, it was not technically feasible to resect any further margins on the left ducts. Although the resection margins were non-tumoral, since the tumor was filling the entire biliary tree up to the point of transection, we elected to give adjuvant chemotherapy (vincristine, actinomycin D and cyclophosphamide) and radiation. The child was well at last follow-up.

Discussion

Obstruction of the biliary tree carries an extended differential diagnosis. Patients often present with vague symptoms including obstructive jaundice, generalized pruritus, pale stools and darker urine. Epigastric pain can occur due to the biliary obstruction. Labs elevate liver enzymes and hyperbilirubinemia. Embryonal rhabdomyosarcoma is a malignant neoplasm with spindle cell morphology and positive markers of striated muscle differentiation. Rhabdomyosarcoma involving the biliary tree is rare. It usually represents the so-called botryoid variant which is characterized by the growth of tumor cambial layer under the epithelial lining. This growth leads to protrusion of tumor masses, still covered by the epithelium, into the ductal lumen with the characteristic gross appearance resembling a bunch of grapes. The Intergroup Rhabdomyosarcoma Studies I-IV, conducted between 1972 and 1997, found that of all reported cases of rhabdomyosarcoma occurring within the first two decades of life, only 0.5% of cases (25 of 4291) involved the intrahepatic or extrahepatic biliary tree [2]. The rare nature of these lesions makes them difficult to diagnose.

Historically, the diagnosis for this malignancy has been made radiographically. Ultrasound and CT typically show intrahepatic biliary ductal dilatation with an intraductal mass, as times with cystic areas of necrosis [3]. These findings are non-specific and cannot differentiate between biliary rhabdomyosarcoma, a choledocal cyst or a different malignancy [4-7]. The advent of cholangioscopy with Spyglass technology facilitated performing a biopsy for tissue diagnosis, which allows for the optimal planning of surgical, chemotherapy and radiation treatment modalities. This is of particular importance as surgical management for biliary rhabdomyosarcoma is quite extensive and may involve aggressive resection requiring a choledochojjunostomy or pancreaticoduodenectomy. Surgery might also be used strictly for staging purposes via exploratory laparotomy or laparoscopy [8]. Concomitant radiographic information can be used to assess tumor extent. It is clear that a diagnostic and treatment algorithm for these cases is forever altered by the advent of this novel technology [9].

Conclusion

Several cases have been described in the literature of biliary rhabdomyosarcoma in the pediatric patient initially misdiagnosed by radiographic imaging alone. Radiographic studies alone are not sufficient for the diagnosis of such lesions. We postulate that the use of cholangioscopy, such as the SpyGlass Direct Visualization System, although not currently approved for use in children, will become the pediatric population. Increased awareness of this resource and its appropriate application in clinical practice is essential.

References