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Recurrent Gallstones in a Patient with a Congenitally Absent Gallbladder

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ABSTRACT

Gallbladder agenesis is a rare congenital anomaly of the biliary tract, due to failure of the gallbladder and cystic duct budding off of the common bile duct during fetal development. Cholangiocarcinoma (CCA) is a malignant tumor arising from the biliary ducts in patients with underlying chronic biliary tract inflammation, primary sclerosing cholangitis or other diseases. Although few studies have reported cases of cholelithiasis in patients with congenital gallbladder agenesis, there is only one other known case of concomitant cholangiocarcinoma and congenital gallbladder agenesis. Herein we present a 79-year-old male who presented with recurrent gallstones and was diagnosed with gallbladder agenesis intraoperatively with pathology consistent with cholangiocarcinoma.

INTRODUCTION

Congenital abnormalities of the gallbladder can be from hypoplasia or agenesis. Congenital gallbladder agenesis (GA) was first reported in 1701 and is due to failure of the cystic duct and gallbladder to bud off in the fourth or fifth week of fetal development. Often, these anomalies are undiagnosed until the patient becomes symptomatic, or incidentally during surgery. The annual incidence of those who are diagnosed intraoperatively or from imaging obtained due to symptomatic gallstones is 0.007-0.0027%, however up to 0.13% are diagnosed during autopsies. The occurrence in men and women are the same when diagnosed from autopsies, however women tend to be more symptomatic and are thus diagnosed more frequently during their lifetime. Overall, there are approximately 10 to 65 cases per year. There are no known familial links, however GA is associated with other developmental conditions such as Klippel Feil syndrome, malrotation of the gut, horseshoe kidney, aberrant left pulmonary artery, anterior abdominal wall defects, heterotaxy syndrome, polysplenia, and asplenia syndrome. Typically, these patients die from congenital defects other than their GA. GA is also associated with congenital genetic syndromes, such as Trisomy 18. Those who are symptomatic, often present with biliary colic in their second or third decade of life, and are diagnosed intraoperatively, as the diagnosis is originally mistaken for cholelithiasis or cholecystitis. Conditions such as liver fluke infection, primary sclerosing cholangitis, chronic viral hepatitis infections and pancreaticobiliary maljunctions can lead to chronic inflammation, potentially causing cholangiocarcinoma. The risk of cholangiocarcinoma in those with pancreaticobiliary maljunction is more than 285 times higher than in the general population. The incidence of cholangiocarcinoma in GA is not known, as only one case report has ever described this relationship.

Test Name	Value (Reference Range)
Total Bilirubin	3.7 mg/dL (0.3-1.1)
Direct Bilirubin	2.5 mg/dL (0-0.2)
AST	119 units/L (13-39)
ALT	234 units/L (7-52)
ALP	214 units/L (34-104)
Lipase	215 units/L (11-82)

CASE PRESENTATION

A 79 YO M PMH presented to the ED with generalized abdominal pain for 6 weeks prior to admission. Pain was associated with nausea, diarrhea, anorexia and unintentional weight loss of 18 lbs. A gallbladder was not identified on CT A/P and an US was performed, which showed shadowing and contracted gallbladder concerning for obstructive choledocholithiasis. Labs were remarkable for elevated LFT's in a mixed hepatocellular-cholestatic pattern with conjugated hyperbilirubinemia. MRCP showed loss of signal in the mid-CBD and small distal filling defects suggestive of small stones. ERCP identified 2 stones in the lower third of the main bile duct and a sphincterotomy with plastic biliary stenting was performed. A laparoscopic cholecystectomy was performed however aborted as no gallbladder was found. Specimens obtained intraoperatively from a perihilar hepatic mass and CBD resulted positive for cholangiocarcinoma. He underwent partial liver resection and gastrojejunostomy and was discharged under medical oncology for further management.

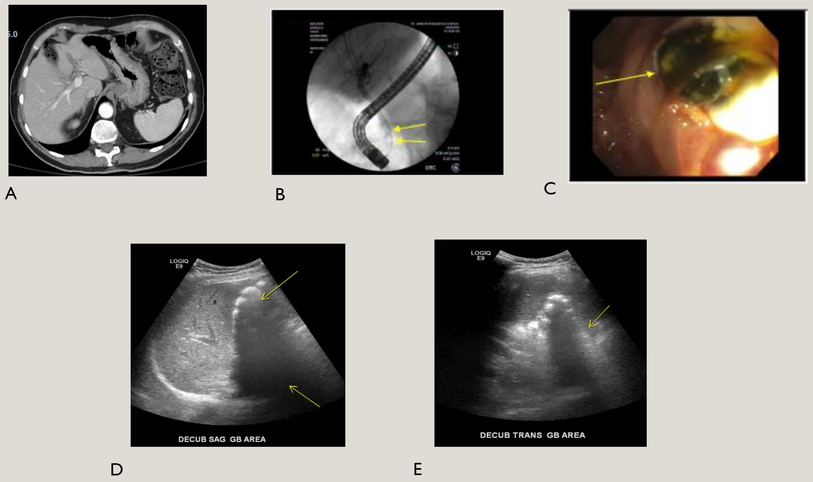

FIGURE LEGEND

A. CT A/P at the level of the absent gallbladder
B. ERCP demonstrating CBD stones (Arrows)
C. ERCP demonstrating CBD stones after balloon sweep
D. US sagittal decubitus of gallbladder area with shadowing (arrows)
E. US transabdominal decubitus of gallbladder area with shadowing (arrow)
Table 1. Admission Labs

DISCUSSION

Despite advances in modern technology the preoperative diagnosis of CGA is low. False positive sonographic results may show a shrunken, sclerotic gallbladder, likely artifact from periportal tissue interpreted as hyperechogenic shadows with cholelithiasis and possible CBD dilatation, as such patients are taken for surgical exploration. Surgeons should thoroughly investigate the intrahepatic, retro-hepatic, retro-duodenal, retro-pancreatic and retroperitoneal regions intraoperatively and send specimens for pathology. The relationship between CGA and CCA is unclear, but chronic inflammation of the hepatobiliary tract in the setting of chronic bile stasis due to biliary dyskinesia likely predisposed our patient to CCA.

CONCLUSIONS

A high index of suspicion is imperative to avoid unnecessary surgical exploration in patients with congenital gallbladder agenesis.

REFERENCES

1. Elzubeir N, Nguyen K, Nazim M. Acute Cholecystitis-like Presentation in an Adult Patient with Gallbladder Agenesis: Case Report and Literature Review. *Case Rep Surg.* 2020;2020:8883239.
2. Joseph JS, Ramesh V, Allaham KK, Ilangovan G, Khan MAB. Gallbladder Agenesis Mimicking Chronic Cholecystitis in a Young Woman. *Cureus.* 2021;13.
3. Agarwal PK. Agenesis of gall bladder: Diagnosed before it is an unpleasant laparoscopic surprise-clinical case report and review. *Int J Surg Case Rep.* 2020;76:144-147.
4. Yoldas O, Yazıcı P, Ozsan I, et al. Coexistence of gallbladder agenesis and cholangiocarcinoma: report of a case. *J Gastrointest Surg.* 2014;18(7):1373-1376.
5. Maemura K, Natsugoe S, Takao S. Molecular mechanism of cholangiocarcinoma carcinogenesis. *J Hepatobiliary Pancreat Sci.* 2014;21(10):754-760.
6. Joliat GR, Shubert CR, Farley DR. Isolated congenital agenesis of the gallbladder and cystic duct: report of a case. *J Surg Educ.* 2013;70(1):117-120.
7. Chowbey PK, Dey A, Khullar R, et al. Agenesis of gallbladder - our experience and a review of literature. *Indian J Surg.* 2009;71(4):188-192.
8. Al-Hakkak SMM. Agenesis of gall bladder in laparoscopic cholecystectomy-A case report. *Int J Surg Case Rep.* 2017;39:39-42.