Case report

Beware of the shrunken gallbladder – Case report of intraoperatively diagnosed gallbladder agenesis

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A R T I C L E   I N F O

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A B S T R A C T

Introduction and importance: Gallbladder agenesis, the congenital absence of the gallbladder, is a very rare biliary anatomical anomaly found in 13 to 65 people per 100,000. Symptomatic patients usually present with abdominal pain, nausea, and emesis, and are often misdiagnosed with gallbladder pathologies leading to unnecessary operations.

Case presentation: We report the case of a 63-year-old Caucasian female patient who presented with recurrent right upper quadrant pain (RUQ) and found to have a contracted gallbladder on ultrasonography (US). A hepatobiliary iminodiacetic acid (HIDA) scan was positive, and the patient was misdiagnosed with chronic cholecystitis. No gallbladder was found on laparoscopy and patient was diagnosed intraoperatively with gallbladder agenesis. Normal biliary anatomy was confirmed intraoperatively using the Firefly technique and there was no ectopic gallbladder on computed tomography (CT) scan obtained postoperatively. Patient currently doing well on conservative pain management.

Clinical discussion: Patients with gallbladder agenesis and normal bile ducts often have symptoms similar to that of usual gallbladder problems. Accurately diagnosing these patients preoperatively remains a challenge.

Conclusion: Symptomatic patients with agenesis of the gallbladder are often diagnosed intraoperatively despite major advances in diagnostic imaging techniques. In these cases, minimizing the risk of injury to the biliary tree is crucial. As our case demonstrates, it is critical for surgeons to become increasingly aware of this rare and important congenital anomaly. A very high index of suspicion is warranted in patients with a shrunken or contracted gallbladder. When in doubt, obtain cross-sectional imaging.

1. Introduction

First described by Lemery in 1701, gallbladder agenesis is a rare anatomical anomaly that results in complete absence of the gallbladder and cystic duct [1]. Its incidence has been reported as 0.007% to 0.0027% with a 3:1 female predominance in patients diagnosed intraoperatively [2]. Approximately half of patients with gallbladder agenesis present with symptoms consistent with gallbladder pathology, such as right upper quadrant abdominal pain, cholangitis, and jaundice [3]. The remaining cases include asymptomatic patients and patients who die from other congenital anomalies during fetal development [4].

Even with advancements in ultrasonography (US), computed tomography (CT), and magnetic resonance cholangiopancreatography (MRCP), the preoperative diagnosis of gallbladder agenesis remains daunting challenge. Unfortunately, for patients presenting with cholecystitis-like symptoms, diagnosis is often made intraoperatively during an attempted cholecystectomy [5]. For this reason, appropriate pre-operative work-up, a high level of suspicion, and avoidance of non-therapeutic surgical intervention is warranted.

We present the case of a 63-year-old female patient misdiagnosed with cholelithiasis and chronic cholecystitis. At laparoscopy, the diagnosis of gallbladder agenesis was made. This case has been reported in line with the SCARE 2020 criteria [6].

Abbreviations: CT, computed tomography; GA, gallbladder agenesis; HIDA, hepatobiliary iminodiacetic acid scan; MRI, magnetic resonance imaging; MRCP, magnetic resonance cholangiopancreatography; RUQ, right upper quadrant; US, ultrasonography.

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2. Presentation of case

A 63-year-old Caucasian female was referred to clinic after a recent visit to the small emergency department (ED) in a rural community with complaints of recurrent right upper quadrant abdominal pain that radiated to her back. Previous episodes of similar abdominal pain had been managed conservatively. The patient had a medical history that was significant for hypertension, gastroesophageal reflux disease, anxiety, and a parathyroid adenoma. Her surgical history included oral surgery and parathyroidectomy. She had no previous abdominal operations. Her medications included an antihypertensive, anti-anxiolytic, probiotic, and proton pump inhibitor.

The patient was hemodynamically stable. She was mildly tender to deep palpation in the right upper quadrant, but Murphy’s sign was not present. Her laboratory findings, including complete blood count, comprehensive metabolic panel and lipase were within normal limits.

An abdominal ultrasound (US) was noted for limited evaluation of the gallbladder due to wall shadow echo complex from probable gallstones filling (Fig. 1). Given the absence of nuclear medicine capability at this facility, the patient was scheduled for an outpatient hepatobiliary scan of the abdomen and pelvis, which confirmed an absent gallbladder (Fig. 5). There was no intrahepatic or extrahepatic ductal dilatation. Given challenges with health insurance coverage, an MRCP could not be obtained. She was prescribed intermittent pain medications and at 6-month follow up, patient was doing very well without complaints.

3. Discussion

Gallbladder agenesis (GA) is a very rare congenital anomaly that was first reported by Lemery et al. in 1701 [1]. Given the rarity of the condition, in symptomatic patients there is usually a low index of suspicion amongst surgeons leading to many patients undergoing unnecessary cholecystectomies. As our patient demonstrates, there is need for surgeons to be particularly suspicious in patients with an incompletely or poorly visualized gallbladder on ultrasonography or if it is found to be shrunken or very contracted.

The exact cause of gallbladder agenesis is not known. It is thought to result from failure of vacuolization of the pars cystica on the ventral mesentery or the failure of proliferation of the gallbladder bud during the 7th week of gestation [7]. Most cases, like ours, are unfortunately still diagnosed intraoperatively. Patients tend to present during the third and fourth decades of life with symptoms consistent with biliary disease [5]. Bennion et al. identified three categories of patients with gallbladder agenesis [8]. The first group (50 %) is of patients with symptoms characteristic of biliary pathologies. The second group (35 %) is asymptomatic and is usually diagnosed incidentally at autopsy. The last group (15 %) is patients with associated severe congenital anomalies, many of whom do not survive past the early years of life.

Our patient is part of the symptomatic category. The most common symptoms include right upper quadrant pain, nausea, emesis, intolerance of fatty foods and heart burn. Jaundice was noted in 27 % of patients in one review [9]. Our patient presented with recurrent right upper quadrant pain and was mistakenly diagnosed with chronic cholecystitis. In gallbladder agenesis, biochemical labs are usually unremarkable [2] [10] [11]. However, there have been cases with elevated liver enzymes and bilirubin, consistent with choledocholithiasis [12].

Ultrasongraphy remains the most common initial imaging modality in the work up of patients with RUQ pain. False-positive cases are often described as having a “shrunken,” “contracted,” or “constricted” gallbladder with hyperchoic “shadowing” suggestive of gallstones [5]. This is consistent with our patient who was thought to have a gallbladder that appeared “very small and contracted and contained some shadowing stone material.” Misdiagnoses with ultrasonography are thought to be due to structures mimicking the appearance of cholelithiasis such as an air-filled duodenum or an overlying transverse colon [13].

Our patient had a HIDA scan performed preoperatively but the diagnosis was assumed to be chronic cholecystitis given the non-visualization of the gallbladder. This is a common mistake that often leads to operative interventions as in our patient. The mistake highlights the need for preoperative cross-sectional imaging in making the correct diagnosis when the gallbladder is not fully visualized.

While advances in diagnostic techniques such as CT, MRI, MRCP, and ERCP have reduced the number of misdiagnosed patients with gallbladder agenesis, symptomatic patients like ours, remain prone to misdiagnoses. Sachin Malde in 2010 published a diagnostic algorithm to assist in the workup of patients with questionable imaging findings [14]. Additional imaging prior to operative intervention is recommended for patients without the WES triad on ultrasonography (visualization of gallbladder wall, echo from stone, an acoustic shadow) and with the presence of a constricted or sclerotic gallbladder.

When the diagnosis of gallbladder agenesis is made intraoperatively, it is critical to rule out an ectopic gallbladder. Areas to investigate

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Fig. 1. Abdominal ultrasound obtain preoperatively. Limited evaluation of the gallbladder due to wall shadow echo complex from gallstones filling gallbladder per radiology report.
Fig. 2. Hepatobiliary iminodiacetic acid (HIDA) scan obtained preoperatively. No visualization of the gallbladder.

Fig. 3. Intraoperative view of the porta hepatis showing the common bile duct (CBD) but no gallbladder visible.

Fig. 4. Near infrared fluorescent cholangiography with indocyanine green (ICG) – the Firefly technique showing good tracer uptake in liver and common bile duct (CBD). Cystic duct and gallbladder not present.
include retrohepatic position, in the retroperitoneum, retropancreatic, retroduodenal, in the falciform ligament or the lesser omentum. There is a high risk of injury to the biliary system especially if there is associated fibrosing of the porta hepatis. If this proves difficult, our advice is to abort the procedure and obtain cross-sectional imaging such as a CT scan or an MRCP to better assess the biliary anatomy.

With a rise in the number of robotic cholecystectomies performed, the use of near infrared fluorescent cholangiography with indocyanine green (ICG) – the Firefly technique – is increasingly common. Maker and Kunda showed that the Firefly technique correctly identified the extrahepatic biliary anatomy in all 35 patients undergoing elective cholecystectomy in their study [15]. This technique is now increasingly employed in lieu of the traditional intraoperative cholangiogram in defining biliary anatomy. In our patient there was no visualization of the cystic duct and gallbladder on Firefly, but a patent common bile duct was fully visualized.

Confirmation of gallbladder agenesis with MRCP is considered the gold standard. Due to insurance coverage challenges, an MRCP was not done in our patient. Despite this, her post-operative CT scan findings combined with the intraoperative Firefly confirm the diagnosis.

There is currently no consensus in the management of symptomatic patients with gallbladder agenesis. Our view is that a conservative approach should be attempted in all patients. This includes use of pain medications and antispasmodics. Though rare, consideration should be given for possible sphincter of Oddi dysfunction. In patients with severe symptoms an ERCP with sphincterotomy should be pursued. Our patient’s symptoms have been controlled conservatively for the past nine months with pain medications as needed.

It is not necessary to convert to an open procedure when the diagnosis of gallbladder agenesis is entertained intraoperatively. Abort the procedure and undertake further imaging studies to confirm agenesis or to plan management if an ectopic gallbladder is found.

4. Conclusion

Agenesis of the gallbladder is a very rare congenital defect with symptomatic patients presenting in a similar fashion to those with gallbladder disease. As in our patient it often leads to unnecessary operations despite advances in diagnostic imaging. It is very important for surgeons to have a high index of suspicion especially in patients found to have a shrunken, contracted, or constricted gallbladder on ultrasound. If diagnosed intraoperatively, it is best to abort the procedure and obtain cross-sectional imaging to minimize injury to the biliary system.

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Consent

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Author contribution

Kiyah Anderson: study design, manuscript writing – original draft
Avery Roland: data curation; writing - editing
Mark Miller: manuscript review and editing
Denis Foretia: study design, manuscript writing and review, overall supervision
All authors approved the final manuscript

Registration of research studies

Not applicable.

Guarantor

Denis A. Foretia.

Declaration of competing interest

None.

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