Gallbladder Hypoplasia, a Congenital Abnormality of the Gallbladder: A Case Report

Christophoros S. Kosmidis
Georgios D. Koimtzis
Maria S. Kosmidou
Fotini Ieridou
Triantafyllia Koletsa
Katerina T. Zarampouka
Eleni Georgakoudi
Isaac Kesisoglou

Corresponding Author: Georgios Koimtzis, e-mail: drgkkoimtzis@gmail.com

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Patient: Male, 62
Final Diagnosis: Gallbladder hypoplasia
Symptoms: Abdominal pain
Medication: —
Clinical Procedure: —
Specialty: Surgery

Objective: Congenital defects/diseases

Background: There are different variations in the anatomy of the gallbladder. Congenital abnormalities of the gallbladder such as agenesis and hypoplasia are rare conditions and difficult to diagnose with imaging studies. Patients are usually asymptomatic or have symptoms that mimic gallstone disease. The diagnosis is often made intraoperatively and is established by histopathological examination.

Case Report: We report a case of a 62-year-old male who had cholelithiasis symptoms and was falsely diagnosed with gallstone disease by abdominal ultrasound scan. The patient underwent an operation which revealed a rudimentary gallbladder. The histology result showed hypoplastic gallbladder tissue.

Conclusions: This case suggests that surgeons need to take into consideration congenital anomalies of the gallbladder intraoperatively in order to avoid any iatrogenic injury to biliary tract during a routine laparoscopic cholecystectomy. Intraoperative cholangiography can be a useful tool to avoid unnecessary surgical risky interventions.

MeSH Keywords: Cholecystectomy, Laparoscopic • Congenital Abnormalities • Gallbladder

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Background

There are several gallbladder congenital abnormalities (Table 1) [1]. One of them is gallbladder agenesis. This condition was first described by Lemery in 1701 [2]. It occurs in 0.01% to 0.065% of the population [3,4]. It develops from failure of normal development of the cystic bud in utero [5]. It is more common in women than in men with a ratio of 3:1 [6]. Gallbladder agenesis is often associated with other malformations such as gastrointestinal, cardiovascular, and musculoskeletal abnormalities [5]. Approximately 40% to 65% of those with gallbladder agenesis have the typical symptoms of choledolithiasis, i.e., right upper quadrant pain, nausea, vomiting, intolerance to fatty food, dyspepsia, and jaundice [7,8]. Symptomatic patients are approximately 25% of the population with gallbladder agenesis and will usually develop symptoms in the fourth or fifth decade of life [9]. Pre-operative diagnosis includes endoscopic retrograde cholangiopancreatography (ERCP) and ultrasound (US) that are usually misinterpreted as a shrunken, scarred, and atrophied gallbladder which is difficult to visualize and therefore patients are scheduled for surgery [10,11]. Evidence has shown that the main tool to establish gallbladder agenesis is magnetic resonance cholangiopancreatography (MRCP) because it allows a biliary cartography without injection of contrast material and should be used preoperatively to complement inconclusive ultrasounds [12]. MRCP can also demonstrate or exclude an ectopic gallbladder, as well. In most cases imaging studies show a shrunken and contracted gallbladder thus making the diagnosis of gallbladder agenesis impossible and therefore a laparoscopic and histopathology investigation is further required to confirm this [9]. Another congenital anomaly of gallbladder is hypoplasia. It has a similar etiology with gallbladder agenesis. Hypoplastic gallbladder can develop due to incomplete development of gallbladder bud or failure of re-canalization of solid primordium [13]. Hypoplastic gallbladder is associated with different conditions such as cystic fibrosis, cholangitis, neonatal hepatitis, and biliary atresia [14]. It is estimated that 33% of patients with a hypoplastic gallbladder are symptomatic in comparison to 54% that remain asymptomatic. Hypoplastic gallbladder is a rare condition that should be considered when the gallbladder cannot be identified intraoperatively [15]. Before proceeding to operate on a patient, the surgeon must evaluate the possibility of an ectopic gallbladder in areas such as the greater omentum or retroperitoneum [16]. In our case report, we present a patient with gallbladder hypoplasia who had a false positive ultrasound report for gallstone disease and was subsequently diagnosed based on histopathological examination.

Case Report

A 62-years-old male presented to our clinic with a two years history of intermittent right upper quadrant pain, nausea after ingestion of fatty food, intermittent vomiting of bilious content for two months, flatulence, and dyspepsia. There was no history of fever or jaundice. He had a past medical history of splenectomy twenty years before, after a traffic accident, and kidney stone disease that was diagnosed in November 2015. The patient was not taking any regular medications.

On clinical examination his abdomen was soft and non-tender and his bowel sounds were normal. Murphy’s sign was also negative. His vital signs were normal with an arterial blood pressure of 130/84 mm Hg, a heart rate of 75 bpm, a temperature of 36.6°C and a respiratory rate of 14 cycles per minute. His laboratory test values were all within normal range with a white blood cells count of 7500/μL, a hemoglobin value of 15.4 g/dL, a platelet count of 343, a urea value of 26 mg/dL, a creatinine value of 0.75 mg/dL, a Na+ value of 137 mmol/L, a K+ value of 4.6 mmol/L and INR value of 0.9. His ultrasound result showed a liver of normal size and structure with normal sized intra- and extra-hepatic bile ducts, a normal portal vein, a shrunken gallbladder filled with gallstones (Figure 1).

Table 1. Congenital abnormalities of the gallbladder [1].

| Anomaly                  | Prenatal age at onset | First appearance | Sex chiefly affected | Relative frequency | Remarks                          |
|--------------------------|-----------------------|------------------|---------------------|--------------------|----------------------------------|
| Absence of gallbladder   | 4th week              | Adulthood, if ever | Female              | Rare               |                                  |
| Duplication of gallbladder| 4th week              | None             | Equal               | Rare               |                                  |
| Deformation of gallbladder| 6th week              | None             | Equal               | Uncommon           |                                  |
| Left-sided gallbladder   | 4th week?             | None             | ?                   | Very rare          |                                  |
| Intrahepatic gallbladder | 2nd month             | None             | ?                   | Rare               |                                  |
| Mobile gallbladder        | 2nd month             | Late adulthood, if ever | Female | Rare | Symptoms result from torsion |
a normal pancreas, a normal prostate gland and urinary bladder, a normal right kidney and a 1.8 cm cyst in the left kidney.

The patient was listed for laparoscopic cholecystectomy but the operation was converted into an open operation due to adhesions that were found and the fact that the gallbladder could not be identified; however, the common bile duct was recognized. Intraoperatively, an unusual structure was noticed between the common bile duct and cystic duct, which was excised (Figure 2). A Redon drain was inserted in the sub-hepatic space. The patient was discharged five days postoperatively and had a good recovery.

The specimen was sent for pathology and was consisting of adipose tissue and measured 3.5×2×0.5 cm. (Figure 3). Cut sections revealed only a duct, 0.6 cm in length. A lymph node was found as well. Hematoxylin and eosin stained sections revealed the presence of a duct, one edge of which was dilated due to biliary sludge (Figure 4), while at the other edge multiple foci of mucous glands were found (Figure 5). These findings were compatible with the presence of the cystic duct and an effort to form the neck of gallbladder during embryogenesis. Hence the diagnosis of gallbladder hypoplasia was set. The lymph node presented reactive lymphadenitis.
Discussion

The gallbladder is formed at six weeks in the human embryo from the caudal part of foregut. Failure of development of the caudal division of primitive hepatic diverticulum leads to gallbladder agenesis [17,18]. Gallbladder agenesis is transmitted as a non-sex linked trait. Conditions that can be associated with an absent gallbladder include cardiovascular and gastrointestinal malformations as well as biliary atresia [19,20]. In addition, gallbladder agenesis has also been found in other syndromes such as Klippel-Feil syndrome, trisomy 18 and G syndrome [13,14]. Hypoplastic gallbladder embryologically develops due to incomplete recanalization of solid primordium [13,21,22].

The gallbladder is a thin walled hollow sac that has several functions: it concentrates and stores bile produced by the liver and empties mucoid bile in response to fatty food [22]. Gallbladder consists histologically of a folded mucosa of simple columnar epithelial cells and fibrovascular lamina propria. It also consists of a deeper muscularis and an external layer of supporting tissue with elastic fibers. The outer surface is covered by serosa. Gallbladder lacks a muscularis mucosae and submucosae [23]. Mucous glands and ducts occur only at the neck of gallbladder secreting mucous into lumen. In cholecystitis muscle layer is thickened by hypertrophy and a pouch of epithelium lined mucosa Rokitansky-Aschoff sinus is usually bulged through it [22,24].

There are very few reports of gallbladder hypoplasia [12,14] in comparison to agenesis that is more commonly reported [25–27]. Evidence has shown that a rudimentary gallbladder in children is characterized as hypoplasia, whereas in adults it can be due to post-inflammatory process. Surgeons may find a gallbladder stump directly attached to hepatic duct or a very short cystic duct. That was the case in our report, where a small, contracted gallbladder was found intraoperatively [28,29].

Gallbladder agenesis has three main categories: those which are asymptomatic (35%), those that are symptomatic (50%) and the last category which includes children with other congenital anomalies (15%) which are usually incompatible with life. In the symptomatic group, patients undergo surgery; the possible mechanisms for right hypochondrial symptoms are due to biliary duct stones, biliary dyskinesia, or non-biliary disorder [30]. Both gallbladder agenesis and hypoplasia can lead to intraoperative injury of the common bile duct or other parts of the biliary tree as a result of false identification of these structures [31]. Thus, a high degree of clinical suspicion is necessary when encountering anatomical variants and the usual structures cannot be properly recognized. The reason for converting a routine laparoscopic to an open cholecystectomy procedure is that there is a different anatomy in gallbladder agenesis resulting in an inability to pull on the gallbladder to dissect the triangle of Calot [32,33].

By reviewing the literature, we found that preoperative investigations failed to demonstrate the right diagnosis such as congenital anomalies of the gallbladder (agenesis); as a result, patients have unnecessarily operations. There are different radiological modalities when investigating gallbladder disease [32–34]. Usually ultrasonography is the gold standard investigation for the diagnosis of gallbladder stones. In gallbladder hypoplasia, ultrasound shows a contracted and shrunken gallbladder; however, this investigation is inconclusive preoperatively and often surgery is required in order to make the diagnosis [19]. In our case, the ultrasound misdiagnosed a contracted lithiasic gallbladder; as a result our patient underwent an operation to remove the gallbladder. Evidence has shown that a loop of gas-containing bowel located in the gallbladder fossa or in sub-hepatic folds can mimic a shrunken gallbladder containing gallstones that can result in a preoperative ultrasound inaccuracy [2]. Lack of awareness of this rare congenital condition amongst surgical and radiological staff was the main reason that we proceeded to an operative intervention and removal of the specimen (rudimentary gallbladder).

Some studies have shown that magnetic resonance cholangiopancreatography imaging (MRCP) and endoscopic retrograde cholangiopancreatography (ERCP) can be used preoperatively to diagnose biliary pathology [2]. MRCP can show if there is an ectopic gallbladder or other anatomical variants or malformations in the biliary system and is also more accurate in the diagnosis of gallbladder agenesis in comparison to ERCP which is more invasive and it can cause an injury to biliary tree [26,35]. ERCP has also been associated with high rates of morbidity and mortality and the unsuccessful rates of cannulation. MRCP is a non-invasive method to delineate biliary tract and should be considered preoperatively in cases where ultrasound is inconclusive [26,36].

A diagnosis of agenesis or hypoplasia of gallbladder can only be confirmed with histology, intraoperative cholangiography, or laparoscopy. If bile ducts are identified, then intraoperative cholangiography should be available prior to definite surgery to define biliary system anatomy. In particular cholangiography is an essential tool to identify stones in the common bile duct, an ectopic gall bladder, an intrahepatic gallbladder, or when gall bladder is buried by extensive adhesions or atrophic due to previous cholecystitis episodes [30,37,38]. In our case, a cholangiogram was not performed because the hepatic duct was thin. Another way of ruling out the presence of an ectopic gallbladder intraoperatively is by the use of a cholecystoscope, which can also ensure that there are no gallstone remnants inside the biliary tree after a common bile duct exploration [39].
There are no specific guidelines to manage patients with gallbladder hypoplasia. The cause of pain in patients with gallbladder hypoplasia is similar to post-cholecystectomy syndrome pain, which occurs due to dilatation of the common bile duct in an effort to store bile, which results in increased pressure in the sphincter of Oddi. It is speculated that the hepatic duct assumes the role of bile storage when an anatomical gall bladder is absent and this predisposes to cholestasis, infection, and cholelithiasis [40]. Conservative management includes smooth muscle relaxants with the options of sphincterotomy if the first option fails [5]. Several studies have shown that patients with symptomatic hypoplastic gallbladder disease were cured after laparoscopic cholecystectomy [29].

Conclusions

Gallbladder hypoplasia and agenesis are rare congenital conditions that require special consideration and remain to be a diagnostic dilemma for surgeons. Surgeons should be aware of these unusual abnormalities to avoid injury to bile duct and other unnecessary complications when operating. Preoperatively investigations such as MRCP and intraoperative cholangiography can be useful tools to avoid any unnecessary surgical risky interventions, especially when ultrasound is inconclusive.

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