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Incidental Intra Laparoscopic Cholecystectomy Diagnosis of a Gallbladder Agenesis, A Rare Diagnosis

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Case Report

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Abstract

Gallbladder agenesis is quite a rare anomaly; it's probably due to an embryologic mishap within the development of the hepatobiliary bud and is predominately associated with other congenital abnormalities. Most patients will stay asymptomatic; however, some will develop symptoms that mimic those of biliary, urinary, or gastrointestinal disorders. Symptoms typically appear within the fourth or fifth decade of patient life. For these reasons, gallbladder agenesis might be misdiagnosed. Leading to unnecessary surgery. This paper reports a case of gallbladder agenesis of a 41-year-old male. Presented with history of right upper quadrant abdominal pain associated with nausea and vomiting. Based on the clinical presentation and ultrasonography findings, symptomatic cholelithiasis was misdiagnosed. He was then seen by the surgical team as a case of acute cholecystitis. Imaging modalities commonly used in investigation of biliary disease symptoms where employed. Ultrasound and MRCP were done. However, findings have led to the diagnosis of acute cholecystitis and eventually to a laparoscopic cholecystectomy. This case has also shown that gallbladder agenesis can still be misdiagnosed, leading to the detection of gallbladder agenesis in the operating room.

Keywords: Gallbladder; Agenesis; Anomaly; Cholecystectomy; Cholecystitis

Abbreviations: AST: Aspartate Transaminase; ALT: Alanine Transaminase; ALP: Alkaline Phosphatase; GA: Gallbladder Agenesis; US: Ultrasonography; T-Bil: Total Bilirubin; CT: Computed Tomography; ERCP: Endoscopic Retrograde Cholangio-Pancreatography; HIDA: Hepatobiliary Imino Diacetic Acid.

Introduction

The absence of the gallbladder, commonly known as gallbladder agenesis (GA), is an extremely unusual anatomical defect. The stated frequency is fewer than one in every 6500 live births [1]. The majority of GA sufferers are asymptomatic their whole lives. As a result, the vast majority of cases are discovered at autopsy or inadvertently when having diagnostic imaging or surgery. However, around 23% [2] to 50% [3] of patients may present with symptoms of right upper quadrant discomfort that may be misdiagnosed as cholecystitis or symptomatic cholelithiasis. Routine preoperative ultrasonography (US) can be deceptive, subjecting patients to unneeded surgical operations.

Case Presentation

A 41-year-old male patient was presented to our office after several past admissions for biliary colic, the last admission was before a month of the current presentation with a diagnosis of acute cholysyctitis. However, in this elective admission for the multiple gallbladder stones laparoscopic cholecystectomy, the patient was doing well, healthy with a stable vital sign and no evidence of acute abdominal conditions. Firstly, the surgical operation was smooth until the surgeon reached the liver and did not find the gallbladder. After several attempts to do adhesolysis, the decision was to convert the operation to the classical open approach. Despite the field was clear and the abdominal cavity was fully visualized, the gallbladder cannot be identified. An intraoperative cholangiogram was ordered the result was absent gallbladder and dilated common bile duct. The stones were extracted, the CBD was repaired, and the wound was closed with an intraabdominal drain. The post operation course went smoothly on day three the drain was removed. A day after, the patient was discharged home.

Investigations

The patient had undergone laboratory work up that documented normal complete blood count, and liver function test for aspartate transaminase (AST), alanine transaminase (ALT), alkaline phosphatase (ALP), and total bilirubin (T-Bil) showed levels of 70.9 IU/L (reference range <=37 IU/L), 86.2 IU/L (reference range <= 42 IU/L), 240 IU/L (reference range 40-129 IU/L), 21.3 umol/L (reference range 5.1-17.1), respectively. Ultrasound of the abdomen showed normal echo texture of the liver, spleen and kidneys. However, the gall bladder was contracted and the common bile duct measures about 1.6 cm with a suspicion of stone impaction distally. There was no evidence of peritoneal free fluid in the abdomen or pelvis. MRCP was subsequently performed and revealed dilated CBD with multiple filling defects seen from the level to the ampullary part consistent with CBD stones and dilated common hepatic duct about 2cm.

Discussion

The gallbladder develops from the caudal part of the hepatic diverticulum in the fourth week of intrauterine life. Gallbladder agenesis or GA is thought to be caused by either a failure of the cystic bud to form or a lack of vacuolation [4]. GA is a very uncommon illness that presents without specific symptoms. It has an incidence of 0.007–0.0027% and a reported 3:1 female predominance of symptomatic cases. Gallbladder agenesis is also linked to other system abnormalities, and it appears that there is a “familiar tendency” [5]. GA can be seen in both children and adults [6]. Moreover, it is linked to a number of other abnormalities, including: Klippel Feil syndrome, malrotation of the gut and horseshoe kidney.

Clinically, the presentation of gallbladder agenesis is classified into three categories:

- Multiple fetal anomalies (15-16%): These patients invariably die during the perinatal period as a result of associated anomalies, and GA was only discovered at autopsy. Most encountered anomalies were cardiovascular, GI, genitourinary, anterior abdominal wall, and anomalies of the central nervous system.
- Asymptomatic group (35%): GA was identified either through autopsy, laparotomy for an unrelated disease, or through screening of patients relatives. These patients don’t have biliary tract symptoms, and no treatment is needed.
- Symptomatic group (50 %): Typically, it is an isolated aberration. This major group first appears in the fourth or fifth decades. In the symptomatic group, common signs are chronic right upper quadrant pain (90%) [6].

Diagnosing gallbladder stands as a challenge, and in many cases, diagnosing the condition without surgery is not possible [7]. With clinical symptoms similar to those of biliary colic including right upper quadrant pain, gallbladder agenesis is commonly misdiagnosed as cholecystitis or choledocholithiasis [8]. For patients with biliary colic, abdominal ultrasound is the diagnostic test of choice. It is also usually the initial investigation done to all patients presenting with symptoms of gallbladder disease. However, ultrasound can definitely be misleading in the diagnosis of patients with gallbladder agenesis. In fact, GA can be often misdiagnosed by ultrasound as having a contracted gallbladder [7]. Studies show that specific signs on the ultrasound should be enough to raise suspicion of gallbladder agenesis; these features have been described as the absence of features of the WES triad (visualization of the gallbladder wall, the echo of the stone, and the acoustic shadow) and the double-arc shadow on ultrasound.

In order to confirm the diagnosis and prevent unnecessary procedures, additional testing, such as a radioisotope hepatobiliary scan, MRCP, or CT is necessary [9]. According to the Malde algorithm, if ultrasound scan is not sufficient for identifying the gallbladder, MRCP, CT and ERCP are the investigations that come next in line, respectively and depending on availability [10]. MRCP is considered the preferred diagnostic method for investigating gallbladder agenesis before surgical interventions. It is also used after surgery and when ultrasonography is inconclusive [11]. The reason for this superiority of MRCP is that it does not rely on the passage of contrast for visualization of the biliary tree and is therefore not affected by biliary stasis [8]. Thus, MRCP allows for a comprehensive evaluation of the biliary system

and is considered the optimum imaging approach in case of gallbladder agenesis.

On the other hand, Hepatobiliary Imino Diacetic acid scan or HIDA scan and endoscopic retrograde cholangio-pancreatography (ERCP) can also be deceptive in cases of gallbladder agenesis. Non-visualization of the gallbladder through these modalities is likely to be interpreted as obstruction of the cystic duct, anatomic differences, or technical errors [12]. This often leads to a diagnosis that is consistent with cholecystitis rather than the correct diagnosis of absence of the gallbladder [12]. Computed tomography (CT) scanning is another technique that can be used to diagnose agenesis of the gallbladder. Nonetheless, the same limitation applies to non-visualization of the gallbladder in which it can be attributed to other explanations [9]. Despite the utilization of modern diagnostic modalities, diagnosis of gallbladder agenesis is still considered extremely difficult and is often made at laparotomy or during attempted laparoscopic cholecystectomy. Ideally, gallbladder agenesis could be diagnosed pre-operatively; nevertheless, it is more common for the diagnosis to be made through an operation [13]. If the diagnosis is established during surgery, the surgeon should first check for ectopic gallbladders. Excluding an ectopic gallbladder with its different variations that include intrahepatic, lesser omentum, retroperitoneum, retro hepatic retro duodenal or retro pancreatic is important. If unable to identify them; intraoperative cholangiography may be reasonable [10]. Additionally, gallbladder agenesis poses difficulties during laparoscopic surgery in which the biliary or portal structures can easily be wounded in the search for a gallbladder that does not exist [5].

Furthermore, due to the rarity of gallbladder agenesis, this condition remains relatively uncommon compared to other biliary diseases. There is a lack of awareness of this condition among medical and surgical professionals, and doctors that could encounter cases of GA are unfamiliar with it and often hesitate to document it [12]. Indeed, many cases end up having open surgery for a concrete diagnosis [11]. In many other cases, patients end up undergoing cholecystectomy despite suggestive findings on ultrasound, MRCP, CT, and ERCP (Table 1).

Diagnostic Modality	Advantages	Disadvantages
Ultrasound Scan	· Non invasive	· Nonspecific (can lead to misdiagnosis)
	· No radiation	· Not sufficient
	· Relatively Cheap	
MRCP	· Non invasive	· Relatively Expensive
	· Optimum diagnostic method before surgical intervention	
	· Does not require contrast and is not affected by biliary stasis	
CT Scan	· Non invasive	· Non specific
	· Useful postoperative modality	· Relatively Expensive
ERCP	□ Useful postoperative modality	□ Invasive
		□ Non specific
		□ Needs Admission to the hospital
		□ More Complications
		□ Relatively Expensive
Surgical Intervention	□ Specific	□ Invasive
	□ Accurate	□ More Complications
	□ Definitive diagnostic modality	□ Needs Admission
	□ Used to rule out ectopic gallbladder	□ Risk of Anesthesia
		□ More post-surgery recovery duration
		□ More Costly

Table 1: Findings on ultrasound, MRCP, CT, and ERCP.

This underlines the vital need for greater awareness and recognition of gallbladder agenesis as a cause of biliary symptoms. When initial radiologic tests and imaging modalities suggest an absent gallbladder, surgeons and

radiologists should attempt to identify gallbladder agenesis pre-operatively and prevent operative intervention [9]. In regards to treatment of gallbladder agenesis, specific guidelines are not identified. Many patients continue to experience biliary colic and abdominal pain. The pain is explained by the increased pressure in the sphincter of Oddi due to dilatation of the common bile duct that occurs in an attempt to store bile in the absence of the gallbladder [8]. In most cases, management is conservative. Smooth muscle relaxants and analgesics are used to relieve the pain [10]. In other instances, sphincterotomy has also been a successful procedure for relieving symptoms of gallbladder agenesis [11].

In our case, the patient was a 41 year-old male, and similar to the classical symptomatic presentation discussed earlier [7], the patient had symptoms of pain and vomiting. He was then seen by the surgical team as a case of acute cholecystitis. Imaging modalities commonly used in investigation of biliary disease symptoms where employed. As described in the case presentation, both ultrasound and MRCP were done; however, findings have led to the diagnosis of acute cholecystitis and eventually to a laparoscopic cholecystectomy. This case has also shown that only through surgery was the right diagnosis of gallbladder agenesis finally reached as was encountered in many cases in literature [11,13].

Conclusion

In conclusion, gallbladder agenesis is a rare condition that could present in both asymptomatic patients and patients with symptoms of biliary colic. GA should be given more attention when gallbladder is not visualized in routine imaging methods. In such cases, MRCP is a promising method of investigation for diagnosing GA pre-operatively. Yet, diagnosis during surgery remains a common and prevalent happening in cases of gallbladder agenesis.

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