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Case Report

Gallbladder and Cystic Duct Agenesis as an Extremely Rare Embryologic Abnormality and Misdiagnosis

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Abstract:

Background: Gallbladder agenesis (GA) is an uncommon congenital condition defined by the absence of the gallbladder and cystic duct resulting from an anomaly in embryonic development. It is often linked with other congenital anomalies, but an isolated variant is very infrequent.

Basic procedures/main findings: The clinical presentation is diverse. In fact, GA is often diagnosed incidentally. Magnetic resonance cholangiopancreatography (MRCP) is regarded as the preferred diagnostic approach, since it circumvents needless and risky surgical procedures in symptomatic patients. This report presents a case of gallbladder agenesis incidentally identified during surgery in a 29-year-old patient.

Conclusions: Although GA is uncommon, clinical awareness of the disorder facilitates accurate preoperative diagnosis and thereby reduces morbidities associated with unnecessary surgical exploration.

Key words: agenesis; gallbladder; cystic duct; embryologic abnormality

Introduction

Developmental abnormalities of the gallbladder are relatively rare and is often encountered during cholecystectomy [1]. Gallbladder agenesis (GA) is an uncommon congenital abnormality. The incidence rate in the general population is documented as 13 to 65 cases per 100,000 individuals. In clinical data, the incidence ranges from 0.007% to 0.0027%, but in autopsy series, it ranges from 0.04% to 0.13% [2]. Isolated GA is an uncommon congenital defect caused by the inability of the cystic bud to form in gestation. Since its first description by Lemery in 1701, various additional cases have been documented [3].

Three clinical categories of GA have been recognized: (1) asymptomatic cases, typically discovered incidentally during laparotomy for unrelated issues (35%), (2) symptomatic cases (50%), and (3) cases in children with multiple fetal anomalies, such as tetralogy of Fallot and pulmonary agenesis, resulting in perinatal mortality (15–16%) [2,3]. The majority of those diagnosed with some GA are asymptomatic. The primary clinical suspicion is often cholecystitis or biliary colic. Symptomatic individuals may exhibit signs of biliary colic, cholecystitis, or even jaundice. A hypothesized mechanism suggests that pain may arise from biliary dyskinesia, Sphincter of Oddi dysfunction, or choledocholithiasis [2,4]. GA is often linked with other congenital anomalies (12.8-30%). It has been documented that this is associated with gastrointestinal, cardiovascular, genitourinary, and skeletal malformations [3]. Even if biliary agenesis is identified during surgery, the surgeon may remain unaware of the associated pathological problems, potentially leading to a delay in appropriate patient management. Biliary agenesis may be erroneously diagnosed radiologically as cystic duct obstruction, acute cholecystitis, or chronic cholecystitis in patients where the gallbladder is not visible during ultrasonography, as atrophied and fibrotic gallbladder tissue is challenging for diagnosis in chronic inflammatory conditions[1].

Case report

In this report, we describe a case of a 29-year-old female patient sent to the radiology department due to suspected cholelithiasis. She had

persistent dyspeptic symptoms characterized by abdominal pain and bloating. She had three analogous episodes of discomfort in the upper-right abdominal quadrant and received treatment with smooth muscle relaxants and nonsteroidal anti-inflammatory drugs (NSAIDs). She had no prior surgical history. No relevant family history was noted. No congenital abnormalities or disorders were documented. She was not prescribed any routine drugs. The patient had a healthy weight, with a BMI of 19.5 kg/m². The physical examination revealed no signs of jaundice or pallor. Upon abdominal palpation in the right hypochondrium, mild tenderness was seen without guarding. The laboratory studies, including liver function tests, were normal.

The abdominal ultrasound revealed a typical echotexture of the solid organs. Nonetheless, the gallbladder was presumed to be sclerotic and atrophic. No indication of peritoneal free fluid was seen. Due to the maintenance of pain, laparoscopic cholecystectomy is indicated. Immediately after placement of the camera and ports, a gallbladder agenesis was noted. The hepatoduodenal ligament, the left half of the liver, as well as the place on the visceral side of the liver where the gallbladder is normally present *(*Fossa for gallbladder, *lat. fossa vesicae fellae)* were examined in detail **(Figure 1).** The operation was completed as an exploration.

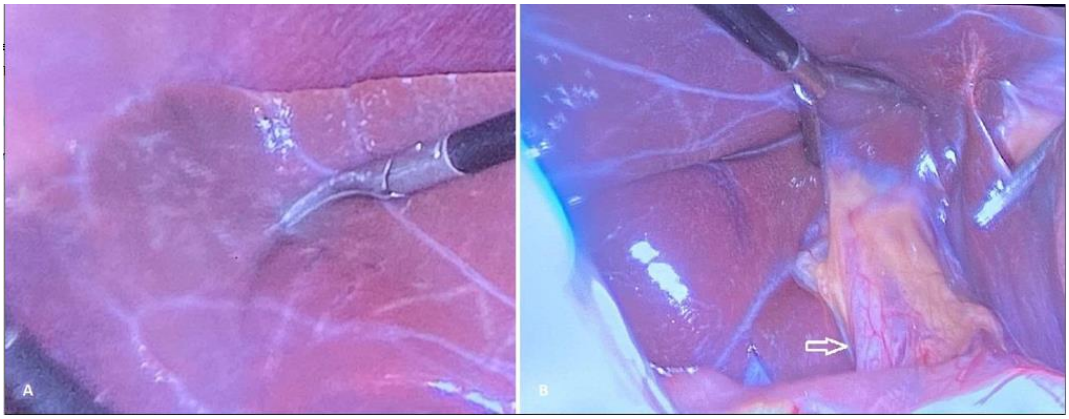

Figure 1: A) Intraoperative laparoscopic view of gallbladder bed; B) Intraoperative laparoscopic view of common bile duct (white arrow)

Magnetic resonance cholangiopancreatography (MRCP) was subsequently performed since the suspicion of gallbladder agenesis was put forth on operation. MRCP was done after overnight fasting. Routine MRCP sequences were performed with GE SIGNA 1.5 Tesla MRI. MRCP showed an empty gallbladder fossa and the gallbladder was not visualized in positions such as the posteroinferior part of the liver, retrohepatic, retropancreatic, retroduodenal, or left side of the liver. The common bile duct, common hepatic duct, and rest of the biliary structures appeared normal and were not dilated. No evidence of choledocholithiasis was observed. The pancreatic parenchyma showed normal signal intensities with a normal-appearing pancreatic duct. No evidence of inflammation or fluid was observed in the peritoneal cavity **(Figure 2).**

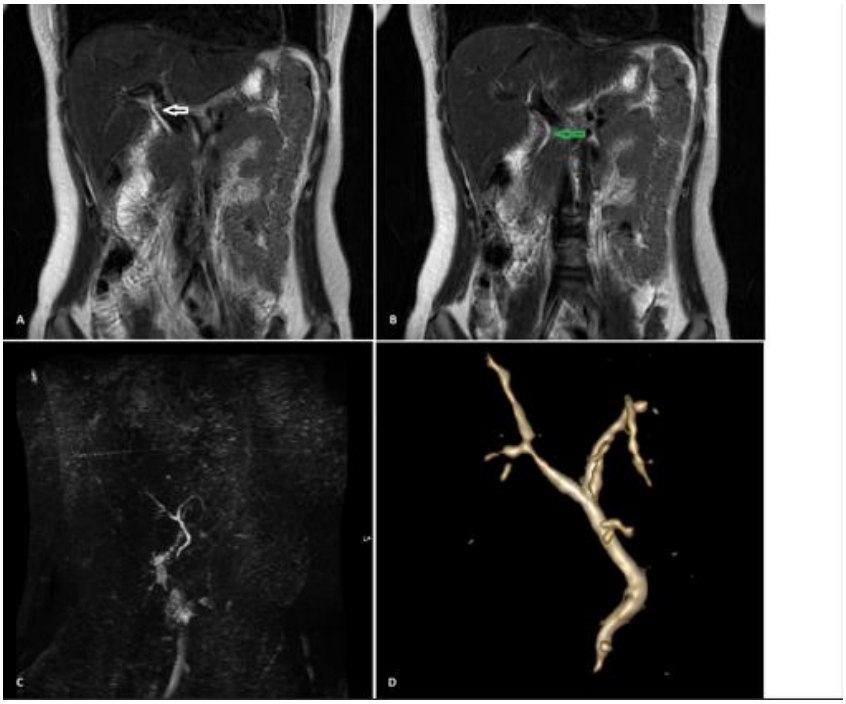

Figure 2: A, B) Postoperative magnetic resonance cholangiopancreatography (MRCP): The absence of the gallbladder and cystic duct; in addition the common bile duct (white arrow) and common hepatic duct (green arrow), respectively, are normal in caliber and course (coronal plane, T2 PROP sequence); C) 3D Oblique MRCP showing the common bile duct and common hepatic duct; D) 3D reconstruction of this anatomical abnormality

Discussion

In past years, all case were found after surgery, despite the high resolution of diagnostic imaging technology. Accurately diagnosing GA pre-operatively in symptomatic patients has proved to be very challenging, resulting in many cases still being identified intraoperatively [2]. In cases when a patient presents signs of biliary colic or upper abdominal discomfort, the preferred first investigation is an abdominal ultrasonography. The "WES" triad (wall-echo-shadow sign – representation of gallbladder Wall, Echo of stone, and acoustic Shadow) has been suggested to differentiate between a constricted gallbladder containing gallstones and bowel loops. Consequently, the results of abdominal ultrasonography may be misleading, requiring MRCP to verify a diagnosis of GA. MRCP, being a non-invasive technique, is the preferred diagnostic method for this type of condition. An ectopic gallbladder may also be found [4]. Since MRCP does not need contrast administration for the biliary and pancreatic ducts visualization, it is unaffected by biliary stasis. Alternative modalities, like endoscopic retrograde cholangiopancreatography (ERCP) and endoscopic ultrasonography, may be used for diagnosis; however, these are invasive procedures that often provide not much data about the existence of ectopic gallbladders [2].

Upon exclusion of choledocholithiasis and confirmation of gallbladder agenesis, conservative therapy may include the administration of smooth muscle relaxants or biliary sphincterotomy for refractory patients [4]. Our patient had a favorable response to conservative therapy with proton pump inhibitors and antispasmodics, which remains controlled in outpatient care. Possible future symptoms were discussed, and a yearly follow-up was advised.

In summary, this case report of a female patient with biliary colic symptoms mimicking chronic cholecystitis, finally diagnosed with gallbladder agenesis, demonstrates a rare although remarkable mimicry of cholecystitis, both clinically and radiographically. GA should be considered when the gallbladder is inadequately visible in standard imaging techniques for individuals with biliary-type pain. Although GA is uncommon, clinical awareness of the disorder, along with appropriate preoperative examinations, facilitates accurate preoperative diagnosis and thereby reduces morbidities associated with unnecessary surgical exploration. Consequently, the integration of clinical and radiological expertise enhances patient care.

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Contributions:

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Footnote

Conflicts of Interest: The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Informed Consent: The authors declare that they have received informed consent from patient to publish images and laboratory findings.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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