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Case Report

The pitfalls of gallbladder agenesis combined with small bowel malrotation

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Abstract

Gallbladder agenesia is a rare congenital anomaly that is frequently overlooked in clinical practice, often misdiagnosed as chronic sclero-fibrous cholecystitis or cholelithiasis. Failure to recognize it intraoperatively can prompt unnecessary and hazardous exploration of the biliary tree and liver, increasing the risk of iatrogenic injuries.

This report describes a rare case of gallbladder agenesia coincidentally discovered during surgery, alongside adult midgut malrotation—both diagnosed intraoperatively. The patient presented with symptoms mimicking biliary colic, which, in 23–55% of gallbladder agenesia cases, lead to symptomatic presentation.

Preoperative ultrasound commonly misinterprets the absent gallbladder as a shrunken, hypoplastic, or contracted organ due to artifacts from intestinal gas or adjacent structures. In the presented case, symptoms and ultrasound misinterpretation were likely attributable to the malrotated jejunum positioned beneath the liver. Magnetic resonance imaging (MRI), particularly MR cholangiography, is recommended when ultrasound findings are inconclusive or suggest a scleroatrophic gallbladder, though misinterpretation remains possible due to the condition's rarity and low clinical awareness.

Intraoperatively, overzealous dissection poses the greatest risk. Authors advocate aborting the procedure upon suspicion of agenesia, followed by postoperative confirmatory imaging (CT or MRI), rather than extensive exploration.

Adult midgut malrotation is also uncommon, with only 10% diagnosed in adulthood. Most cases are asymptomatic or cause vague abdominal symptoms (pain, nausea, bilious vomiting), often mistaken for biliary pathology. The patient exhibited duodenal nonrotation with right jejunal positioning and normal colon (DNJRCR type). In asymptomatic incidental findings, a conservative approach is recommended.

In summary, heightened suspicion for gallbladder agenesia is essential in ambiguous ultrasound reports. Preoperative MRI can prevent unnecessary surgery, while intraoperative recognition should prompt procedure termination and postoperative imaging to avoid iatrogenic complications.

Keywords

gallbladder agenesis, bowel malrotation, laparoscopy, pitfalls, misdiagnosis.

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PENSOFT.

Introduction

Gallbladder agenesis (GA) is a relatively rare anomaly characterized by a complete absence of the gallbladder. It has been reported in 0.007%–0.03% of the cases, or in nine out of 10,000 cases.[1] The rate is higher in autopsy series (0.04%–0.1%), or 1 in 6,334 live births.[2,3] Despite being well known, this anomaly is rarely considered in daily practice and is often misdiagnosed as chronic sclero-fibrous cholecystitis or cystic lithiasis. If unrecognized during the operation, it could lead to unnecessary and potentially dangerous explorations of the liver and biliary tree with a high risk for iatrogenic injuries.

Herein, we report a case of GA diagnosed intraoperatively and discuss the possible pitfalls and mistakes in diagnosis and management.

Case report

A 67-year-old female patient was admitted to the hospital with a four-year history of pain and discomfort in the right upper quadrant, without concurrent symptoms of nausea and vomiting. She had arterial hypertension, type II diabetes, and a thyroidectomy for multinodular goiter in the past. The laboratory values were normal. The US was performed by a gastroenterologist and demonstrated a contracted gallbladder with stones. The physical examination was unremarkable. Laparoscopy revealed a complete absence of gallbladder with dilated common bile duct (CBD) and small bowel malrotation with a normal position of the large bowel ***(Figs 1, 2)****. The operation was terminated without additional exploration. The postoperative contrast CT scan demonstrated GA associated with non-rotation of the duodenum and malrotation of the small bowel* ***(Fig. 3)****.*

Discussion

The first description of GA is attributed to Lemery in 1701.[4] According to other sources, the first report came from Bergman in the same year.[3] Up to 1947, Latimer et al. reported a total of 73 cases. As of today, roughly 500 cases have been reported in the literature, including newborn and autopsy series[5], but generally, the condition is increasingly reported worldwide after the implementation of laparoscopic surgery.

In 2005, based on MEDLINE and PubMed, Peloponissios et al. published a narrative review of the literature from 1960 to 2003 and reported two cases from their institution.[1] They found that 23% of the cases with GA are symptomatic, in contrast to a review from 1945, wherein Dixon et al. reported 58% (60 cases, 24 of them diagnosed intraoperatively).[6] According to the review by Bennion et al. from 1988, 55% of the cases were symptomatic.[2] The most frequent manifestations were right upper abdominal pain (90%), nausea or vomiting (66%), fatty food intolerance (38%), and jaundice (36%), which was also confirmed by more recent reports.[4,7,8] Other authors also report jaundice in one-third of the patients.[7,9,10] One-third of the patients had dilated CBDs with stones in 85% of them.[2]

The clinical manifestation resembles the post-cholecystectomy syndrome and can be attributed to biliary dyskinesia or CBD stones (seen in 1/3 of the cases) or due to intestinal malrotation, as in the presented case.[2] The dilated CBD without stones can be attributed to the higher

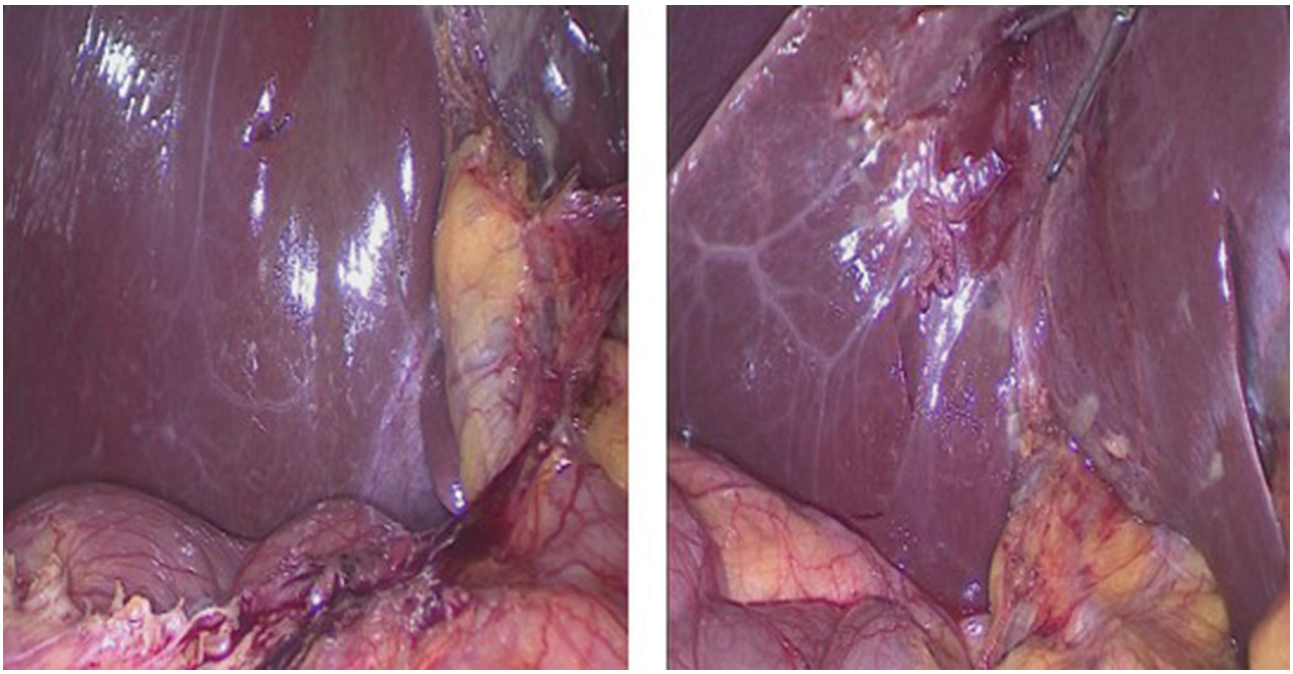

Figure 1. Absence of gallbladder with dilated common bile duct and subhepatic jejunal loops.

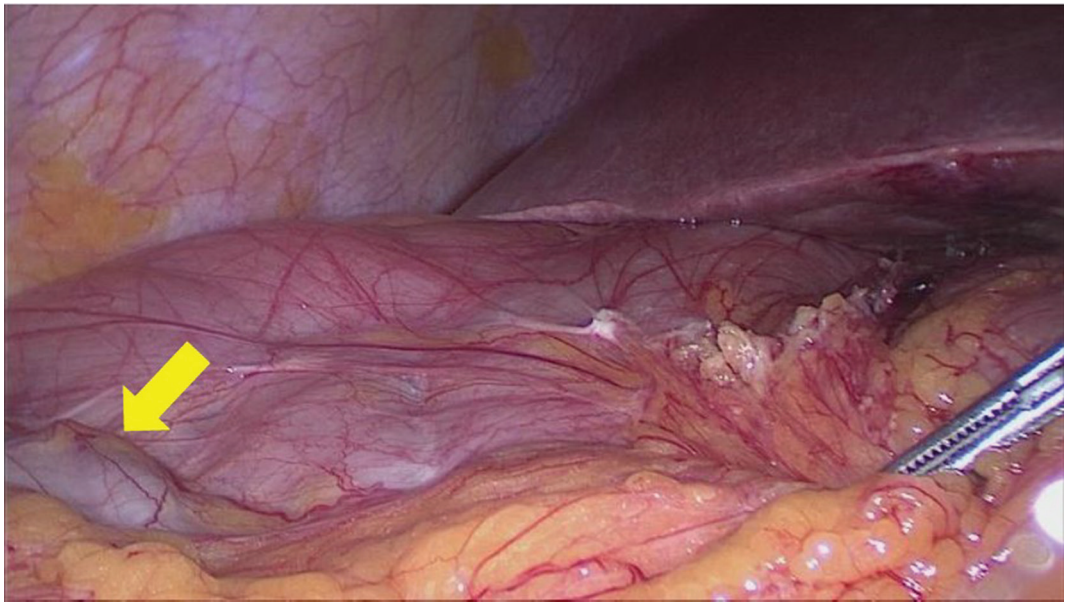

Figure 2. The right location of the jejunum and hepatic flexure (arrow).

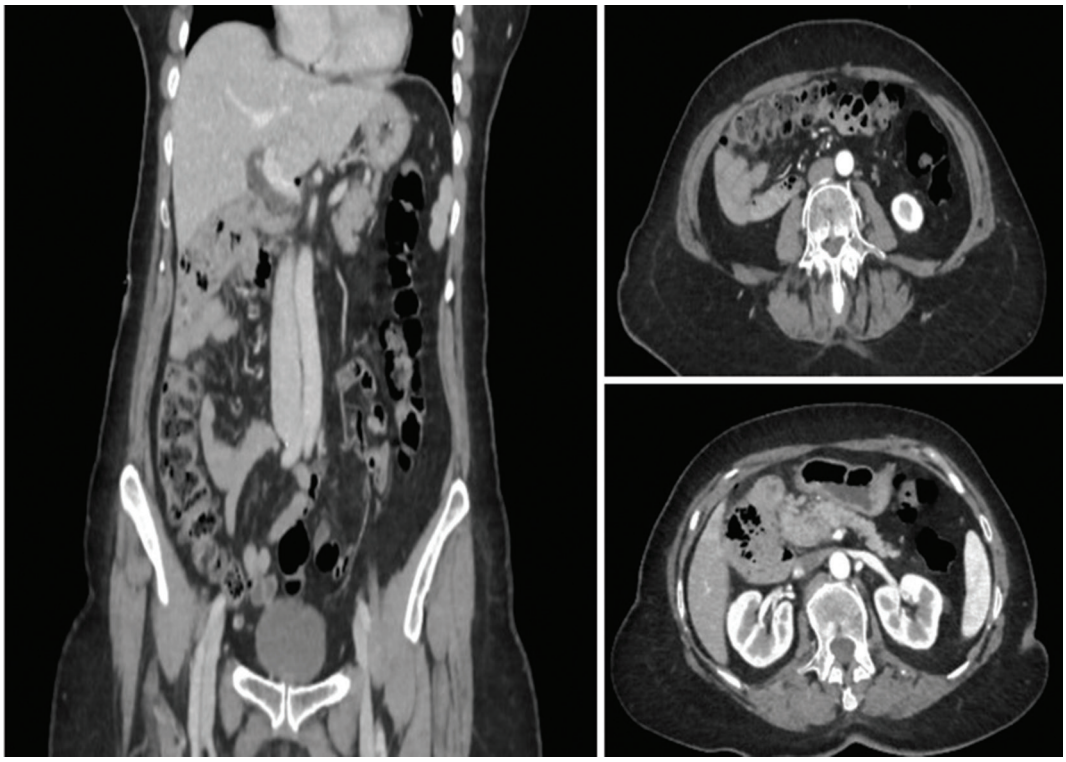

Figure 3. A contrast CT demonstrated GA, dilated CBD, non-rotation of the duodenum, and small bowel malrotation.

pressure in the duct and is similar to the moderate dilatation seen after cholecystectomy.

Although the US has high specificity and sensitivity for calculous cholecystitis (up to 98%), diagnosing the cases with fibro-sclerotic chronic cholecystitis with shrunken and contracted gallbladder remains difficult. Peloponissios et al. demonstrated well the difficulties of US diagnosis in such cases—despite the usefulness of McDonald's WES triad (wall, echo, and shadow) and "double arc shadow," in some cases, including GA, the diagnosis is hampered by the examination conditions and operator's experience.[1,11] In most reports of GA, the preoperative US showed "*shrunken gallbladder with hyperechoic shadows*," "*hypoplastic, contracted gallbladder,*" "*small gallbladder with small stones and diffused thickening of the wall,*" "*scleroatrophic gallbladder,*" or "*wall echo shadows in the gallbladder fossa*."[1,4,7,9,12,13] Gotohda reported a "*high echoic area without acoustic shadow*."[14] Nevertheless, the authors operated on

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the patient despite the absence of a gallbladder on CT and ERCP, presuming the diagnosis of chronic cholecystitis with an incarcerated cystic stone. On the other hand, in the literature, there are increasing reports of correct preoperative diagnoses.[7,8,15] Recently, Pinto et al. demonstrated an accurate diagnosis in a woman in a 17-week pregnancy.[16]

We agree with Peloponissios et al. that the main obstacle to the correct diagnosis is the misperception of shadowy opacities as stones due to intestinal gas artifacts or other adjacent structures.[1,17] According to Hammond, GA "is not a diagnosis that one should make preoperatively without support from corroborative imaging tests."[17] On the other hand, the non-visualization of the gallbladder is more frequent in the presence of a contracted or porcelain gallbladder, emphysematous cholecystitis, or air due to a fistula between the gallbladder and gut, or microgallbladder associated with cystic fibrosis.[17] Therefore, in such cases, it is advisable to perform additional imaging, especially MRI. However, misinterpretation is possible, most likely due to the GA's rarity and a lack of awareness of the condition in routine daily practice. Calder et al. reported a 28-year-old woman with GA who had seven US scans, three CT scans, and three MRIs over 4 years and who was, after all, operated on with a preoperative diagnosis of "cholelithiasis."[18] We speculate that the complaints and misinterpretation of the US scan are probably due to the subhepatic location of the jejunum.

The main intraoperative pitfall is overzealous exploration because it can lead to severe iatrogenic injuries.[1,9] Some authors recommend conversion to open surgery with an exploration of the liver, the falciform ligament, the lesser omentum, and the Kocher maneuver to search for gallbladder ectopia.[1,17] We share the opinion of other authors that this is unnecessary and the operation should be terminated with subsequent postoperative CT or MRI.[8,9] Intraoperative cholangiography is an invasive procedure that, if available, could be replaced with infrared fluorescent cholangiography with indocyanine green (ICG).[13] ERCP can easily manage one-third of patients with common bile duct stones.

AG is associated with other malformations in 13% of the children (two-thirds with cardiovascular malformations).[2] Midgut malrotation is observed in 1 of 6,000 births.[18] Although 85% of the cases are diagnosed during the first year, the malrotation "is not just a pediatric surgeon's problem."[19] About one-half of the adults with malrotation had biliary tree abnormalities.[20] Approximately 57% of the adults are asymptomatic.[21] The rest usually manifests with vague abdominal symptoms (mild pain, nausea, bilious vomiting), frequently attributed to biliary tract disease, as in our case and those reported by Calder et al., or with bowel obstruction.[18,21,22] CT allows a correct diagnosis of intestinal malrotation in adults. Xiong et al. validated a CT classification by assessing five points.[21] Only 25% of the patients had duodenal non-rotation as in the presented case, which was classified as DNJRCR (nonrotation of the duodenum, right location of the jejunum and cecum). In this variant, the cecum can be high or normal, as in our case. In asymptomatic patients, when encountered incidentally during the operation, we recommend a conservative approach advocated by others.[19]

Conclusion

GA is a rare and often misdiagnosed condition. If not recognized during the operation, extensive exploration of the liver and biliary tree increases the risk of iatrogenic injuries. The operation should be terminated with subsequent postoperative CT or MRI when GA is suspected.

A high index of suspicion for GA is needed in cases with inconclusive US findings or reports of shrunken or contracted gallbladder. The MRI cholangiogram is highly recommended in this scenario to avoid unnecessary operative interventions.

Ethical approval

Not applicable

Ethical statements

The authors declared that no clinical trials were used in the present study.

The authors declared that no experiments on humans or human tissues were performed for the present study.

The authors declared that no informed consent was obtained from the humans, donors or donors' representatives participating in the study.

The authors declared that no experiments on animals were performed for the present study.

The authors declared that no commercially available immortalized human and animal cell lines were used in the present study.

Conflict of interest

The authors have declared that no competing interests exist.

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Use of AI

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Data availability

All data used are referenced or included in the article.

Author contributions

All authors made significant contributions as follows: conceptualization: GP, VM, and KK; data curation: DS, MT, and MK; formal analysis, funding acquisition, and investigation: GP, DS, NI, KK, and VM; methodology: MT, VM, MK, and NI; project administration: GP, VM, and RC; resources, software, and supervision: GP, VM, and KK; validation, visualization, writing–original draft: GP, VM, MK, NI, KK, DS, and MT; writing–review and editing: GP, VM, MT, and KK.

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