Asian Journal of Case Reports in Surgery

Volume 7, Issue 1, Page 235-239, 2024; Article no.AJCRS.115622

Agenesis of Gall Bladder: A Case Report

Rohit Saha [a*], Uttam Konwar [b++], Diganta Borgohain [b++], Razvi Das [b#], Deeparani Pegu [b†] and Surya Kumar Saikia [b†]

[a] PGT General Surgery, Assam Medical College and Hospital, Dibrugarh, Assam, India.
[b] Department of General Surgery, Assam Medical College and Hospital, Dibrugarh, Assam, India.

Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

Article Information

Open Peer Review History:
This journal follows the Advanced Open Peer Review policy. Identity of the Reviewers, Editor(s) and additional Reviewers, peer review comments, different versions of the manuscript, comments of the editors, etc are available here: https://www.sdiarticle5.com/review-history/115622

Case Report

Received: 21/02/2024
Accepted: 24/04/2024
Published: 29/04/2024

ABSTRACT

Background: Agenesis of the gall bladder is a rare condition, majority of which are usually diagnosed during intraoperative procedure because of low incidence of suspicion and high rate of false positive results for cholecystitis on ultrasound imaging.
Methodology: A 25 years old female patient presented with right upper quadrant pain, whose clinical features and abdominal ultrasound suggested chronic calculous cholecystitis. After pre-operative evaluation and preparation, she was planned for laparoscopic cholecystectomy.
Results: The patient underwent elective operative intervention on 17th June 2023. Intra-operatively, the liver and the extra-hepatic biliary tree were thoroughly inspected, but no gall bladder was detected anywhere in the undersurface of the liver. Decision was made to stop the procedure avoiding any further exploration thereof. Post-operatively the patient was subjected to magnetic resonance cholangiopancreatography (MRCP), which confirmed agenesis of gall bladder.

[++] Associate Professor;
[#] Assistant Professor;
[†] Registrar;
**Corresponding author: E-mail: rohitsaha2008@gmail.com;*

Conclusion: The accuracy of abdominal ultra-sonography (USG) in the diagnosis of gall bladder disease is only 97% and a second USG screening or MRCP has got a definite role to play in doubtful cases.

Keywords: Agenesis; gall bladder; MRCP.

1. INTRODUCTION

"Developmental abnormalities of the gall bladder are relatively rare. Agenesis of gall bladder is three times more common in females than males" [1,2]. "Patients typically present with symptoms akin to biliary colic or cholecystitis with symptoms such as dyspepsia, abdominal pain, nausea, vomiting, intolerance to fatty food and so on" [1,3]. "Most cases are sporadic (around 70%) and there is very little literature on familial links" [4]. "Since its first description by Lemery and Bergman in 1701 and 1702 respectively, a number of cases have been published, with a reported incidence of 0.01 to 0.06%" [5,6]. "Combined with the rarity of the condition, the diagnosis of cholecystitis is infrequently made. Upper abdominal ultrasonography is a first line imaging for the diagnosis of cholelithiasis with a sensitivity of 97%, but in gall bladder agenesis, the sensitivity decreases to 61%, and erroneous report of cholelithiasis or shrunken gall bladder suggestive of chronic cholecystitis is often made" [2,7]. "Due to these misinterpreted reports patients undergo unnecessary surgery and may encounter iatrogenic biliary tract injuries and portal injuries, due to excessive dissection to find out the absent gall bladder" [8]. "Sometimes conversion to open exploration is made. Preoperative magnetic resonance cholangiopancreatography (MRCP) and endoscopic ultrasound (EUS) should be considered. And when such condition is encountered intraoperatively, intraoperative cholangiography and intraoperative ultrasound can be done to rule out agenesis and ectopic gall bladder" [9].

2. CASE REPORT

A 25-year-old female presented to the hospital with history of right upper abdominal pain and vomiting which was on and off since 2 years. She was otherwise fit and healthy. Her blood tests showed normal liver function tests (LFTs). She had undergone an ultrasonography (USG) of the whole abdomen which gave an impression of a contracted gallbladder. There was no definite wall hyperaemia and probe tenderness was present. She was diagnosed with acute on chronic cholecystitis and antibiotics were commenced. Symptoms improved so she was subsequently discharged with plan for an elective laparoscopic cholecystectomy after 4 weeks of discharge.

After 4 weeks a repeat USG was done which confirmed chronic calculous cholecystitis with a 1 cm stone. An elective laparoscopic cholecystectomy was planned. Intraoperatively, the gallbladder was not seen. The examination of right and left subhepatic space, and portal region was done. The anatomy looked unremarkable other than the absence of a gallbladder. A second opinion was taken from another senior surgeon who agreed with the findings and the procedure was converted to open cholecystectomy to exclude any ectopic position of the gall bladder. Even after opening the abdomen, the gall bladder was neither seen nor any stone was felt and hence the intended operation was abandoned.

The patient was discharged home on recovery and was planned for an outpatient MRCP and repeat LFT. On MRCP no gallbladder or cystic duct was identified and was consistent with a diagnosis of a congenital agenesis. Common bile duct (CBD) was normal with no evidence of biliary stricture or choledocholithiasis. The repeat LFT was normal. She remained well postoperatively with no further symptoms. She was discharged with surgical follow-up. She was reviewed on outpatient basis following these investigations.

3. DISCUSSION

"The liver and gall bladder develop in the fourth week of intrauterine life" [10]. "The gall bladder originates from the cystic bud as a ventral outgrowth from the caudal region of the foregut. The vacuolation of hyperplastic epithelium starts at about the seventh week during which the gall bladder and cystic duct develop a lumen" [11]. "Though the exact pathogenesis is unknown, gall bladder agenesis is thought to be due to the failure of the cystic bud to develop further or to the failure of vacuolation" [10].

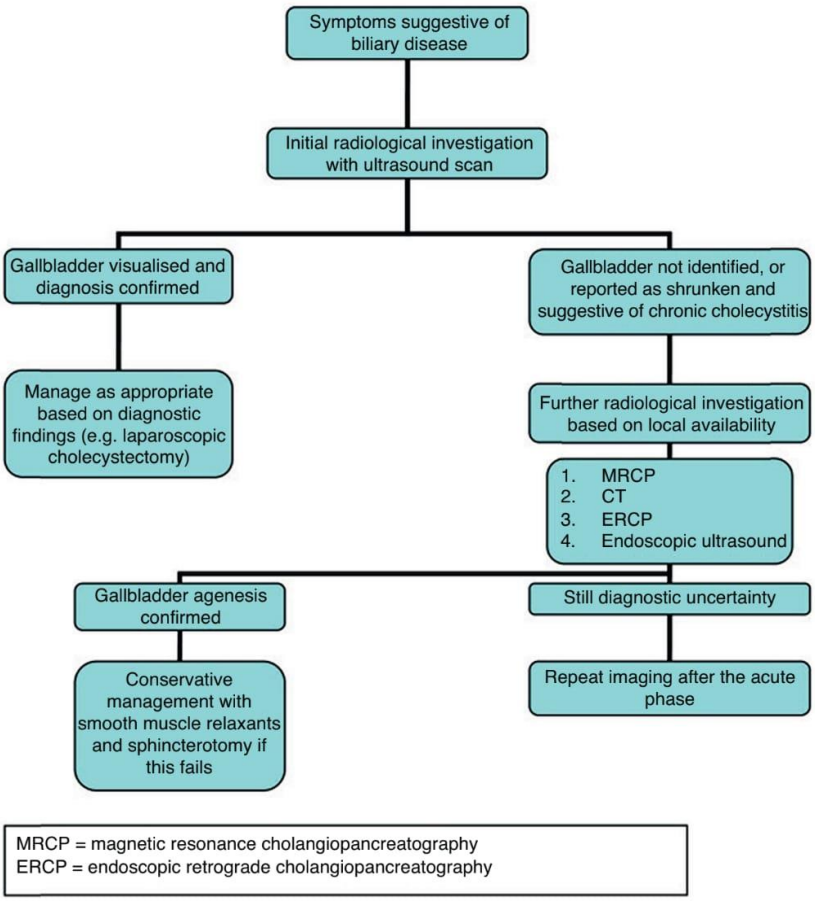

Fig. 1. Symptoms suggestive of billary disease

Kabiri H et al in their study suggested that “gall bladder agenesis is a rare congenital anomaly characterized by the absence of the gall bladder in conjunction with a normal bile duct system”. “The prevalence range is 0.007 to 0.13 % with an incidence of 0.007 to 0.027% in surgical cases and 0.04 to 0.13% in autopsy reports” [12].

Bennion RS et al and Gilbert Scott F explained the agenesis of gall bladder by two developmental theories [11,13]:

1. Failure of hepatic diverticula to develop into gall bladder
2. Failure of recanalization of cystic duct and gall bladder.

According to Winter RB et al agenesis of gall bladder may present as [14]:

1. Neonates with multiple fetal anomalies (15 – 16%): in these patients, agenesis usually diagnosed on autopsy because of death in perinatal period due to associated GIT, GUT, CVS anomalies.
2. Asymptomatic (35%): in these patients, agenesis recognized at autopsy and during laparotomy for other cause.
3. Symptomatic (40 – 60%): In these patients agenesis is discovered on USG abdomen, MRCP, EUS, and during laparoscopy for evaluation of (colicky) pain in right hypochondrium (90%), dyspepsia, vomiting.

“Agenesis of gall bladder is associated with congenital syndromes such as cerebrotendinous xanthomatosis, G-syndrome, Klippel-Feil syndrome, trisomy 18 and some cases reported after thalidomide therapy” [5,15,16,17].

“When a patient presents with symptoms of biliary colic or upper abdominal pain, the first investigation of choice would be an abdominal ultrasonography. In order to allow the gall bladder to distend and to reduce the amount of gas present in the bowel and assist in

distinguishing bowel loops from an absent gallbladder, the ultrasonography should be performed after fasting. The “WES” triad (wall-echo-shadow sign - demonstration of gallbladder Wall, Echo of stone, and acoustic Shadow) has been proposed to differentiate between a contracted gallbladder with gallstones and bowel loops” [11,18].

Latimer EO et al said that “MRCP and Endoscopic Retrograde Cholangiopancreatography (ERCP) provide excellent alternatives to open exploration and intra-operative cholangiography” [19]. “Though ERCP contributes little in diagnosis of agenesis because non-visualization of gall bladder is interpreted as cystic duct obstruction” [20]. “If the diagnosis is made pre-operatively or the symptoms continue postoperatively, a possible conservative treatment for this symptomatic group includes smooth muscle relaxants, and if this fails, sphincterotomy” [19].

“Preoperative MRCP should be considered in cases of USG diagnosis of non-visualization of gall bladder” [7]. “Other diagnostic modality includes EUS, intra-op ultrasound and selective arteriography can be used for agenesis. But their availability is less” [20]. “There were some cases reported in which the agenesis of gall bladder was diagnosed preoperatively and the operation was avoided” [21]. Malde S et al suggested “a decisional tree for the investigation of suspected gallbladder agenesis in an attempt to identify this rare condition pre-operatively, thereby preventing the unnecessary operative intervention as was seen in this case” as shown in the Fig. 1 [20].

4. CONCLUSION

When non-visualization of gallbladder is present during laparoscopy or open exploration intra-operative cholangiogram, intraoperative ultrasound and postoperative MRCP or endoscopic ultrasonography (EUS) can help in the confirmation of agenesis or ectopic gallbladder.

CONSENT

As per university standard patient written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

As per international standards or university standards, written ethical approval has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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Peer-review history:
The peer review history for this paper can be accessed here:
https://www.sdiarticle5.com/review-history/115622