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Agenesis of the gallbladder, a rare congenital anomaly that can mimic chronic cholecystitis: case report

Abstract

Agenesis of the gallbladder is a rare congenital malformation caused by the failure of the cystic bud to develop in the gallbladder, which may or may not be associated with other biliary malformations. It is an anomaly of variable symptomatology, with 50% of patients presenting symptoms mimicking biliary colic; it may be discovered by chance during surgery, while 15% may present fatal fetal malformations. When symptomatic, it is interpreted as chronic cholecystitis, with a narrowed appearance of the gallbladder on abdominal ultrasound, as the biliary structures may resemble those of a gallbladder, demonstrating the need for radiologists and surgeons to be aware of this diagnosis and its management, in order to avoid unnecessary surgery. In this article, we present a case of gallbladder agenesis in a young man who presented with hepatic colic-like symptoms suggestive of cholecystitis. The gallbladder was not well visualized on ultrasound, simulating chronic cholecystitis, and a Bili MRI concluded that the gallbladder was absent, thus avoiding surgery.

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Sawssen Ben Marzouk, Wael Ferjaoui, Adel Jelassi, Mohamed Hedi Mannai, Mohamed Bechir Khalifa

Department of General surgery, Military University Hospital, Faculty of medicine of Tunis, University of Tunis El Manar, Tunisia

Correspondence: Wael Ferjaoui, Department of General surgery, Military University Hospital, Faculty of medicine of Tunis, University of Tunis El Manar, Tunisia, Tel +216 52430099; Email farjaouiwae4@gmail.com

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Introduction

Agenesis of the gallbladder was first reported in 1701 by Lemery et al.[1] It is a malformation of the biliary tree that may be associated with several genitourinary, gastrointestinal or cardiovascular malformations.[2] Most patients with gallbladder agenesis are asymptomatic, although 50% may present with symptoms such as abdominal pain in the right hypochondrium, dyspepsia, nausea and vomiting.[3,4] Abdominal ultrasonography, with a sensitivity of 60% in the detection of congenital malformations,[5] is the first examination requested for this symptomatology, and may show findings compatible with chronic cholecystitis, a narrowed or sclero-atrophic gallbladder. If this is the case, Bili MRI should be performed.[6] If agenesis is discovered intraoperatively, further dissection is unnecessary to avoid complications such as biliary wounds.[7]

Clinical case

Our 30-year-old patient presented with nausea and abdominal pain in the right hypochondrium. Physical examination showed no mucocutaneous jaundice or pallor with slight tenderness of the right hypochondrium. Laboratory tests, including liver function tests, showed a slight elevation of YGT and PAL. Abdominal ultrasonography showed a sclero-atrophic gallbladder, with fine intra- and extra-hepatic bile ducts and a thin duct of Wirsung. The ultrasound appearance and cholestasis justified a Bili MRI (Figure 1) concluding to a homogeneous liver with regular contours and no detectable focal lesions ,the vesicular loge was free in relation to agenesis, the bile ducts were not dilated, the pancreas was normal in appearance with a fine Wirsung.

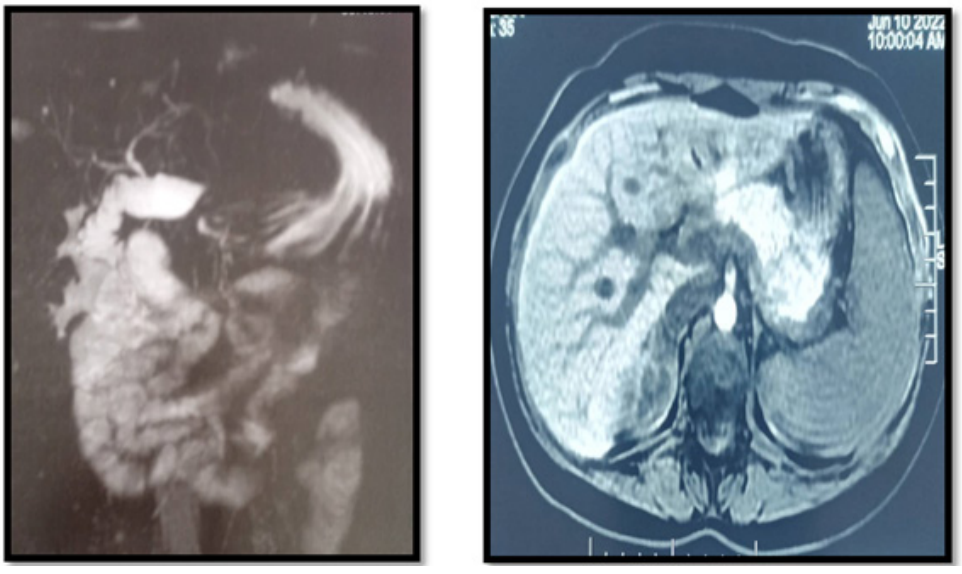

Figure 1 MRI photo showing missing gallbladder.

An abdominal CT scan revealed no ectopic location of the gallbladder; the diagnosis was agenesis of the gallbladder; the pain improved with antispasmodic medication.

Discussion

The exact etiology of gallbladder agenesis is unknown. The gallbladder is formed from a hepatic bud, whose failure to develop leads to gallbladder agenesis. It results either from a defect in cystic bud formation, or from a lack of vacuolization.[8,9] 500 cases have been reported in the literature, with a predominance of females.[10] The incidence of vesicular agenesis ranges from 0.0007 to 0.00027.[6] Most cases have been diagnosed intraoperatively.[11] For some authors, there is a hereditary component, as reported by Wilson et al.[12] This congenital anomaly may be isolated or associated with other congenital anomalies such as biliary, gastrointestinal and genitourinary malformations. According to Stevenson et al, 70% of cases are isolated.[13] Diagnosis of vesicular agenesis in adults remains difficult due to the variety of symptomatology. In 2015, a classification according to symptomatology was proposed: type I for symptomatic agenesis and type II for asymptomatic forms.[14] The symptomatology is explained by dysfunction of the sphincter of Oddi, which has a common embryological origin with the gallbladder,[15] and stasis of the bile duct, often dilated and assuming the function of bile storage.[16] The symptomatology most frequently reported by patients is hepatic colic,[17] followed by nausea and vomiting, dyspepsia and jaundice. In gallbladder agenesis, liver function tests are normal, but elevated liver enzymes or bilirubin may be observed, as in the case of our patient.[18] Some ultrasound scans performed on patients with vesicular agenesis, given that this is the first examination to be requested in cases of hepatic colic, contribute to vesicular lithiasis or a sclero-atrophic vesicle appearance, these misinterpretations are due to the radiologist considering periportal tissue, subhepatic peritoneal folds, duodenum or liver lesions as vesicular lithiasis, explaining the symptomatology reported by the patient.[19] These diagnostic errors, due to the appearance of the ultrasound, highlight the importance of Bili MRI in avoiding unnecessary surgery.[17] Bili MRI can also be used to confirm or rule out an ectopic location of the gallbladder, as in our case, where it is very useful, showing an empty vesicular compartment with no other ectopic location. Despite the development of imaging techniques, intraoperative diagnosis remains the most frequent circumstance of discovery. It is difficult to make a preoperative diagnosis in symptomatic subjects.[20] Malde has shown the need for further radiological examination, such as Bili MRI, CT scan, endoscopic retrograde cholangiopancreatography (ERCP) and endoscopic ultrasound, if the gallbladder is not visualized on ultrasound.[21] When laparoscopic surgery is performed, the risk of iatrogenic injury is high, especially for a surgeon with little experience of hepatobiliary surgery. Dissection can be performed in search of ectopic gallbladder locations, which are usually the falciform ligament, the leaflets of the lesser omentum, and intra- and retrohepatic locations. Intraoperative ultrasonography and cholangiography can be used to search for an ectopic gallbladder.[22] And to limit the risk of iatrogenic injury such as wounds to the biliary tree liver and hepatic vessels caused by dissection. According to some authors, this type of dissection is dangerous and should be avoided.[23] Post-operative exploration includes a CT scan of the abdomen and pelvis, which confirms the absence of a gallbladder and allows a search for an ectopic gallbladder, and at best a Bili MRI.[24]

Once the diagnosis has been made preoperatively, no treatment is necessary if the patient is asymptomatic. However, in patients with symptomatic biliary disease, prescription of antispasmodics can improve symptoms,[22] Sphincterotomy may be performed if conservative treatment is ineffective.[25] In the literature, there is no guideline for the management of symptoms associated with gallbladder agenesis. Similarly, there are no guidelines for follow-up, but abdominal ultrasound is sufficient to look for dilatation of the bile ducts.[26]

Conclusion

Agenesis of the gallbladder is a rare malformation that can mimic vesicular lithiasis or cholecystitis. The challenge is always to establish the diagnosis of GA before the patient undergoes unnecessary laparoscopic or laparotomy surgery. We must not hesitate to ask for a Bili MRI if the diagnosis is suspected preoperatively, or an ultrasound scan with intraoperative cholangiography confirming the vacuity of the vesicular loge and allowing us to look for an ectopic localization.

Acknowledgments

None.

Conflicts of interest

The authors declare no conflicts of interest.

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