

CASE REPORT

Congenital agenesis of the gallbladder: a UK case report

Jenna L. Scobie* and Simon R. Bramhall

General Surgery, Hereford County Hospital, Hereford, UK

*Corresponding address. Hereford County Hospital, Postgraduate Medical Centre, Stonebow Road, Hereford HR1 2ER, UK.
Tel: +44-(0)-7725947662; Fax: 01432 364102; E-mail: jenna.scobie@nhs.net

Abstract

Congenital absence of the gallbladder has a reported incidence between 0.013 and 0.075% (Singh *et al.*, Congenital absence of the gallbladder. *Surg Radiol Anat* 1999; 21: 221–4). With fewer than 500 cases reported in the literature, it is a well-recognised yet rare embryological malformation. Gallbladder agenesis becomes symptomatic in almost a quarter of cases mostly presenting as cholecystitis or cholecystolithiasis. In this case report, a 24-year-old Caucasian male presented with intermittent right-sided abdominal pain with no associated symptoms. On a background of no past medical history, his pain was presumed to be biliary colic. The rarity of these case reports shows that there is a lack of awareness of gallbladder agenesis when thinking of differential diagnoses. There has also been no conclusive gold standard investigation decided, upon which results are satisfactory enough to avoid surgery. Therefore, as in this case report, this group of patients often undergo unnecessary operations as often the condition is not considered and precise diagnosis pre-operatively is very difficult.

INTRODUCTION

A well-recognised yet rare embryological malformation, gallbladder agenesis becomes symptomatic in 23% of cases [1], which mostly present as cholecystitis or cholecystolithiasis. With imaging becoming more readily available, the incidence of agenesis is increasing. It is typically more common in females at a ratio of 3:1 and often presents in the 2nd and 3rd decades. Most cases are sporadic (around 70%), and there is very little literature on any familial links [2]. The pathogenesis of this abnormality of embryological development is as of yet unknown.

CASE REPORT

A 24-year-old male patient was admitted with a slightly atypical presentation of biliary colic. Investigations, whilst inconclusive, also indicated that the cause could be gallstones. On a background of no previous medical or surgical history, a laparoscopic exploration of the abdomen identified that there was no gallbladder, and this was subsequently confirmed with magnetic resonance cholangiopancreatography (MRCP). The patient wished for

further investigation of his symptoms and went on to have upper gastrointestinal (GI) endoscopy.

The patient presented with a 3-day history of intermittent abdominal pain starting in the right iliac fossa but moving to the right upper quadrant (RUQ). He suffered 3–4 episodes per day of sharp pain, which was worse on movement. He had no other associated symptoms, and on examination, the abdomen was soft with right-sided tenderness.

He had no past medical history of note and was usually fit and well.

An ultrasound of the abdomen (Fig. 1) reported the gallbladder not positively identified, but there is significant acoustic shadowing suggestive of a gallbladder contracted around gallstones. The common bile duct measured 5.9 mm with no biliary tree dilatation.

Laboratory testing revealed bilirubin level 35 mg/dL, ALT 114 IU/L and otherwise normal.

Patient was consented for a laparoscopic exploration and cholecystectomy to treat suspected biliary colic secondary to gallstones. At operation, no gallbladder could be identified either in the correct place or an ectopic site, and therefore the operation was abandoned.

Received: December 23, 2015. Revised: April 27, 2016. Accepted: May 2, 2016

© The Author 2016. Published by Oxford University Press.

This is an Open Access article distributed under the terms of the Creative Commons Attribution Non-Commercial License (<http://creativecommons.org/licenses/by-nc/4.0/>), which permits non-commercial re-use, distribution, and reproduction in any medium, provided the original work is properly cited. For commercial re-use, please contact journals.permissions@oup.com

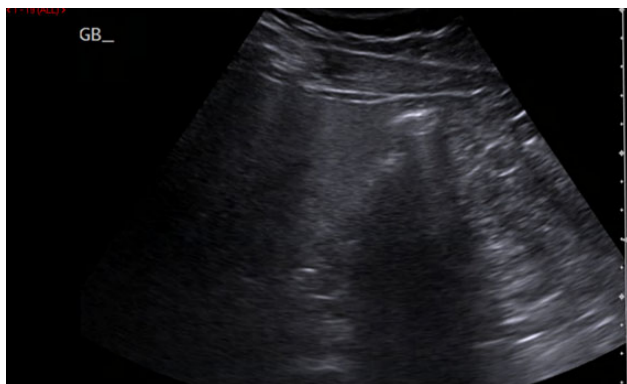


Figure 1: Ultrasound scan indicating no confirmed gallbladder but acoustic shadowing usually suggestive of a gallbladder contracted around gallstones.

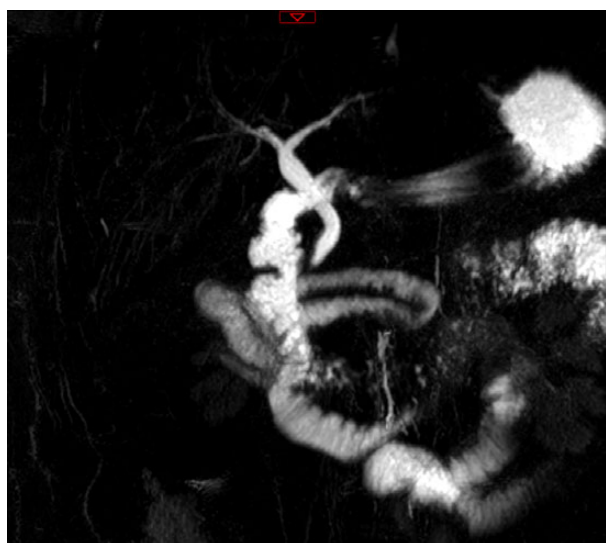


Figure 2: MRCP indicating no gallbladder positively confirmed and no duct dilatation or stones present.

An MRCP (Fig. 2) was done post-operatively, which showed no significant intra-hepatic biliary dilatation or hepatic lesions. No calculi were identified in the common hepatic duct, and no gallbladder was identified. The deranged liver function tests improved throughout admission, and he was discharged with follow-up upper GI endoscopy as the patient wished for further investigation of his symptoms.

DISCUSSION

In individuals with gallbladder agenesis, the bile duct system often remains normal. There are several classification methods; the most commonly used being the Bennion *et al.* [3], which divides cases into three groups: (i) multiple foetal malformations, (ii) asymptomatic and (iii) symptomatic. These groups comprise 12.9% (rising to 30% if including biliary atresia), 31.6% and 55.6% of cases, respectively. The symptomatic group includes varying symptoms, which would be expected from a present gallbladder such as jaundice and its associated features of RUQ pain, onset of pain after fatty foods, dyspepsia and vomiting.

There is no conclusive evidence that directs which imaging is best to carry out to confirm diagnosis. The problem is often

misdiagnosed with the result being an unnecessary operation. One study has found that a CT scan was the most successful at correct pre-operative diagnosis [4]. However, this study had a small sample size compared with others that stated USS and MRCP are most beneficial [5]. The only specific and clear pathway identified in research for this case report is the Malde algorithm. Malde suggests that if an ultrasound scan does not clearly identify the gallbladder, the next most appropriate investigation in order of accuracy are MRCP, CT and ERCP, respectively—depending on what is available in the clinical setting. Interestingly, he further suggests that if results of imaging remain to be inconclusive, they should be repeated again once the acute phase of the illness or symptoms have resolved [6]. Ultrasound scanning is not sufficient alone as there are multiple variables affecting the quality of scans produced, for example, the habitus of the individual. Other literature suggests that hepatobiliary iminodiacetic acid scans are not helpful as in cases of cystic duct obstruction there may also be the incorrect appearance of gallbladder agenesis [2].

Due to unawareness of agenesis, many cases still end up having open surgery for a concrete diagnosis. During exploration of the abdomen, it is important to exclude an ectopic gallbladder with common variations including placement being intra-hepatic, left-sided, within the falciform ligament or even retroperitoneal [7]. Even after surgical exploration, a further scan is often advised for re-confirmation of diagnosis, although there are divided opinions on whether this should be an ultrasound or MRCP [8].

Management is conservative and often with the use of smooth muscle relaxants; however, sphincterotomy has been successful in relieving symptoms in a small number of reported cases [2]. This may be due to the common bile duct dilating as a means of storing bile in those who do not have a gallbladder. Thus, they have a higher sphincter of the Oddi pressure, which could explain the relief by sphincterotomy [9].

Whilst previously non-visualization of the gallbladder prompted conversion to open surgery, developments in imaging and increasing numbers of reports on agenesis have now deemed theatre unnecessary with a moving preference towards non-invasive investigation [10].

CONCLUSION

The diagnosis of gallbladder agenesis should be considered in cases which present atypically or when diagnosis of biliary symptoms is inconclusive from gold standard investigations. However, correct diagnosis remains difficult pre-operatively; therefore, the pros and cons of repeated investigations versus immediate operation should be considered.

ACKNOWLEDGEMENTS

S.R.B. critically revised the manuscript for important intellectual content and made the final approval of the version to be published.

CONFLICT OF INTEREST STATEMENT

None declared.

FUNDING

There is no funding to report for this publication.

ETHICAL APPROVAL

No ethical approval is required.

CONSENT

I can confirm written patient consent has been obtained for the publication of this case report and the figures within.

GUARANTOR

Guarantor for this case report is J.L.S.

REFERENCES

1. Serour F, Klin B, Strauss S, Vingrad I. False-positive ultrasonography in agenesis of the gallbladder: a pitfall in the laparoscopic cholecystectomy approach. *Surg Laparosc Endosc* 1993;**3**:144–6.
2. Kasi PM, Ramirez R, Rogal S, Littleton K, Fasanella K. Gallbladder agenesis. *Case Rep Gastroenterol* 2011;**5**:654–62.
3. Bennion RS, Thompson JE, Tompkins RK. Agenesis of the gallbladder without extrahepatic biliary atresia. *Arch Surg* 1988;**123**:1257–60.
4. Cho CH, Suh KW, Min JS, Kim CK. Congenital absence of gallbladder. *Yonsei Med J* 1992;**33**:364–7.
5. Orlando R, Candiani F, Lirussi F. Agenesis of the gallbladder associated with Gilbert's syndrome. *Surg Endosc* 2001;**15**:757.
6. Malde S. Gallbladder agenesis diagnosed intra-operatively: a case report. *J Med Case Rep* 2010;**4**:285.
7. Sherson ND. The absent adult gallbladder. *Aust N Z J Surg* 1970;**39**:225–61.
8. Castorina S, Scilletta R, Domergue J. Gallbladder agenesis: laparoscopic views of a significant diagnostic challenge. *Surg Radiol Anat* 2014;**36**:619–20.
9. Meshkinpour H, Mollot M, Eckerling GB, Bookman L. Bile duct dyskinesia: clinical and manometric study. *Gastroenterol* 1984;**87**:759–62.
10. Balakrishnan S, Singhal T, Grandy-Smith S, El-Hasani S. Agenesis of the gallbladder: lessons to learn. *JLS* 2006;**10**: 517–9.