CASE REPORT

Rare case of gallbladder agenesis presenting with pancreatitis

Luke Thornton, Yan Li Goh, Mark Lipton, Andrew Masters

Arrowe Park Hospital,
Merseyside, UK

Correspondence to
Yan Li Goh,
yanli.goh@doctors.org.uk

Accepted 10 July 2016

SUMMARY

Gallbladder agenesis (GA) is a rare congenital abnormality with an incidence of 0.01–0.09%. Majority of GA exist alone although it can be associated with other systemic malformations involving the gastrointestinal, genitourinary, cardiovascular and skeletal systems. It is thought that biliary and pancreatic pathologies coexist and this is the second case reported in the literature of GA presenting with pancreatitis.

BACKGROUND

Gallbladder agenesis (GA) was first reported by Bergman in 1702 and has since been described in 415 case reports in the literature.¹ This rare congenital anomaly has an approximate overall incidence of 0.01–0.09%.² Majority of GA exist alone although GA can be associated with congenital malformations of other systems involving the gastrointestinal (imperforate anus, tracheoesophageal fistula), genitourinary (abnormal internal genitalia, renal agenesis), cardiovascular (septal defects, single umbilical artery) and skeletal systems (cleft lip and palate, vertebral anomalies).²

It is thought that biliary tract and pancreatic pathologies coexist but this is the second case of GA and pancreas divisum associated with pancreatitis reported in the literature. The first case reported by Amadon in 1933 was GA associated with chronic pancreatitis.³

CASE PRESENTATION

A 39-year-old male who is otherwise fit and well initially presented to an admitting hospital with epigastric pain and nausea. On examination, he was very tender in the epigastrium with no signs of peritonism. His only previous surgery included tonsillectomy as a child. He smokes e-cigarettes and drinks ~8 units of alcohol per week.

His blood tests on admission showed an elevated white cell count of $15 \times 10^9/L$, C reactive protein of 63 mg/L and amylase of 1500 IU/L. His liver function test was normal. Abdominal ultrasound scan performed was unable to visualise the gallbladder as it was thought to be contracted, as shown in (figure 1). MR cholangiopancreatography (MRCP) reported features consistent with acute pancreatitis. The gallbladder could not be identified and there was mild prominence of the common bile duct but no intraductal filling defects.

The patient was referred to our hospital for consideration of laparoscopic cholecystectomy. A repeat in-house ultrasound scan reported a collapsed

Figure 1 Ultrasound scan showing the collapsed gallbladder.

gallbladder with gallstones. The patient underwent laparoscopy with a view to a laparoscopic cholecystectomy 2 weeks later. Intraoperative findings showed an absent gallbladder and cystic duct as shown in figure 2. There were no features of previous inflammation in the right upper quadrant (RUQ). These images were reviewed retrospectively by our in-house radiologist after the laparoscopy which confirmed an absent gallbladder. A repeat in-house MRCP performed postoperatively confirmed GA and pancreas divisum, as shown in figure 3. The patient was referred to the hepatobiliary team for consideration of endoscopic retrograde cholangiopancreatography (ERCP) and sphincterotomy of the accessory papilla. He is well and asymptomatic at present.

DISCUSSION

This patient has two congenital abnormalities, GA and pancreas divisum, both could contribute to his presenting diagnosis of pancreatitis. The incidence of GA is 0.01–0.09%. The liver bud derived from the embryonic foregut divides into two to form the primordium of the liver and the primordium of the gallbladder and cystic duct during the fourth week of embryological development in utero. Vacuolation of the gallbladder and cystic duct occurs by week 7. Failure to develop at any stage results in GA.² Pancreas divisum, on the other hand, is a congenital abnormality when the ventral and dorsal components of the pancreatic ducts fail to fuse at week 7 leading to the main duct of Wirsung draining through the small accessory papilla. It occurs in 4% of the population.⁴

Patients with GA can be symptomatic or asymptomatic. Classification of GA by Tang *et al*⁵ is shown below:

CrossMark

To cite: Thornton L, Goh YL, Lipton M, *et al*. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-216510

Figure 2 Laparoscopy demonstrating an absent gallbladder and cystic duct.

Type 1 Symptomatic

- A. With fatal congenital malformations
- B. Without fatal congenital malformations

Type 2 Asymptomatic.

If symptomatic, these patients most commonly present with RUQ pain (96%), nausea and vomiting (15%); reduced appetite (16.7%) and diarrhoea (12%) (Tang). The patient in this case has two congenital abnormalities that could contribute to the presentation of pancreatitis—two theories have been proposed:

1. The increase in bile pressure and lack of protective mucus in an absent gallbladder causes pancreatitis³
2. Stenosis of the accessory duct of Santorini that occurs during failure of fusion in pancreas divisum.⁴

As in this case, the diagnosis of GA is challenging and can lead to misleading radiological assessments and unnecessary surgery. On ultrasound scan, severe acute or recurrent cholecystitis can lead to a shrunken gallbladder; poor preparation can

lead to poor distension of the gallbladder which could be confirmed as an absent gallbladder. Opacifications thought to represent gallstones could actually be intestinal gas or hepatic peritoneal folds producing false-positive results.

Malde's decision tree for investigating GA recommends that either MRCP, a CT scan, ERCP or endoscopic ultrasound scan should be performed when the gallbladder cannot be identified or is shrunken on ultrasound scan. In this case, MRCP was unable to confirm the diagnosis of GA preoperatively. Extensive dissection intraoperatively to look for the gallbladder is discouraged; therefore another MRCP was performed postoperatively to further confirm the diagnosis of GA given the intraoperative findings.⁶

Learning points

- ▶ The diagnosis of GA and pancreas divisum is rare with an incidence of less than 1%. Clinicians need to be aware of this and perform thorough pre-operative imaging to avoid unnecessary surgery.
- ▶ Clinicians should be aware that investigation for GA can be challenging and lead to misleading radiological assessment and interpretation as demonstrated in this case.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Latimer EO, Mendez FL Jr, Hage WJ, *et al.* Congenital absence of gallbladder: report of three cases. *Ann Surg* 1947;126:229–42.
- 2 Turkel SB, Swanson V, Chandrasoma P, *et al.* Malformations associated with congenital absence of the gallbladder. *J Med Genet* 1983;20:445–9.
- 3 Amadon PD. Agenesis of the gall bladder associated with pancreatitis. *Am J Surg* 1933;19:263–7.
- 4 Warshaw AL, Richter JM, Schapiro RH. The cause and treatment of pancreatitis associated with pancreas divisum. *Ann Surg* 1983;198:443–52.
- 5 Tang LM, Wang XF, Ren PT, *et al.* The diagnosis of gallbladder agenesis: two cases report. *Int J Clin Exp Med* 2015;8:3010–16.
- 6 Malde S. Gallbladder agenesis diagnosed intra-operatively: a case report. *J Med Case Rep* 2010;4:285.

Figure 3 MR cholangiopancreatography confirming an absent gallbladder and cystic duct.

Copyright 2016 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow