

Agenesis of the gallbladder



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Agenesis of the gallbladder

Agenesis of the gallbladder is a rare anomaly that is usually asymptomatic, but sometimes the patients may have symptoms compatible with a biliary disorder like cholelithiasis. Its preoperative diagnosis is often difficult, especially if displastic cyst is associated and simulates the gallbladder.

When the diagnosis is doubtful its confirmation and treatment of displastic cyst require open surgery, careful dissection of the common bile duct to avoid biliary lesions and intraoperative ultrasonography or cholangiography to be performed to exclude other associated anomalies.

The Authors describe the case of agenesis of gallbladder and displastic cyst associated and comment on its clinical, diagnostic, and therapeutic aspects.

Key words: Agenesis, Displastic cyst, Cholecistectomy, gallbladder.

Introduction

Agenesis of the gallbladder is a rare congenital condition in adult patients, especially in absence of other malformations of the biliary tree ¹.

The incidence rate is from 0,09 to 0,016% in necropsies. In 40-70% of cases this anomaly is associated with other gastrointestinal, skeletal, cardiovascular, and genitourinary malformations ².

The absence of gallbladder, with normal bile ducts, occurs in 13 to 65 per 100,000 population, probably from failure of the gallbladder bud to develop or vacuolize in the 4th week of intrauterine life ^{3,4}.

Cases diagnosed through operations present a female predominance of 3:1, although in autopsies the proportions are equal between the genders.

Usually adult are asymptomatic but they may present abdominal pain in the upper right quadrant and more rarely jaundice ⁵.

To our knowledge, this is the first case of gallbladder agenesis and displastic cyst combined. Such association

may be incidental or they could represent different expression of the same congenital anomaly of the ductal plate.

Case report

A 58-year-old white woman, who have had an episode of colic pain in the upper right quadrant of the abdomen over the past ten days, was admitted in Gastroenterology Department. She was seen in consultation because ultrasound exam demonstrated normal gallbladder and displastic cysts of the liver. She was referred to our service and her physical examination was normal, an other ultrasonography confirmed the previous exam, on the contrary preoperative CT scan described two main displastic cysts of the liver with normal biliary tree and two others small displastic cysts on the 4th segment of the liver. The patient was operated on by laparotomy, laparoscopy was excluded because of doubtful diagnosis. The exploration of the peritoneal cavity was normal and one big displastic cyst, with gallbladder shape, was found on inferior aspect of the 4th and 5th segments of the liver. The common bile duct was dissected from confluence of the hepatic duct to the duodenum. Intraoperative ultrasonography and cholangiography, via puncturing the bile duct using a fine needle, did not locate gallbladder or cystic duct and demonstrated normal intra and extrahepatic biliary tree. The displa-

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sic cyst, separated by an avascular and soft tissue from the liver, was removed and the two small cysts of the 4th segment was unroofed. The patient was discharged after 13 days, without any complications. She stays well and has not other episode of colic pain after 2 months of follow-up.

Discussion

During liver development hepatoblasts differentiate into hepatocytes and biliary epithelial cells (BEC) that delineate the intrahepatic, extrahepatic bile ducts and the gallbladder. The transcription factors that control the development of the biliary tract are unknown, but the oncut transcription factor HNF6 is expressed in hepatoblasts and in the gallbladder primordium. Clotman shows, analyzing the phenotype of *Hnf6*(-/-) mice without gallbladder and abnormal biliary tract, that HNF6 is expressed in the BEC and stimulate the *Hnf1beta* promoter, essential for differentiation and morphogenesis of the biliary tract. He concludes that bile duct development is controlled by a *HNF6*→*HNF1beta* cascade⁶.

Agenesis of gallbladder is rarely associated with other malformations^{5,7}.

Farrant has found only 2 on 35 babies, studied by ultrasound scanning, with agenesis of gallbladder and biliary atresia combined⁸. Approximately 220 cases have been reported in the literature. Most of these are from necropsy studies and many of these were newborns with more serious anomalies. Adult patients with agenesis of gallbladder are 36-46 years old⁵. Lithiasis of the common bile duct is present in 25-50% of cases² and they have not any characteristic symptomatology, but some patients develop symptoms related to biliary tree disease, some-

times due to calculi in the common bile duct^{7,9}.

Richards and coworkers have studied the symptomatology in 44 patients: dyspepsia was presents in 15 (34%); 24 (54%) had colic pain suggestive of biliary disease, 12 (27%) had jaundice, due to common duct stones in eight⁴.

The preoperative diagnosis is extremely difficult and the absence of the gallbladder is often an intraoperative finding and the diagnosis will be sure only after explorative surgery.

Imaging techniques have sensitivity less than 100% for identification of this organ.

Fischella and Belli underline the inaccuracy of currently used diagnostic tests^{2,10}.

Although ultrasonography is considered to have a 95 per cent sensitivity for the diagnosis of cholelithiasis, a small contracted gallbladder associated with stones and chronic cholecystitis will be difficult to visualize with ultrasonography^{3,5}. By ultrasonographic exploration performed on 1823 patients Senecail found morphologic variations and abnormalities in more than 33% of gallbladders, topographic ones in about 3.5% of observations and only 3 cases of real duplication of the gallbladder¹¹. Ectopic gallbladder locations include intrahepatic, lesser omentum, retroperitoneal, within the falciform ligament, retroduodenal, and retrohepatic areas¹². Displasic cyst on inferior aspect of the fourth and fifth segment of the liver, that simulate normal gallbladder as in our patient, can cause diagnostic pitfalls.

The diagnosis will be confirmed by explorative laparotomy with carefull dissection of the whole common bile duct and intraoperative ultrasonography or cholangiography are necessary to exclude other associated biliary tract malformations or aberrant localization^{7,9}. Gotohda thinks that, when gallbladder is not visualized by imaging techniques, it may be better to perform laparoscopy for observation before performing laparotomy, in order

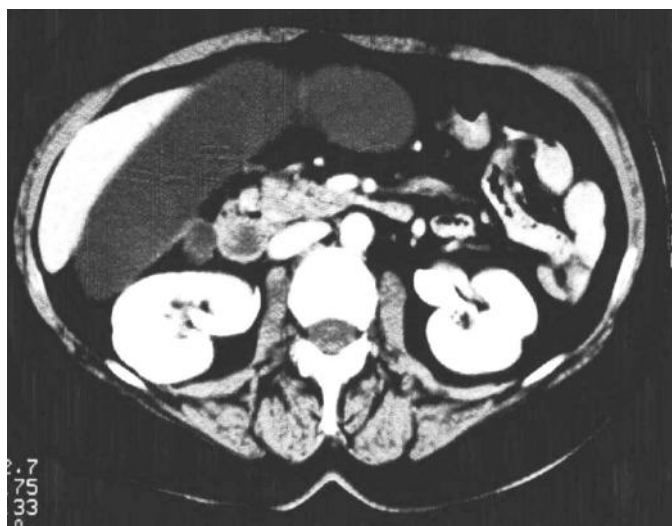


Fig. 1: CT Abdomen: displasic cyst of the liver simulates the gallbladder.



Fig. 2: Intraoperative finding: agenesis of gallbladder and presence of big displasic cyst.

to reduce surgical stress¹³. For unexplained reasons our patient, as well as majority of cases in literature³, became asymptomatic after surgical exploration and removal of the displastic cyst, the only possible cause of pain, probably due to distension of its wall.

We conclude that a perfect knowledge of the normal anatomy and congenital variation of biliary tract is essential in prevention of operative bile duct injury¹⁴. When there are doubts about diagnosis on biliary disease and displastic cysts are associated, laparoscopic approach might be risky, because it is necessary carefully dissection of the common bile duct, ultrasonography and colangiography exams are mandatory¹ and open surgery should be considered before undesirable complications occur¹⁶.

Riassunto

L'agenesia della colecisti è una anomalia rara e generalmente asintomatica, ma talvolta i pazienti possono avere sintomi compatibili con patologie della via biliare come la colelitiasi. La sua diagnosi preoperatoria è spesso difficile, soprattutto se associata ad una cisti displasica che simula la colecisti.

Quando la diagnosi è dubbia la sua conferma e il trattamento della cisti displasica richiedono un intervento chirurgico a cielo aperto, e per evitare lesioni è necessaria una attenta dissezione della via biliare e l'esecuzione di una ecografia e colangiografia intraoperatoria per escludere altre anomalie associate.

Gli Autori descrivono un caso di agenesia della colecisti con associata una cisti displasica e commentano gli aspetti clinici, diagnostici e terapeutici.

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