Agenesis of the gallbladder

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Introduction

Agenesis of the gallbladder is a rare congenital condition in adult patients, especially in the 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 displasic 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 displasic 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 displasic cysts of the liver with normal biliary tree and two others small displasic 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 displasic cyst, with gallbladder shape, was found on inferior aspect of the 4th and 5th segments of the liver. The common bile duct was dissec-
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 onecut 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 cascade 6.

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 combined 8. 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 old 5. Lithiasis of the common bile duct is present in 25-50% of cases 2 and they have not any characteristic symptomatology, but some patients develop symptoms related to biliary tree disease, sometimes 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 eight 4.

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 Senecal 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 gallbladder 11. Ectopic gallbladder locations include intrahepatic, lesser omentum, retroperitoneal, within the fallicorn ligament, retroduodenal, and retrohepatic areas 12. 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 careful 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
to reduce surgical stress. For unexplained reasons our patient, as well as majority of cases in literature, 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 injury. 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 mandatory and open surgery should be considered before undesirable complications occur.

**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.

**Bibliografia**


