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Pediatrics

Incidentally detected gallbladder agenesis in a child: the importance of identifying anatomic structure

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The absence of a gallbladder is a very rare anomaly. While it is usually asymptomatic, it can cause biliary colic symptoms. For these reasons, gallbladder agenesis can be misdiagnosed as a hepatobiliary disease and is diagnosed correctly after surgery. This condition may also be detected through an autopsy for other causative diseases. Abdominal ultrasonography is used as a diagnostic method to detect gallbladder agenesis. Hepatobiliary scintigraphy, magnetic resonance cholangiopancreatography, and endoscopic cholangiopancreatography are also used to make a more accurate diagnosis. In the emergency room, however, gallbladder agenesis can still be misdiagnosed as acute or chronic cholecystitis, leading to the detection of gallbladder agenesis in the operating room. Although some cases of gallbladder agenesis detected in adults during surgery have been reported in Korea, there are no reports of gallbladder agenesis in pediatric patients to date. This paper reports a case of gallbladder agenesis in a symptomatic child that was detected incidentally by a radiographic examination.

Keywords: Gallbladder agenesis; Magnetic resonance cholangiopancreatography; Pediatrics

INTRODUCTION

Gallbladder agenesis is a highly rare disease with an incidence of 0.01-0.02%.[1,2] Approximately 500 cases of gallbladder agenesis have been reported worldwide so far. Gallbladder agenesis is difficult to preoperatively diagnose in most cases. As congenital gallbladder agenesis can exhibit the general symptoms of hepatobiliary diseases and is detected after surgery, an accurate differential diagnosis is important. Cases of gallbladder agenesis detected through an autopsy have also been reported.[3] Although cases of gallbladder agenesis detected in adults during surgery have been reported in Korea, gallbladder agenesis in pediatric patients has not been reported. We report, here, a case of gallbladder agenesis detected incidentally, in addition to presenting a literature review.

CASE REPORT

A 13-year-old boy was visited emergency room with symptom of periumbilical abdominal pain, vomiting, nausea. He did not have underlying disease, operation history and family history. On physical examination, he did not have fever or jaundice. His abdomen was soft and flat with normoactive bowel sounds. Definite focal tenderness and rebound tenderness were absent on the whole abdomen and no abdominal mass was palpable. Routine blood test results were as follows: white blood cells, 7,370/mm^3 (neutrophils, 56%; lymphocytes, 35%; monocytes, 5%); hemoglobin, 14.5 mg/dL; hematocrit, 39%; and platelet, 385,000/mm^3. The blood chemistry test results were all normal with the following values: total bilirubin, 0.9 mg/dL; direct bilirubin, 0.2 mg/dL; aspartate aminotransferase, 22 IU/L; alanine aminotransferase,

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Capsule Summary

What is already known in the previous study

Gallbladder agenesis is very rare disease that can cause biliary colic symptoms. Hence, it can be misdiagnosed as a hepatobiliary disease and is diagnosed correctly after surgery.

What is new in the current study

Although many cases of gallbladder agenesis detected in adults during surgery have been reported in Korea, there are no reports of gallbladder agenesis in pediatric patients to date. In this case, gallbladder agenesis was detected incidentally by a radiographic examination in a symptomatic child.

12 IU/L; amylase, 69 U/L; lipase, 13 U/L; and alkaline phosphatase, 426 IU/L. No abnormal findings were detected on plain abdominal radiographs. An emergency abdominal ultrasonography (Fig. 1) performed to diagnosis causes of abdominal pain. In abdominal ultrasonography, we could not find gallbladder in expected anatomical lesion and we found that intrahepatic and extrahepatic ducts were found to be slightly dilated. In abdominal computed tomography (Fig. 2), there were no visible gallbladder and no other bowel and sold organ abnormality. Magnetic resonance cholangiopancreatography was performed to identify gallbladder agenesis and hepatic duct (Fig. 3). The patient was eventually diagnosed with gallbladder agenesis and minimal dilatation of intra and extrahepatic duct system. After supportive treatment, the patient is currently in good condition and is being regularly followed at the outpatient department. The patient's legal guardians provided written informed consent.

DISCUSSION

Gallbladder agenesis, which occurs in the absence of hypoplasia of the extrahepatic biliary system, is a highly rare disorder with an incidence of 0.01-0.02%.[1-3] Gallbladder agenesis can be diagnosed at all ages, and the mean age at the time of diagnosis is 46 years.[4] Although the cause for the development of this condition is not known, disruption of embryonic development is considered to be the main cause of gallbladder agenesis. During this developmental period, the liver, biliary tract, gallbladder, and biliary ducts develop in the hepatic diverticulum. The hepatic diverticulum grows and connects with the intestines to form the extrahepatic biliary system. As this connection becomes narrower, the extrahepatic biliary system forms, followed by the biliary ducts and gallbladder. When the formation of the biliary ducts and the gallbladder is disrupted during this developmental stage, gallbladder agenesis can result without any dysplasia of the extrahepatic biliary system. As

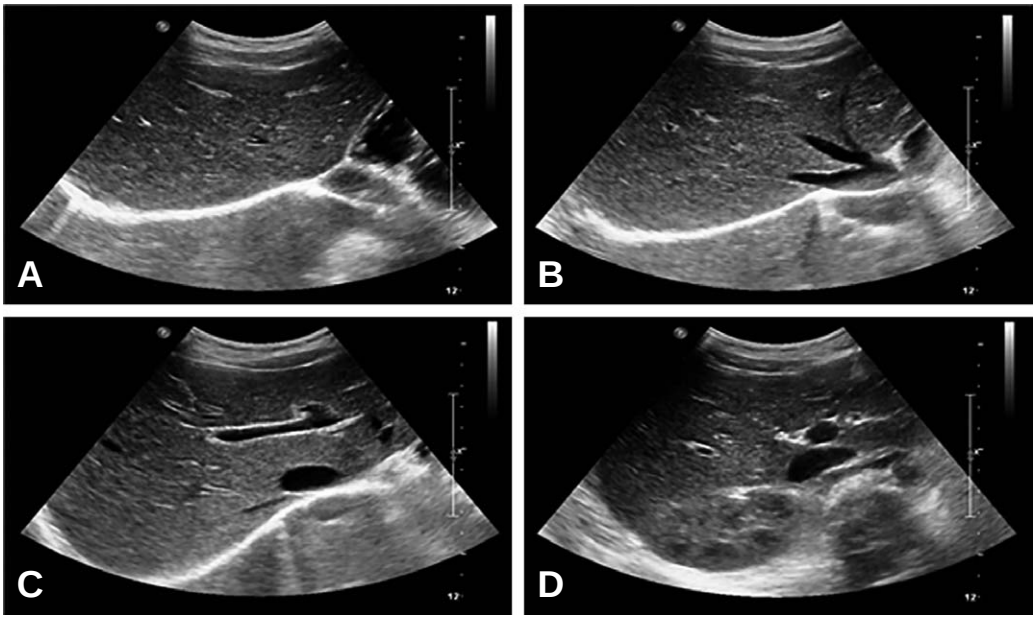

Fig. 1. (**A**-**D**) Abdominal ultrasonogram showing no visible gallbladder in the expected anatomical regions.

some studies report that gallbladder agenesis can be familial, it is important to check whether there are other family members who have been diagnosed with this malformation.[2,3,5]

Gallbladder hypoplasia and gallbladder agenesis may be associated with extrahepatic biliary atresia or cystic fibrosis.[6] Bennion reported that patients with gallbladder agenesis can be largely divided into three groups.[1,5] The first group includes patients with gallbladder agenesis accompanied by other major defects and malformations such as cardiac septal defects, duodenal atresia, imperforate anus pancreatic divisum, and renal agenesis.[1] The

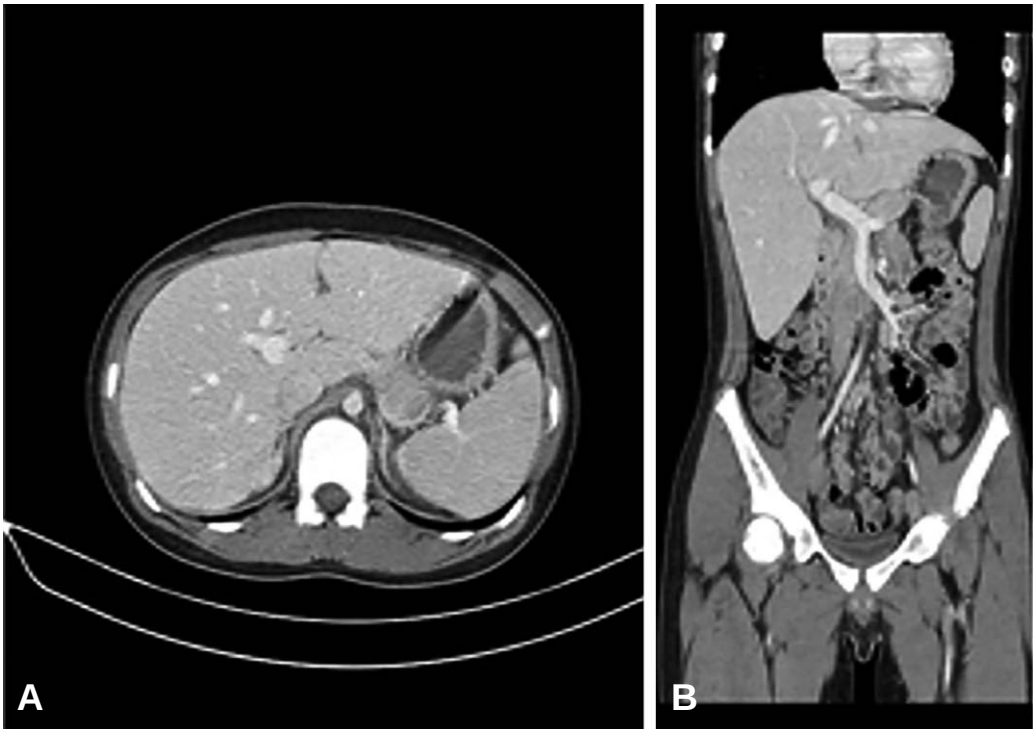

Fig. 2. (**A**, **B**) Abdominal computed tomography image showing no visible gallbladder.

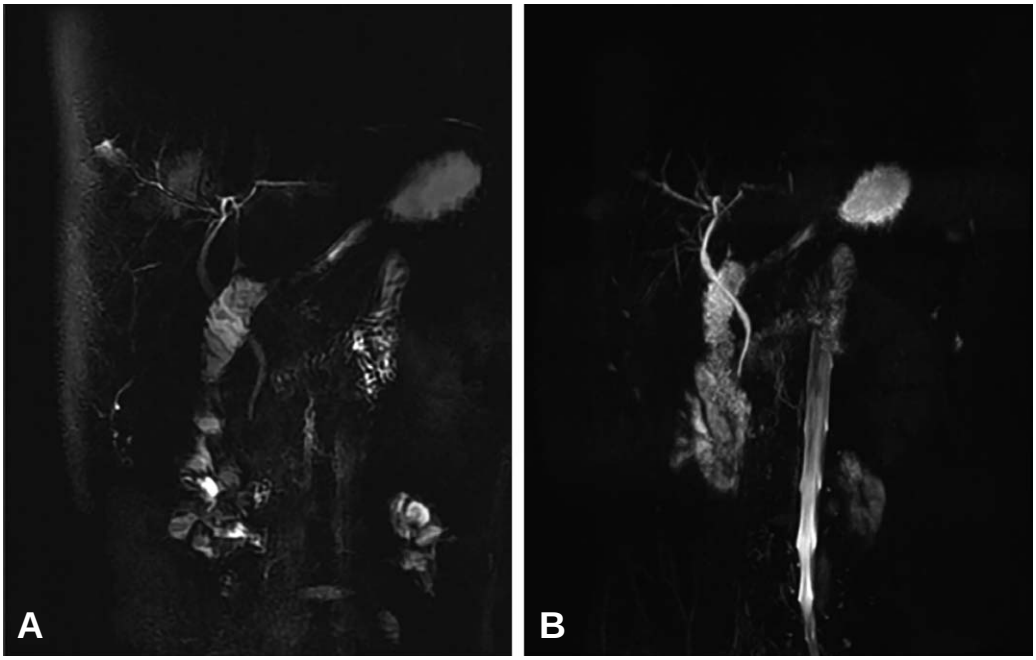

Fig. 3. (**A**, **B**) Magnetic resonance cholangiopancreatography image showing no visible gallbladder.

second group includes patients with asymptomatic gallbladder agenesis. Gallbladder agenesis in these patients are usually detected during laparotomy or an autopsy. The third group is composed of patients with symptomatic gallbladder agenesis. In this group, symptoms such as right upper abdominal pain, nausea, vomiting, and inability to digest fatty foods can be observed, while jaundice is rarely reported.[5]

The patient in this case report had symptoms of acute enteritis such as periumbilical pain, vomiting, and nausea when he was admitted to the emergency room. As whether these symptoms were caused by gallbladder agenesis or by acute enteritis could not be clearly determined, the patient can be classified into the second or third group in the above-mentioned classification. After a supportive treatment, the patient discharged with improved state. But the symptomatic gallbladder agenesis can recur at any time, so continuous follow-up monitoring is necessary even after an adult. Fortunately, there was no problem with fat digestion in his past history. If related symptoms occur, it must be identified whether they are caused by gallbladder agenesis or not. In most cases of gallbladder agenesis detected in pediatric patients are usually detected in an autopsy for infants with fetal defects accompanying the disease, within 1 year after birth. Cerebrotendinous xanthomatosis and G syndrome are syndromes characterized by multiple anomalies that may be due to gallbladder agenesis in pediatric patients. Gallbladder agenesis has also been reported in trisomy 18 and Klippel-Feil syndrome.[4]

Most pediatric patients with gallbladder agenesis are asymptomatic, and 25-50% show accompanying symptoms.[4,7] Bennion et al.[5] investigated 208 cases of symptomatic gallbladder agenesis and reported right upper abdominal pain in 91% and nausea and vomiting in 66.3% of all cases. Of the patients, 37.5% showed resistance to fatty foods, 32% showed common bile duct expansion, and 27% showed gallstones.[5]

These accompanying symptoms occur in patients with gallbladder agenesis because in the presence of gallbladder agenesis, bile excretion becomes functionally or structurally impaired, and this leads to the morphological expansion of the extrahepatic biliary system.[3] In addition to the expansion of the extrahepatic biliary system, biliary colic symptoms such as cholestasis, infection and stone formation can rarely occur owing to the sphincter of Oddi dysfunction and biliary dyskinesia.[8,9] Smooth muscle relaxants and analgesics can relieve these symptoms. For 90% of patients who do not show symptom improvements, sphincter removal can be used to achieve symptom relief.[3]

The medical imaging techniques used to diagnose gallbladder agenesis include hepatobiliary scintigraphy and endoscopic cholangiopancreatography. However, the diagnosis of gallbladder agenesis is reported to be difficult even when these techniques are used. Abdominal ultrasonography can misidentify the tissues around the liver hilar region or peritoneal folds in the lower region of the liver as a shrunken gallbladder and misdiagnose gallbladder agenesis as chronic cholecystitis. When the gallbladder is not visualized in cholecystography or hepatobiliary scintigraphy, cholecystitis is diagnosed, surgery is performed, and gallbladder agenesis is eventually detected during surgery.[3,8]

Especially in emergency rooms, abdominal pain is the cause of many patients. Symptomatic gallbladder agenesis requires differential diagnosis from acute cholecystitis, cholangitis, acute enteritis, and appendicitis. In an emergency room where radiologist does not exist at night, the emergency medical doctor will perform screening abdominal ultrasonography. Generally, in a collapsed gallbladder, physicians can see gallbladder wall through ultrasound. However, as mentioned earlier, the tissues around the liver hilar region or peritoneal folds in the lower regions of the liver can be misidentified the shrunken gallbladder. Doctors who have not experienced gallbladder agenesis think gallbladder has shrunk because of not fasting and can be misdiagnosed as other diseases. Therefore, it is important to confirm the NPO (nothing by mouse) time before abdominal ultrasound. If the gallbladder is not visible or looks like a shrunken gallbladder despite the proper NPO time, physicians should suspect gallbladder agenesis. Owing to the limitations of these diagnostic methods in emergency room, the accurate judgment of the surgeon is important to prevent unnecessary operations. If the gallbladder is not clearly identified, an accurate diagnosis must be made through repeated tests or other medical imaging tests mentioned above. Recently, magnetic resonance cholangiopancreatography has been used as a good noninvasive diagnostic tool to examine the hepatobiliary system and ectopic gallbladder.[4]

In conclusion, gallbladder agenesis is a highly rare dis-

ease that is usually asymptomatic but can cause symptoms such as right upper abdominal pain, vomiting, nausea, and jaundice. Owing to these symptoms, it is sometimes misdiagnosed as a hepatobiliary disease and is incidentally detected during surgery or in an autopsy. Therefore, accurate diagnosis of gallbladder agenesis using image tools like hepatobiliary scintigraphy, magnetic resonance cholangiopancreatography, and endoscopic cholangiopancreatography is important before surgery. The patient in this case report was suspected as having a gallbladder agenesis on abdominal ultrasonography, which was confirmed using magnetic resonance cholangiopancreatography. As gallbladder agenesis has never been reported in pediatric patients in Korea, we report a gallbladder agenesis case here with a literature review.

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CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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