

CASE REPORT

CONGENITAL AGENESIS OF THE GALLBLADDER – A RARE CONGENITAL ANOMALY

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ABSTRACT

Congenital absence of the gallbladder is rare among biliary abnormalities, and its preoperative diagnosis has been considered very difficult. Many of these patients develop a typical symptomatology that leads them to Operation Theater. If an operative procedure is done, it is better to remain at the level of laparoscopy because further surgical investigation may lead to detrimental biliary tract injuries. Establishment of a preoperative diagnosis using noninvasive imaging modalities permits to avoid a risky surgical procedure. We encountered a patient with congenital absence of the gallbladder and suggest a possible preoperative diagnosis of the abnormality.

Keywords: congenital absence of the gallbladder, preoperative diagnosis

INTRODUCTION

Congenital absence of the gallbladder is rare among biliary abnormalities. Agenesis of gallbladder (AGB) is a very rare anatomical abnormality first reported by Lemery in 1707¹ or by Bergman in 1702². The incidence in the general population is reported as 13-65 cases/100000³. In clinical series, the incidence is 0.007-0.0027%, while in autopsy series it is 0.04-0.13%⁴. AGB may be associated with other system malformations as gastrointestinal, genitourinary, cardiovascular, musculoskeletal or some congenital syndromes⁵. Since then several cases have been reported, most of which were unexpectedly found at laparotomy for cholecystolithiasis, cholecystitis or during autopsy. A preoperative diagnosis has been considered very difficult. Preoperative diagnosis includes imaging modalities that may be falsely interpreted (U/S, CT, ERCP, etc.). The main tool to establish pre- and postoperative diagnosis of AGB is MRCP. Once a surgeon meets such a situation, it is wiser to do nothing further in the operating theater and to try to establish the accurate diagnosis postoperatively mainly by using MRCP. We encountered a patient with congenital absence of the gallbladder. Here, we suggest a possible preoperative diagnosis of the abnormality, and review the literature.

CASE REPORT

The patient was 38-year-old female. The patient was diagnosed as having cholelithiasis and contracted gall bladder in previous two abdominal ultrasound examinations in other clinics. The patient was referred to our hospital for close examination. A luminal organ corresponding to the gallbladder was not detected by abdominal ultrasonography in the gallbladder fossa. However, no strong sound shadow, which appears when the gallbladder is filled with stones, was noted. As with abdominal ultrasonography, abdominal contrast CT did not reveal a luminal organ corresponding to the gallbladder in the gallbladder fossa. The gallbladder was not found in the abdominal wall or cavity. The patient underwent CT scan abdomen and pelvis in our department and GB was not visualized in abdomen on CT scans. For further evaluation MRCP study was advised and on MRCP also gall bladder was not visualized. Although the bile duct was clearly demonstrated by MRCP up to its periphery, neither the cystic duct nor gallbladder was shown. In the compressed images, abnormalities showing stones in the common bile duct were not observed. These findings, together with the absence of the gallbladder in the gallbladder fossa suggested that ectopic gallbladder was unlikely and we diagnosed the disease as con-

genital absence of the gallbladder. For about 6 months after this diagnosis, the patient has had no subjective symptoms. Thus an unnecessary laparotomy was prevented.

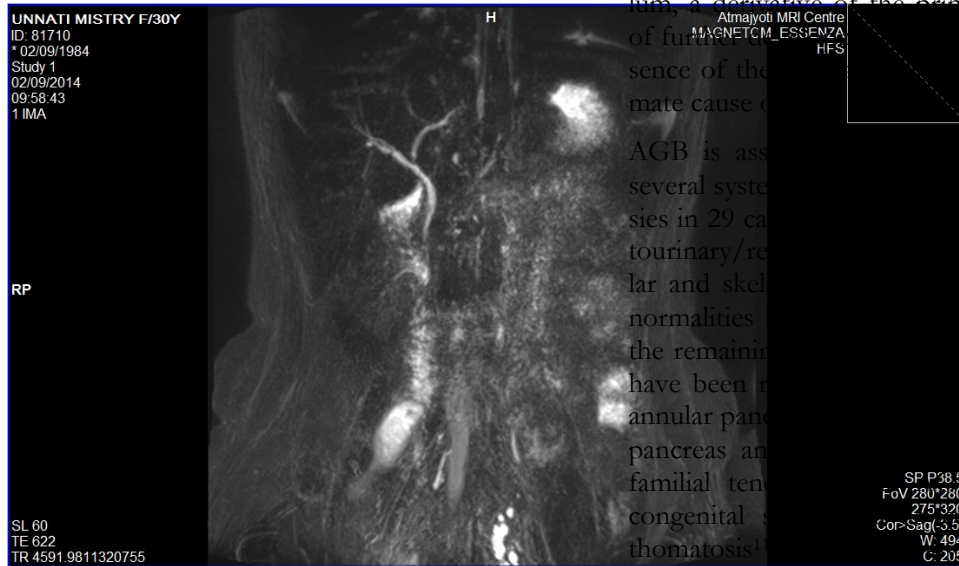


Figure 1: MRCP images showing absence of gall bladder

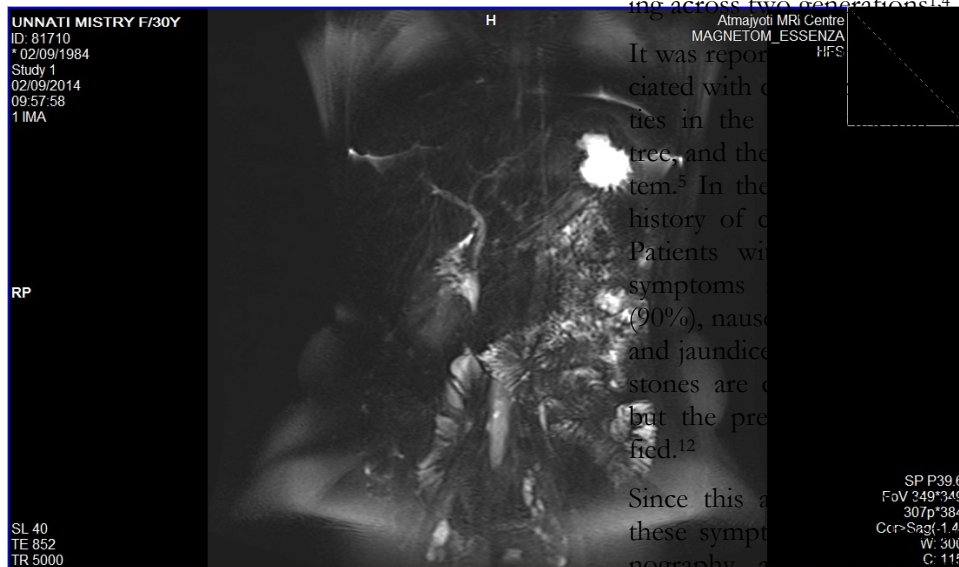


Figure 2: MRCP images showing absence of gall bladder

MRCP images are showing right & left hepatic duct combine to make the common hepatic duct (figure 1). There is absent of gall bladder & cystic duct so the common hepatic duct directly open into 2nd part of duodenum at ampulla of Vater. There is no e/o any other abnormality is seen in biliary tract (figure 2).

DISCUSSION

Clinical detection of this abnormality is incidental, and many asymptomatic patients may exist. During embryonic development, the gallbladder normally arises as a bud from the hepatic diverticulum, a derivative of the primitive foregut. Failure of further development of the primitive foregut result in complete absence of the gallbladder and cystic duct. The ultimate cause of this abnormality remains speculative.

AGB is associated with other malformations in several systems. In a review of autopsies in 29 cases of congenital malformations in the genitourinary/reproductive system, 8 had cardiovascular and skeletal abnormalities, 5 had abdominal wall and in the remaining 16 cases, AGB was alone⁸. There have been reports of syndromes such as AGB, annular pancreas, and congenital hypothyroidism⁹, or AGB, annular pancreas and congenital hypothyroidism¹⁰. There may be a familial tendency as AGB is associated with congenital syndromes such as xanthomatosis¹¹, Marfan syndrome, Klippel-Feil syndrome and trisomy 18¹¹. Some authors reported AGB as a result of thalidomide therapy¹¹. AGB may be inherited with a non-sex-linked heredity, with several familial cases observed, including across two generations¹⁴.

This abnormality is often associated with other congenital abnormalities in the biliary system, hepato-biliary system, and the gastrointestinal system.⁵ In the present case, there was no family history of congenital absence of the gallbladder. Patients with this abnormality have subjective symptoms such as upper quadrant pain (90%), nausea and vomiting (37%), and jaundice (37%). Gallstones are common cause these symptoms, but the precise cause remain to be clarified.¹²

Since this abnormality is often accompanied by gallstones, the abdominal ultrasonography and cholangiography show findings similar to cholecystitis, patients with this abnormality are often diagnosed as having cholecystitis and undergo laparotomy. In patients with this abnormality, peri portal tissues and sub hepatic peritoneal folds are sometimes revealed by ultrasonography as a thick, contracted, shrunken or scarred gallbladder, and the absence of the gallbladder by cholangiography suggests that a non-functioning gallbladder exists. In the present patient, high echo areas were observed by ultrasonography in the gallbladder fossa, but chronic cholecystitis could

not be ruled out solely by this observation, as has been reported.

Congenital absence of the gallbladder, which is usually benign, is unlikely to transform into malignant disease. It was reported that manual detailed examination during laparotomy and intra operative cholangiography are required for the diagnosis of this abnormality. It is not wise to convert a laparoscopic to an open-access procedure. It is better to remain at the level of a simple laparoscopy and to establish the accurate diagnosis of AGB post-operatively by imaging modalities, mainly by MRCP^{13,14}. However, these procedures are inadequate for the diagnosis of ectopic gallbladder.

The preoperative diagnosis of congenital absence of the gallbladder is considered difficult, and many recent studies on its diagnostic problems have indicated the need for adequate diagnostic methods.^{2,4,13}

Diagnosis of congenital absence of the gallbladder is possible using a combination of imaging techniques including MRCP.¹⁵ Patients showing no past symptoms in whom the gallbladder is not observed even by MRCP, which can visualize the intra hepatic bile duct, should be periodically observed due to the possibility of congenital absence of the gallbladder. However, several problems remain for patients with symptoms. Symptomatic patients with congenital absence of the gallbladder are often diagnosed as having cholecystitis and are subjected to treatments required for cholecystectomy. Therefore, minimally invasive therapy such as laparoscopic cholecystectomy should be chosen as the first treatment method.

In some cases, patients showed improvement after laparotomy or responses to conservative therapy despite the absence of the gallbladder. It was also reported that symptoms were improved by sphincterotomy in patients who did not respond to conservative therapy¹⁶. Due to the variety of responses of patients with symptoms, no standard treatment method has been developed, and establishing a standardized system for diagnosis may be difficult. Further studies are required to establishment of diagnostic procedures.

CONCLUSION

The patients without gallbladder are usually operated because a “false” interpretation of ultrasonography. When ultrasonography reveals a “sclerotic” gallbladder or in cases of non-visualization in other imaging modalities, the need of further preoperative investigation must be in

the surgeon’s mind. It seems that MRCP is the most accurate non-invasive diagnostic tool to establish the diagnosis of AGB and to avoid unnecessary and detrimental risky surgery when combined with the other non-invasive investigations.

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