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GALLBLADDER AGENESIS; A RARE ENTITY. ITS DIAGNOSTIC CHALLENGE AND LITERATURE REVIEW

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ABSTRACT

Agenesis of the gallbladder and cystic duct is one of the biliary system's rarest malformations, with a reported frequency of 0.007% to 0.027%. It may present as asymptomatic condition that is typically discovered as an incidental finding on imaging modalities. There are also symptomatic instances where patient may present with symptoms like biliary colic that may be linked to cholelithiasis, cholecystitis or cholangitis misleading to various investigations and treatments. The clinical diagnosis of agenesis of the gallbladder is quite difficult since it is an exceptionally infrequent congenital condition with masked clinical and radiologic characteristics. Here, we discuss the case of a 27-year-old female who arrived to the emergency department with complain of right upper quadrant pain, jaundice, and abnormal bowel habits for few months, aggraveted for past one week. An ultrasound of her upper abdomen revealed the absence of GB, which was verified by her CT scan and MRCP. Gallbladder agenesis is a relatively rare condition that could require needless diagnostic procedures and treatments. The gold standard test is the MRCP. Despite its benign nature, smooth muscle relaxants are chosen for conservative treatment in symptomatic patients. This study's objective is to discuss the most crucial aspects of gallbladder agenesis and to help clinicians in the early diagnosis detect GB agenesis as soon as possible to prevent catastrophic events and misleading to various investigations.

KEYWORDS: Gallbladder Agenesis (GA), Magnetic Resonance Cholangiopancreatography (MRCP), Biliary Colic, Ultrasound, Congenital anomaly, Hepato-biliary.

INTRODUCTION

Agenesis of the Gallbladder is an uncommon disorder with an incidence rate of 0.01–0.075%.[10] It is caused by the inability of the GB and its cystic duct to separate from the common bile duct (CBD) during embryonic development in the 3rd and 4th week of pregnancy.[3,7] Most cases are asymptomatic and the signs of biliary illness are present in roughly 23% of individuals.[1] In some cases, the gallbladder may not always be visible in the GB fossa and may be mistakenly reported during ultrasonography (USG) of the abdomen as contracted GB.[1,7,3] Based on these incorrectly reported cases, these individuals could undergo needless surgery and may suffer iatrogenic portal and biliary system damage from the extensive dissection required to identify the missing and ectopic gallbladder.[4,5] Preoperative imaging such as MRCP and EUS should also be taken into consideration to avoid adverse outcomes.[1] This study's objective is to discuss the most crucial parts of gallbladder agenesis and to help clinicians in the early diagnosis detect GB agenesis as soon as possible to prevent catastrophic events.

CASE REPORT

We present here a case of a 27-year-old female with no known co-morbid, who presented to emergency department of Liaquat National Hospital with complaints of right upper quadrant pain on and off for past few months, aggraveted for about a week. The pain was colicky in nature, intermittent, and radiating to the back and it was accompanied by nausea. These symptoms were worsened by meals and particularly by fatty food. She also had jaundice from 15 days and altered bowel habitus.

Her past medical and surgical history was unremarkable. no previous imaging was available and there was no relevant family history.

On clinical examination her skin and sclera was yellow. On abdominal examination there was slight tenderness in right upper quadrant. Her baseline investigations were done which were in normal range.

An abdominal ultrasound was performed (figure 1a, 1b, 1c), which revealed that the gallbladder was not visualized despite the fact that the patient had been

fasting for prolong time. The possibility of contracted, atrophic or congenitally absent gallbladder was raised.(fig;), rest of the scan was unremarkable.

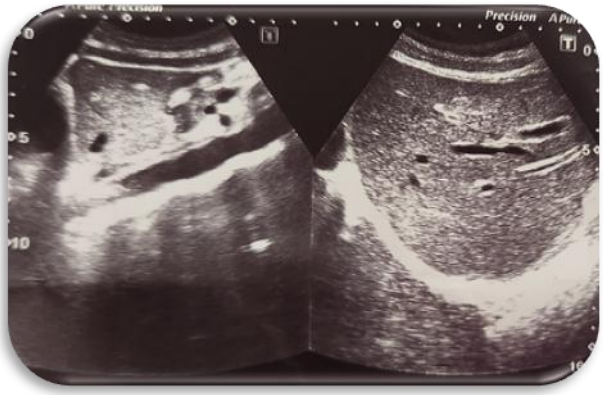


Figure 1a

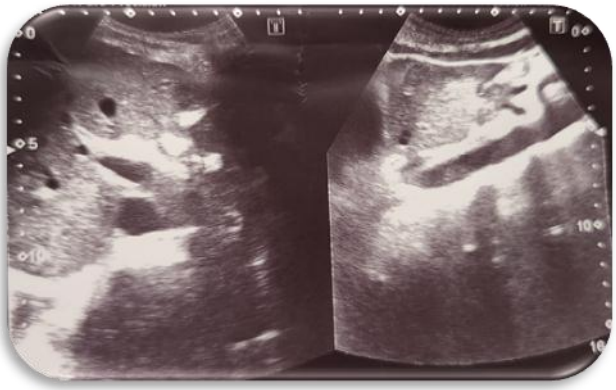


Figure 1b

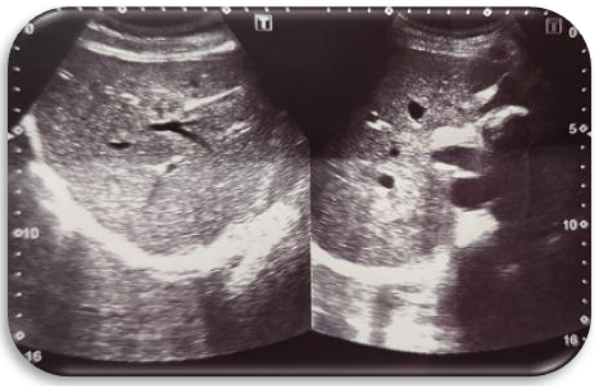


Figure 1c

In the next step an abdominal CT scan was advised to visualize gallbladder in gallbladder fossa or elsewhere in ectopic locations, biliary system or any other pathology in right hypochondrium or elsewhere in abdomen. CT scan of abdomen likewise revealed non-visible gallbladder in its typical location, i.e gallbladder fossa, and was not seen in any of the ectopic sites as well. (figure 2a, 2b, 2c, 2d, 2e, 2f).

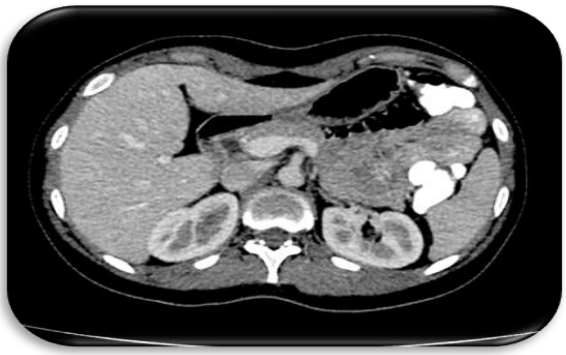


Figure 2a

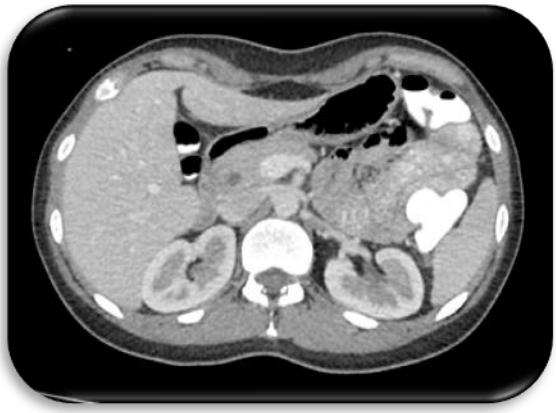
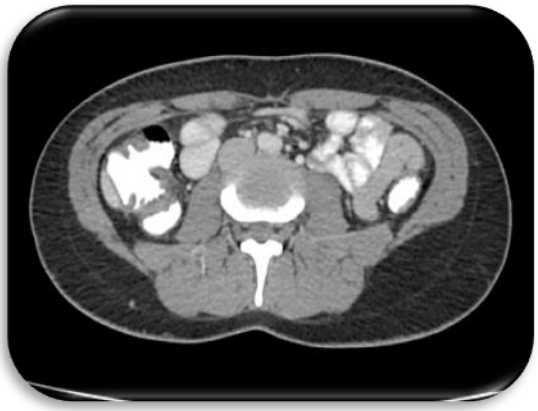


Figure 2b



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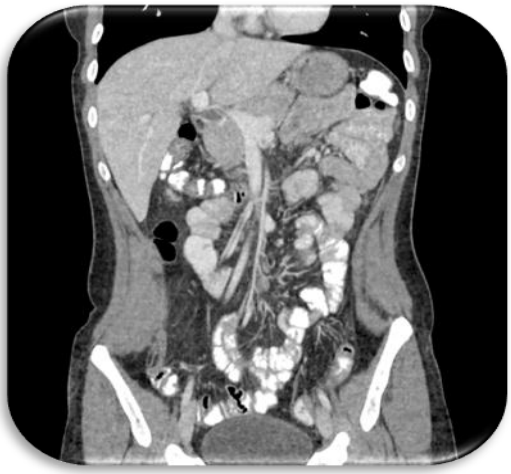

Figure 2d

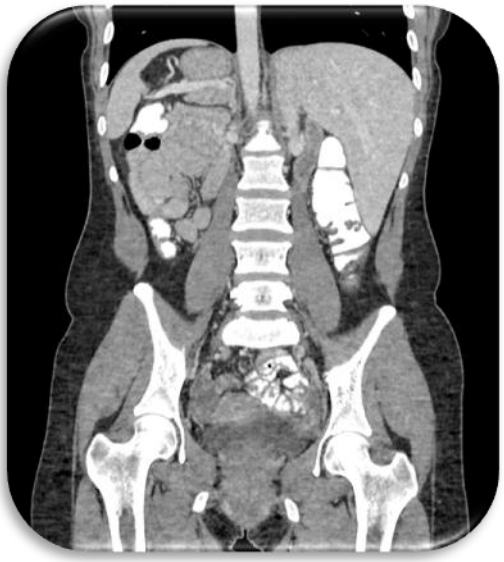

Figure 2e

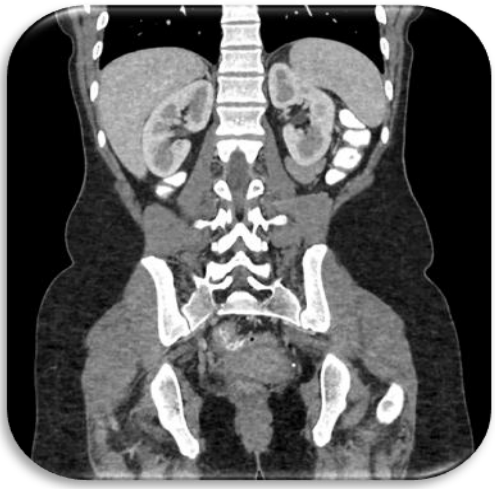

Figure 2f

In light of the non-visualization of the gallbladder, a magnetic resonance Cholangiopancreatography (MRCP) was recommended based on the clinical picture and further imaging like ultrasound, and computed tomography (CT): neither of the tests detected the gallbladder and cystic duct. No morphological changes were detected by MRCP.

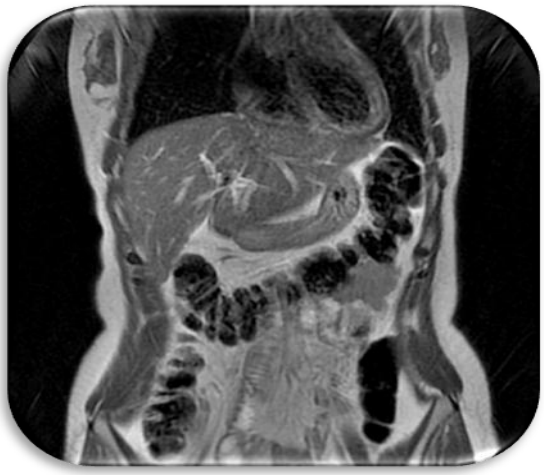

Figure 3a

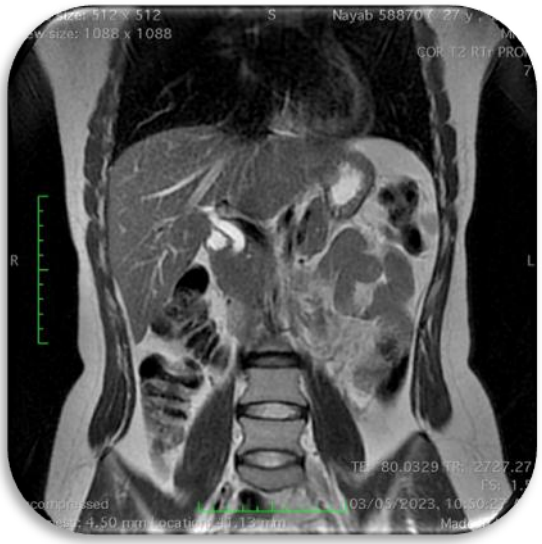

Figure 3b

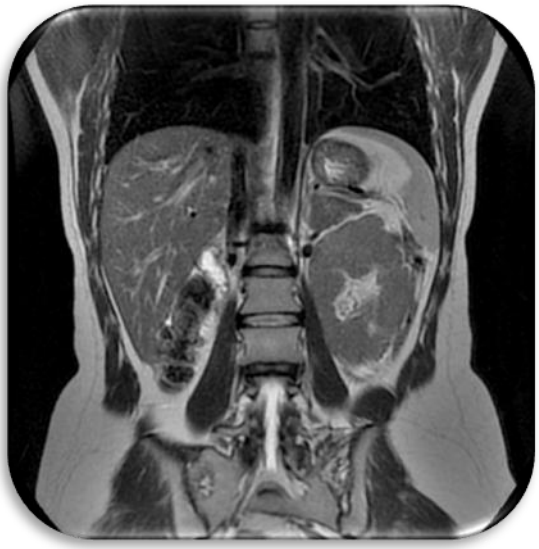

Figure 3c

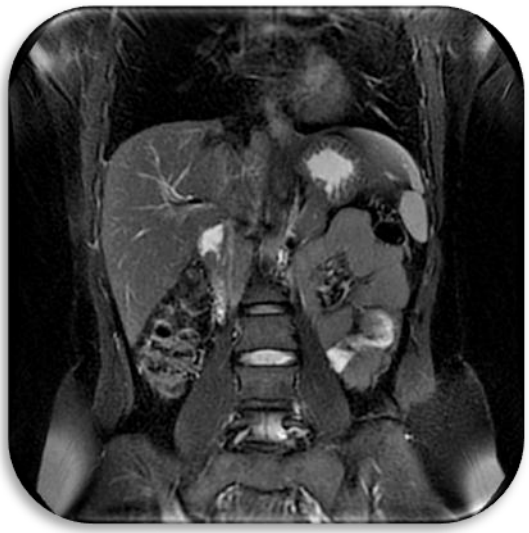

Figure 3d

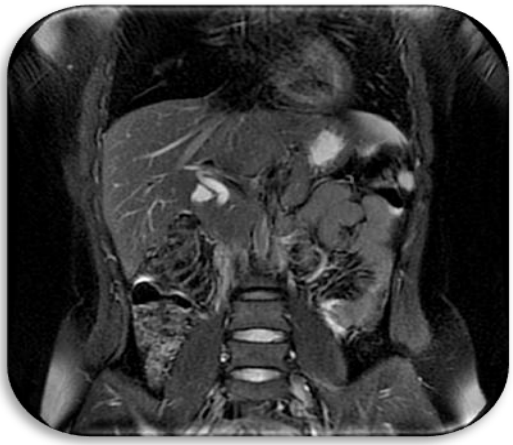

Figure 3e

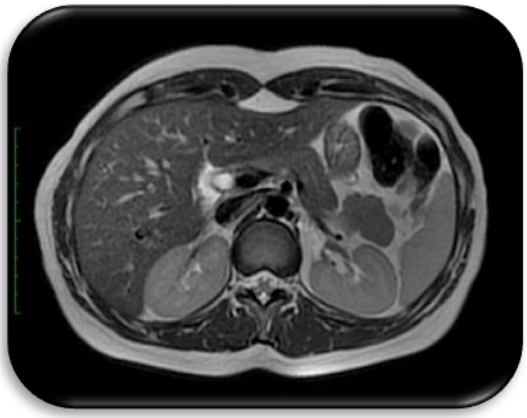

Figure 3f

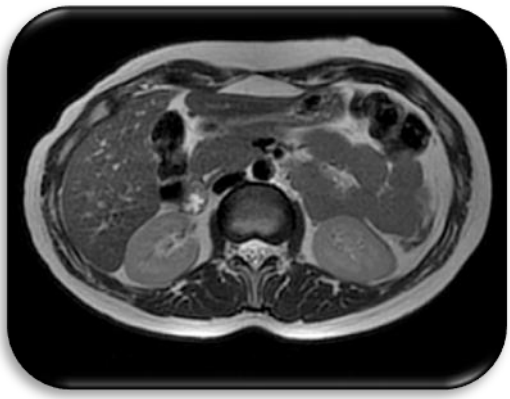

Figure 3g

Figure 3h

The patient responded well to conservative therapy with smooth muscle relxant and was released in good health within a day, with two weeks of uneventful follow-up jaundice was also resolved. She was further advised yearly follow-up with ultrasound.

DISCUSSION

Congenital absence of the gall bladder, a rare developmental disorder that affects less than one in every 6500 live births, is also known as Gallbladder Agenesis (GA). Our research indicates that Lemery and Bergman described the first GA case in 1701-1702.[1,10] Agenesis of the gallbladder occurs between 0.007 and 0.0027% of all instances within clinical settings, and between 0.04-0.13% of the time in postmortem cases. It is more prevalent in females as ratio of 3:1.[3,4,5] GA would be discovered as a standalone entity in 70%–87.2% of instances (just 0.01–0.06%) , of which 31.6% were asymptomatic and 55.6% exhibited symptoms; in the remaining 12.8%–30% of cases, it occurs in conjunction with other congenital changes of the biliary system.[6]

This anatomical abnormality's pathogenesis has two hypothesis. According to the first, the abnormality was caused by the foregut's hepatic diverticulum bud failing to mature properly. Inaccuracy in the canalization of the cystic duct and gallbladder is suggested as a potential contributing factor in the second theory.[5,12] The development of the liver's endocrine system is thought to have been disrupted. The extrahepatic biliary tract is first formed by the hepatic diverticulum's growth and connection with the intestines, followed by the gallbladder. The cystic bud, which forms as a ventral protrusion from the caudal area of the foregut in the fourth week of life in utero, is where the gallbladder gets its start. During the seventh week of development, the hyperplastic epithelium vacuolates. A lumen forms in the cystic duct and gallbladder. It was proposed that agenesis of the gallbladder results from an abnormality at any point in this process. Gallbladder agenesis without extrahepatic biliary tract dysplasia would result from a disturbance before the gallbladder has developed.[8,9] Gallbladder agenesis may be linked to further biliary congenital malformations and/or extra-biliary conditions, such as those of the gastrointestinal, genitourinary, musculoskeletal, or cardiovascular systems.[5]

The gallbladder's normal placement is in the gallbladder fossa, which lies beneath the right lobe of the liver and between segments IV and V.[12,13] Ectopic gallbladder is seen mostly under the left lobe of liver, followed by intrahepatic, transverse, and retroplaced (retrohepatic or retroperitoneal). The falciform ligament, suprahepatic, and the abdominal wall are other rare ectopic locations for the gallbladder.[13]

Often, the biliary system is unaffected by gallbladder agenesis. There are many categorization techniques; the most used is the Bennion et al classification, which categorizes cases into three groups: 1; Multiple foetal abnormalities, 2; asymptomatic, and 3; symptomatic conditions.[11]

The symptomatic group comprises a variety of complaints that would be thought from a gallbladder that is present, such as jaundice and its accompanying characteristics of right upper quadrant pain (90%), nausea and/or vomiting (66%), fatty food intolerance (37.5%), dyspepsia, bloating, or jaundice rarely.[9,11] These symptoms, which are comparable to those of other common biliary tract diseases, are frequently explained by biliary dyskinesia (retrograde sphincter of Oddi contraction), extrabiliary symptoms, or primary choledocholithiasis.[9,13]

Ultrasonography is a very sensitive (95%) diagnostic technique for detecting gallbladder disorders, but it also relies extensively on the operator and the environment of the test. On ultrasonography, a "hyperechoic material in gallbladder fossa, constricted, shrunken, scarred, sclerotic, or atrophied gallbladder " was reported in most of the cases of gallbladder agenesis. In our case gallbladder was not visualized despite the fact that the patient had been fasting for prolong time. The possibility of contracted, atrophic or congenitally absent gallbladder was raised. Additionally, a CT scan is not particularly helpful since gallbladder agenesis or cystic duct blockage might prevent the gallbladder from being seen. To fully identify intrahepatic or ectopic gallbladder elsewhere, though, is helpful.[4,5]

MRCP is the investigation of choice for identifying gallbladder agenesis. It is non-invasive and does not need contrast injection hence does block biliary flow. This approach can be used to examine the biliary tree and an ectopic gallbladder can also be detected. ERCP and EUS are other options to localize the gallbladder at its anatomical position, however they are invasive and cannot detect ectopic gallbladder.[1,7,9,11]

There are no clear suggestions in the literature regarding methods to manage gallbladder agenesis, even though the use of hyoscyamine extended-release pills twice day may assist to reduce symptoms. When considering functional abdominal pain syndrome connected to visceral hypersensitivity as the origin of inexplicable biliary colic, the administration of hyoscyamine in conjunction with low-dose antidepressants such as amitriptyline can be effective in preventing any return of episodes. If no symptoms emerge, no therapy or decisive surgery is necessary since the individual with gallbladder agenesis is healthy and the prognosis is great. Based on our case, we recommended cautious medical therapy and a yearly US follow-up.[4,6,11]

CONCLUSION

Gallbladder is a rare congenital anomaly that should be kept in mind by radiologists to avoid any misdiagnosis when an ultrasonography suggests that the GB is not visible. In the case of clinical suspicion, MRCP is

regarded the gold standard test because it reveals correct anatomic features of the biliary system, avoiding needless investigations or surgery. In asymptomatic cases, no therapy or extra investigations are advised. In symptomatic cases, such as ours, the treatment is mostly conservative, consisting of smooth muscle relaxants. It has good progmosis and the patient leads a healthy life.

Author's contribution: All authors contributed equally in the work.

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Conflict Of Interest: None. Consent for Publication: the patient's permission has been obtained.

REFERENCES

1. Singh S, Koley A, Yadav GD. Gall Bladder Agenesis: Very Rare but to Be Kept in Mind. Available at SSRN 4437954, 2023 May 4.
2. Neuberg M, Detry O. Gallbladder agenesis. Acta Chirurgica Belgica, 2023 Mar 4; 123(2): 218-9.
3. Rivera A, Williamson D, Almonte G, Rivera AM, Hidalgo GA. Preoperative Diagnosis of Gallbladder Agenesis: A Case Report. Cureus, 2022 Oct 27; 14(10).
4. Agarwal PK. Agenesis of gall bladder: diagnosed before it is an unpleasant laparoscopic surprise-clinical case report and review. International Journal of Surgery Case Reports, 2020 Jan 1; 76: 144-7.
5. Ismail IB, Rebii S, Zenaidi H, Zoghlami A. Gallbladder agenesis in the elderly: a diagnostic challenge. The Pan African Medical Journal, 2020; 37.
6. D'Orazio B, Famà F, Martorana G, Di Vita G, Geraci G. Gallbladder agenesis: report of a preoperative diagnosis with magnetic resonance cholangiopancreatography. Cureus, 2020 Aug 10; 12(8).
7. Kasi PM, Ramirez R, Rogal SS, Littleton K, Fasanella KE. Gallbladder agenesis. Case reports in gastroenterology, 2011; 5(3): 654-62.
8. Cegarra-Besora A, González-Álvarez P, Montraveta-Querol M, Bovo MV. Case report of a gallbladder agenesis, a diagnostic challenge. International Journal of Surgery Case Reports, 2022 May 1; 94: 107026.
9. Bianco G, Frongillo F, Agnes S, Nure E, Silvestrini N. Gallbladder agenesis: a case report and brief review. Annals of Hepato-biliary-pancreatic Surgery, 2018 Aug 1; 22(3): 292-5.
10. Meshkati Yazd SM, Bayati H, Nabavizadeh SS, Shahriarirad R. Gallbladder Agenesis without Additional Biliary Tracts Abnormality. Case Reports in Gastrointestinal Medicine, 2022 Jun 23; 2022.
11. Scobie JL, Bramhall SR. Congenital agenesis of the gallbladder: a UK case report. Oxford Medical Case Reports, 2016 Aug 1; 2016(8).
12. Piltcher-da-Silva R, Sasaki VL, Felisberto DE, Bodanese BC, Piltcher-Recuero M, Bodanese BV, Bettini LF, Aguilera YS, da Costa MR, Coelho JC. Gallbladder agenesis a rare and underdiagnosed congenital anomaly: a case report and literature review. Journal of Surgical Case Reports, 2022 Nov; 2022(11): rjac505.
13. Ismail IB, Zenaidi H, Sghaier M, Rebii S, Zoghlami A. Cholecystitis in a midline gallbladder: A rare ectopic location. International Journal of Surgery Case Reports, 2022 Apr 1; 93: 106969.