

1 **A CHOLEDOCHAL CYST RESULTING IN OBSTRUCTIVE JAUNDICE IN A CASE**
2 **WITH GALLBLADDER AGENESIS: Report of a Case and Review of the Literature**

3 **Abstract**

4 Co-existence of gallbladder agenesis and choledochal cyst is a highly rare congenital anomaly
5 of extrahepatic biliary system. Literature presents only five cases with this combination. Here,
6 we report the sixth case. Gallbladder agenesis is usually asymptomatic, while choledochal
7 cyst often presents symptoms before adolescence. This is the report of a 42-year-old female
8 patient with gallbladder agenesis and choledochal cyst leading to obstructive jaundice.
9 Radiological diagnosis of choledochal cysts is not difficult. However, in cases with
10 gallbladder agenesis, cystic dilatation in the choledochus may be misdiagnosed as mislocation
11 of the gallbladder or contracted and/or sclero-atrophic gallbladder.

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13 **Özet**

14 Safra kesesi agenezisi ve koledok kisti birlikteliği oldukça nadir görülen ekstrahepatik biliyer
15 sistemin konjenital anomalileridir. Literatürde şimdiye kadar bu kombinasyonun olduğu
16 yalnızca beş vaka bildirilmiş. Burada altıncı vaka rapor edilmiştir. Safra kesesi agenezisi
17 genellikle semptom vermemesine karşın koledok kisti çoğunlukla adölesan çağa kadar
18 semptom verir. Bu yazıda, tıkanma sarılığına neden olmuş koledok kisti ve safra kesesi
19 agenezisi olan 42 yaşında bir kadın hasta sunulmuştur. Koledok kistlerinin tanısı radyolojik
20 olarak zor değildir. Ancak safra kesesi agenezisi olan vakalarda koledoktaki kistik dilatasyon
21 safra kesesi yerleşim anomalisi, kontrakte ve/veya sklero-atrofik safra kesesi şeklinde
22 yorumlanabileceğinden yanlış tanıya yol açabilir.

23 **Key Words :** Gallbladder agenesis, Choledochal cyst, Obstructive jaundice

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29 **INTRODUCTION**

30 Agnesis of the gallbladder (AGB) and choledochal cyst (CC) are among the rare conditions
31 of congenital extrahepatic biliary tree anomalies. AGB results from the failure of the cystic
32 bud to develop in the 4th week of the intrauterine life. It normally develops from the caudal
33 part of the hepatic diverticulum.^[1,2] CCs are single or multiple dilatations of the intrahepatic
34 or extrahepatic biliary tree. Here, we report choledochal cyst complications presenting as
35 obstructive jaundice in a patient with gallbladder agnesis. This combination is very rare and
36 a review of literature revealed only five such cases.^[3,4]

37 **CASE REPORT**

38 A 42-year-old woman admitted to the emergency department with the complaints of
39 unrelenting pain in the right upper abdomen, post-prandial nausea and vomiting and loss of
40 weight for the last 2 months. She had a medical history of severe and colicky right upper
41 abdominal pain accompanied by intolerance to fatty foods for the last 2 years. Moreover, she
42 had jaundice for the last 10 days. On the abdominal examination, no masses or organomegaly
43 was noted, but there was mild tenderness in the epigastric and right upper quadrant areas
44 without rebound or rigidity. In the clinical examination, the patient was jaundiced and
45 cachectic.

46 He results of the laboratory tests were as follows: white blood cells: 10.2×10^3 /mcL,
47 hemoglobin: 12.3 g/dL, blood urea nitrogen: 16 mg/dL (normal range:1–50 mg/dL),
48 creatinine: 0.6 mg/dL (normal range: 0.5-1.2 mg/dL), aspartate aminotransferase (AST): 337
49 U/L (normal range. 1–38 U/L), alanine aminotransferase (ALT): 339 U/L (normal range: 1–
50 41 U/L), alkaline phosphatase (ALP): 385 U/L (normal range: 35–129 U/L), gama-
51 glutamyltranspeptidaz (GGT): 830 U/L (normal range: 5-61 U/L), total bilirubin 7.4 mg/dL
52 (normal range: 0–1.2 mg/dL), and direct bilirubin: 5.2 mg/dL (normal range: 0-0.2 mg/dL).

53 Ultrasonography (US) showed minimal dilatation of intrahepatic bile ducts. The gallbladder
54 was located in the hilum of the liver, not in its normal anatomic location, and multiple
55 millimetric stones were detected. The abdominal computerized tomography (CT) revealed an
56 ectopic gallbladder containing tiny stones (**Figure 1**). In the endoscopic retrograde
57 cholangiopancreatography (ERCP), intrahepatic bile duct dilatation, ectopic gallbladder with
58 location anomaly of the ductus cysticus were visible. In the same session bile duct stone
59 extirpation was performed using ERCP.

60 The patient underwent laparotomy, during which the gallbladder was not found despite a
61 meticulous search of the abdomen, either by direct vision or by the use of the intraoperative
62 ultrasound probe. On the other hand, a Type 1 CC was detected based on the Todani
63 classification of choledochal cysts (**Figure 2**) (5). The diagnosis was confirmed through
64 intraoperative cholangiography (**Figure 3**). Cyst excision with Roux-en-Y
65 hepaticojejunostomy was performed. Histologically, the wall of the common bile duct was
66 composed of chronically inflamed fibrous tissue with no evidence of malignancy.

67 The patient had cholangitis on the postoperative day 60 and was treated medically. In the 8-
68 month follow-up period, the patient was complaint free.

69 **DISCUSSION**

70 Congenital anomalies of extrahepatic biliary systems are rare. These anomalies may be
71 isolated or combined.^[3] Bergman^[6] was the first author to describe this anomaly in human
72 beings in 1701. AGB is a rare congenital biliary anomaly with an incidence of 0.01-0.06%. In
73 a recent review of autopsies, the incidence of AGB has been reported to be about 1/6334 live
74 births.^[7]

75 Despite having no characteristic symptomatology, possible mechanisms of AGB symptoms
76 have been described as primary duct stones, biliary dyskinesia, or non-biliary disorder. While
77 many adults with AGB remain asymptomatic for life, about 23% of affected individuals

78 present with biliary symptoms, including 90% with right upper quadrant pain, 60% with
79 nausea and vomiting, 37% with food intolerance, 35% with jaundice, and 30% with
80 dyspepsia.^[3]

81 The diagnosis of AGB is difficult with routine investigations and may be misleading,
82 particularly in cases of contracted or ectopic gallbladder. Considering these reasons, in 1967
83 Frey et al.^[8] suggested laparotomy as the only method of diagnosis for AGB. Similarly, in
84 our patient, absence of gallbladder could not be diagnosed preoperatively, but it was
85 determined during the laparotomy. This was evaluated as an abnormal location of the
86 gallbladder because of the presence of a CC. The AGB is usually diagnosed incidentally
87 during investigation or surgery for another disease. Tabibian et al reported a coexistence of
88 AGB and CC in a patient underwent surgery for duodenal obstruction. We determined
89 similarly a AGB coexisting with CC in a patient underwent surgery for obstructive jaundice.
90 If the diagnosis of AGB is made during operation, the surgeon must prove AGB by
91 thoroughly examining the most common sites for ectopic gallbladder, which are intrahepatic,
92 retrohepatic, on the left side, or within the leaves of the lesser omentum or within the
93 falciform ligament, retroduodenal, retropancreatic and retroperitoneal.^[9]

94 Choledochal cyst (CC) is another congenital anomaly of the biliary tree. It was first defined
95 by Vater and Ezler in 1723.^[3] Whereas CC is a rare medical condition with an incidence of 1
96 in 100,000–150,000 live births in the western population, it is remarkably higher in Asian
97 populations with a reported incidence of 1 in 1000, and about two thirds of cases in Japan.^[10]

98 CC is more common in women, with a male to female ratio of 1:3 to 4.^[11] Alonso-Lej et al.
99 established the first classification system for CC in 1959, followed by Todani et al. modifying
100 this system in 1977. Now, the classification system of Todani et al is the most commonly used
101 by clinicians (**Table 1**).^[5]

102 Clinical symptoms can develop at any time; however, 80% of patients present symptoms
103 before the age of 10 years. Only a minority of patients present the classic clinical triad of
104 abdominal pain, jaundice, and abdominal mass. The clinical presentation mostly depends on
105 age, with abdominal pain as the most frequent presenting symptom in adults and jaundice in
106 infants.^[11] Our patient, who was asymptomatic until the age of 42, had choledochal cyst that
107 caused episodic abdominal pain, jaundice, nausea, and vomiting for the last 2 months.
108 Presentation, diagnosis, and prognosis of CC largely depend on its size and location, and
109 type. Our patient had coexisting CC and AGB; thus, preoperative imaging methods were of
110 limited use.
111 Complete cyst excision and Roux-en-Y hepaticojejunostomy is the standard procedure for
112 Type I CC. Our patient underwent total cyst excision and hepaticojejunostomy with
113 laparotomy.
114 Complications of CC, which are the result of stasis, include cholangitis, stone formation,
115 recurrent pancreatitis, cirrhosis, and portal hypertension. Cholangiocarcinoma is another
116 serious complication, with even higher risk after drainage procedures.^[11] Particularly in case
117 of Type I CC, to avoid high risk of cholangiocarcinoma, complete cyst excision and Roux-
118 en-Y hepaticojejunostomy has been advocated, as was performed in our patient. Operation
119 should be performed as soon as the diagnosis of CC is established because of its serious
120 complications even when it is not symptomatic.
121 Radiological diagnosis of CC is not difficult. However, in cases with AGB, the cystic
122 dilatation in the choledochus, as was in our case, may be misinterpreted as location anomaly
123 of the gallbladder (ectopic or in the hiler of the liver), or contracted and/or sclero-atrophic
124 gallbladder, and it may lead to diagnostic challenges or misdiagnosis. Therefore, when the
125 gallbladder is not detected in its normal location or size, AGB and CC coexistence should be
126 kept in mind. In determination of the etiology for jaundice and pancreatitis, particularly in

127 young patients, the gallbladder anatomy should be well evaluated and in differential
128 diagnosis, potential presence of a congenital CC should be kept in mind.
129 Goel et al.^[4] reported the fourth case of CC and AGB coexistence in 1994. Only one such
130 case has been reported since 1994 to date.^[3] To our knowledge, the case reported here is the
131 sixth case of CC and AGB coexistence.

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205 **Table 1. Classification of Choledochal Cysts**

Type I	Dilatation of hepatic and common bile ducts (comprising 80-90% of cases)
Type II	Diverticulum of the common bile duct
Type III	Intraduodenal common bile duct dilatation (termed choledochocele)
Type IV -a	Intrahepatic and extrahepatic bile duct dilatation
Type IV- b	Multiple extrahepatic cysts
Type V	Intrahepatic bile duct dilatation (as in Caroli's Disease)

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227 **Figure Legends**

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229 **Figure 1:** Milimetric densities compatible with stone formations in the liver hilus.

230 **Figure 2:** Gallbladder agenesis and choledochal cyst

231 **Figure 3:** Gallbladder agenesis and choledochal cyst on the cholangiography

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