Case report of a gallbladder agenesis, a diagnostic challenge

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ABSTRACT

Introduction and importance: Agenesis of the gallbladder is an underdiagnosed entity. It can be an asymptomatic pathology, usually diagnosed incidentally as a finding on imaging techniques or in necropsies. There are symptomatic cases presenting as biliary colic too, attributed to cholecystitis or cholangitis, leading to exploratory surgeries.

Case presentation: We present a 14-year-old boy admitted to the outpatient Pediatric Gastroenterology Department with a history of daily abdominal pain for the past 2 months. Blood and fecal tests were normal. Coeliac disease, inflammatory intestinal diseases and Helicobacter pylori infection were ruled out. An abdominal US was informed of a hypodistensed gallbladder with no other findings. A CT scan was performed, which reported of an absent gallbladder. Upon these findings, a Magnetic Resonance Cholangiopancreatography (MRCP) was carried out, confirming the diagnosis of agenesis of the gallbladder.

Clinical discussion: Its diagnosis still poses a challenge, as an absent gallbladder may not always be apparent by US, with the MRCP being the gold-standard test for examining the biliary tract. The prognosis is usually good, and the treatment of choice consists of antispasmodics when needed. There are few cases reported in literature, for this reason, our article discusses the most important aspects of agenesis of the gallbladder in order to suspect it, request the appropriate tests and saving unnecessary surgical interventions.

Conclusion: Gallbladder agenesis is a rare entity that can lead to unnecessary diagnostic tests and interventions. The MRCP is the gold-standard test. Despite its benign nature, symptomatic treatment might be required in order to improve the patient’s quality of life.

1. Introduction and importance

Agenesis of the gallbladder is an underdiagnosed entity unfamiliar to physicians. Most cases are asymptomatic, diagnosed as an incidental finding on imaging techniques or in necropsies. Sometimes it can present as a biliary colic, leading to unnecessary surgeries to treat suspected cholecystitis or cholangitis. With only a few cases reported in the literature, the present article intends to review the most important aspects of agenesis of the gallbladder in order to guide clinicians in its diagnosis and management.

2. Case presentation

We present the case of a 14-year-old boy admitted to the outpatient Pediatric Gastroenterology Department with a history of daily abdominal pain for the past 2 months.

Previous medical history includes an abdominal ultrasonography (US) 3 months earlier informing of a hypodistensed gallbladder with no other findings. There was no other relevant personal or familiar history to be mentioned.

He complained of colic epigastric pain irradiated to the right hypochondriac region, which improved after eating, without pyrosis. He explained a brief period of vomiting at the onset of symptoms, now resolved. He presented a constipated habit, without blood, mucus, acholia or choluria. Asthenia, anorexia and a weight loss of 4 kg were noted, without fever or other symptoms. Physical examination revealed no findings.

Blood tests were taken, including a complete blood count and a chemistry panel assessing renal, hepatic, pancreatic and thyroid hormones, all normal. Coeliac disease was ruled out by negative abdominal pain for the past 2 months.

Abbreviations: MRCP, (Magnetic Resonance Cholangiopancreatography); PPIs, (Proton-pump inhibitors); US, (Ultrasonography).

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determination of anti-transglutaminase antibodies and normal duodenal biopsy. Inflammatory intestinal diseases were also ruled out by normal values of acute phase reactants in plasma and fecal calprotectin. *Helicobacter pylori* antigens in feces were negative, with an unremarkable upper endoscopy. An abdominal US was performed, with no changes from the previous one. The study was completed by a CT scan that informed of an absent gallbladder without other abnormalities. Upon these findings, a Magnetic Resonance Cholangiopancreatography (MRCP) was carried out, confirming the diagnosis of agenesis of the gallbladder (Fig. 1).

Different lines of treatment were introduced, including conventional analgesia, prokinetics, PPIs (proton-pump inhibitors), low-fat diet and spasmolytics, with no improvement. Given the persistence of symptoms with no organic alterations found despite the gallbladder agenesis, a psychogenic component was suspected. Thus, the patient was referred to Psychiatry for further evaluation, which the family refused, discontinuing treatment at our center after three years of follow-up.

### 3. Clinical discussion

The agenesis of the gallbladder is an extremely rare disease, with an estimated prevalence of 0.01–0.06% [1–4], and very few pediatric cases described in the literature. It is believed to be the result of an interruption of the liver embryonic development. The hepatic diverticulum grows and connects with the intestines, thus forming first the extrahepatic biliary tract and last the gallbladder. A disruption before the gallbladder has been formed would lead to a gallbladder agenesis without an added extrahepatic biliary tract dysplasia [1,3–5].

It can present on its own or in association with other malformations and syndromes. When solitary, it is usually asymptomatic, diagnosed incidentally as a finding on imaging techniques or in necropsies [2,5–7]. Mild symptomatic cases presenting as biliary colic have also been described, leading to unnecessary surgeries [2–7]. In such cases, pain is believed to arise from an added sphincter of Oddi dysfunction, either worsening biliary ectasia and favoring gallstone formation in the common bile duct or causing a biliary dyskinesia with painful spasms of the Oddi sphincter [1,3,4,6].

Up to date its diagnosis still poses a challenge, as an absent gallbladder may not always be apparent by US, with the MRCP being the gold-standard for examining the biliary tract [1–7]. Importantly, fasting should be ensured prior to radiologic examination in order to avoid false images in empty gall bladders. The prognosis is usually good, and antispasmodics are the treatment of choice [2–4,6]. Thus said, no clinical guidelines or consensus have been found in the literature on its management. In our patient, the persistence of disproportionate and drug-resistant pain made us suspect the possibility of an added functional disorder.

### 4. Conclusion

Gallbladder agenesis is a rare entity in Pediatrics which should be taken into account in the differential diagnosis of upper digestive tract symptoms. MRCP imaging showing an absent gallbladder is the gold standard in its diagnosis after other aetiologies have been ruled out. Awareness of this pathology may spare unnecessary surgeries and/or invasive procedures. Despite its benign nature, symptomatic treatment might be required in order to improve the patient’s quality of life.

### Consent to participate

Verbal informed consent was obtained from patient's tutors for publication of this case report and accompanying images. Informed consent has been recorded in the patient’s medical history.

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**Fig. 1.** A, Magnetic Resonance Cholangiopancreatography (MRCP) showing a normal biliary tract. B, our patient’s MRCP, with an absence of gallbladder, confirming the diagnosis of agenesis.
CRediT authorship contribution statement

Anna Cegarra: diagnosis and clinical management of the case, literature review, write the manuscript in Spanish
Pablo Gonzalez: diagnosis and clinical management of the case, literature review, translation of the manuscript into English
Montserrat Montraveta: diagnosis and clinical management of the case, literature review, manuscript correction and validation
Victoria Bovo: diagnosis and clinical management of the case, literature review, manuscript correction and validation

Declaration of competing interest

The authors have no conflict of interest to declare.

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