CLINICAL VIGNETTE

Gallbladder Agenesis

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Introduction

A 28-year-old man with no significant past medical or surgical history presented to his primary care provider with complaints of right upper quadrant pain after fatty meals. He described 1 to 2 years of intermittent sharp right upper quadrant pain, occurring 2 to 3 hours after eating, and lasting several hours. His symptoms would occasionally wake him from sleep and occurred every other month. He denied heartburn, nausea, vomiting, diarrhea, or other associated symptoms. His symptoms resolved with avoidance of fatty meals. He denied prior cholecystectomy.

His physical exam was unremarkable. Blood tests including complete blood count, comprehensive metabolic profile, and lipase were normal. An ultrasound did not visualize a gallbladder but was otherwise unremarkable showing normal liver, bile ducts, and visualized pancreas. Because of his symptoms, there was concern for a contracted gallbladder, and a HIDA scan showed non-visualization of the gallbladder despite morphine augmentation. MRCP confirmed congenital absence of the gallbladder and cystic duct in the absence of prior surgery.

Discussion

Gallbladder agenesis is rare congenital anomaly characterized by absence of the gallbladder with a normal bile duct system.[1] This is most often sporadic and occurs at an estimated prevalence of 0.04% based on a postmortem study.[2] It results from failure of the cystic bud to develop *in utero.* Congenital absence of the gallbladder may be an isolated finding, but it has been associated with other anomalies including skeletal, cardiac, genitourinary, and gastrointestinal anomalies.[3]

Patients with gallbladder agenesis can be classified into one of three categories based on data gathered at UCLA from 1955 to 1987:[4] 1) Multiple congenital malformations (13-30%) including patients with biliary atresia or other deformities, frequently associated with demise shortly after birth; 2) Asymptomatic type (30% of cases) where absence of the gallbladder is discovered incidentally during surgeries or autopsy; and 3) Symptomatic type (55%) including biliary symptoms such as right upper quadrant pain, nausea, vomiting, intolerance to fatty foods, dyspepsia, jaundice, and choledocholithiasis. The pathophysiology of symptoms in gallbladder agenesis is similar to post-cholecystectomy syndrome. Stasis may occur in the bile duct, which may be dilated, and can produce clinical features of cholecystitis, cholangitis, and choledocholithiasis.[1]

Misdiagnosis may lead to unnecessary surgery. Abdominal ultrasound may incorrectly interpret gallbladder agenesis as a contracted, fibrosed gallbladder, which may lead to a scheduled cholecystectomy. Excessive exploration during surgery may increase the risk of iatrogenic injury. Nuclear medicine scanning modalities that show non-visualization of the gallbladder may be interpreted as chronic cholecystitis as this would be more common than the rare entity of gallbladder agenesis. Given the frequency of misdiagnosis that may lead to unnecessary surgery, it has been recommended that patients with ultrasonographic findings of absent or atrophic gallbladder undergo further examinations such as CT and magnetic resonance cholangiography.[5]

This condition may be discovered during laparoscopic exploration. It has been proposed that open surgery is then indicated to evaluate for ectopic locations of the gallbladder.[5] Ectopic gallbladders may be located in the liver, between the leaves of the lesser omentum, in the retroperitopeum and retrohepatic region, within the falciform ligament, or in the retroduodenal and retropancreatic; all areas that are difficult to reach with a laparoscope.

Gallbladder agenesis is a rare congenital abnormality that can present a diagnostic challenge. With recognition of this entity, unnecessary operations may be avoided using non-invasive or minimally invasive radiologic techniques to confirm this diagnosis.

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