ABSTRACT

Background: Complete absence of a hemidiaphragm or diaphragmatic agenesis in adulthood is rare with only one previous report in the literature. Its significance in relation to performing laparoscopic procedures has not been documented previously.

Methods: We report a case of previously undiagnosed diaphragmatic agenesis in adulthood precluding laparoscopic cholecystectomy and comprehensively review the literature for papers relevant to diaphragmatic agenesis in adulthood.

Results: Diaphragmatic agenesis in adulthood may complicate and preclude laparoscopic cholecystectomy. The principles of investigation and management of diaphragmatic agenesis complicating laparoscopic surgery are discussed.

Conclusions: In adults with diaphragmatic agenesis and intrathoracic abdominal viscera precluding laparoscopic cholecystectomy, conservative management is recommended.

Key Words: Diaphragmatic agenesis, Laparoscopic cholecystectomy.

INTRODUCTION

Growth failure of the diaphragm during embryogenesis (diaphragmatic agenesis [DA]) is associated with significantly higher morbidity and mortality rates in the perinatal period compared to the more common posterolateral (Bochdalek) defect in congenital diaphragmatic hernia, even following surgical repair.\(^1\) While the clinical course of diaphragmatic agenesis in neonates is well documented, there is a paucity of information on the significance of this condition in adulthood. We report the case of an adult patient in whom previously asymptomatic unilateral DA precluded laparoscopic cholecystectomy. To the authors’ knowledge, only one other case of asymptomatic unilateral DA presenting in adulthood has been reported.\(^2\)

CASE REPORT

A 41-year-old male presented with a two-year history of progressively worsening fatty food intolerance, flatulence, dyspepsia and intermittent right upper quadrant pain radiating to his scapula. It had become increasingly severe and caused sleep disturbance. He had no other co-morbid medical conditions and an unremarkable past medical history. On admission, physical examination revealed absent breath sounds and dullness to percussion in the right lung base but was otherwise unremarkable. Liver function tests revealed a slightly elevated gamma-glutamyl-transaminase (55 IU/L) but was otherwise normal. Postero-anterior and right lateral plain film chest radiographs were suggestive of a right diaphragmatic hernia. Abdominal ultrasound was confirmatory of right diaphragmatic herniation of the abdominal viscera, making it impossible to visualize the gallbladder. Laparoscopy revealed right hemidiaphragm agenesis with an elevated and rotated intra-thoracic liver (Figure 1). Bowel and omentum were in the right hemithorax (Figure 2). Furthermore, the central tendon of the diaphragm was absent with a pericardial defect (Figure 3). The gallbladder was not visualized at laparoscopy, precluding laparoscopic cholecystectomy. He was followed on an out-patient basis and was asymptomatic until seven months later when he presented with a five-day history of anorexia, jaundice and constant right
upper quadrant pain radiating to his right subscapular region. Abdominal ultrasound revealed dilatation of the intra- and extra-hepatic bile ducts suggestive of choledocholithiasis. Endoscopic retrograde cholangiopancreatography (ERCP) demonstrated common bile duct dilatation with choledocholithiasis. ERCP with endoscopic sphincterotomy, stone extraction and stenting of the common bile duct was performed. Following ERCP, he was commenced on gallstone dissolution therapy with ursodeoxycholic acid (250 mg twice daily orally).

DISCUSSION

Bingham, in 1959, first distinguished DA as a distinct anatomical entity from the more common posterolateral congenital diaphragmatic defect (Bochdalek) in an infant who died shortly after birth. Until a decade ago, DA was considered exceedingly rare with only one case report of survivors. There has been an increased incidence of DA in neonates with congenital diaphragmatic hernia since the early 1980s due to improved perinatal care. The reported incidence is 27-31% of cases of congenital diaphragmatic hernia. Experience on the management of neonatal DA as a distinct clinical entity is thus increasing. Consequently, the incidence of adults with DA is likely to increase, and the management dilemmas of the case presented here may occur more frequently in the future.

Diagnosis of DA in adults is based on clinical findings and imaging studies. Dullness to percussion and absent or diminished breath sounds in a lung field may indicate a congenital diaphragmatic hernia. Imaging studies (chest radiography, and abdominal ultrasound) are confirmatory. In adults with DA presenting with cardiorespiratory symptoms, diagnostic and therapeutic approaches differ from those in neonatal DA.

Figure 1. Liver and omentum in right hemithorax via laparoscope.

Figure 2. Large bowel with attached omentum within right hemithorax via laparoscope.

Figure 3. Pericardial defect revealing myocardium as seen from abdomen at laparoscopy.
piratory or gastrointestinal symptoms, the main principle of diagnosis should be to systematically investigate the gastrointestinal tract to rule out pathology in the herniated intrathoracic viscera.

Surgical repair of DA in adulthood is complex. Difficulties arise in deciding the operative approach, reducing the densely adherent intra-thoracic visceral contents and repairing the extensive diaphragmatic defect. Although the patient had no other comorbid medical condition, we decided on conservative management because aggressive surgical intervention would have involved an open cholecystectomy through a thoracoabdominal incision with difficulty in safely reducing the intra-thoracic liver and reconstructing the diaphragm. Furthermore, gallstone extraction was successfully performed by ERCP, and the patient had survived into adulthood without medical problems related to the DA. Emphasis was therefore placed on managing the choledocholithiasis endoscopically, supplemented with gallstone dissolution therapy. This minimalist approach was successful without exposing the patient to an extensive precarious surgical intervention to repair the diaphragmatic defects. The patient remains well and asymptomatic on follow-up of 36 months.

**CONCLUSION**

In adults with DA precluding laparoscopic cholecystectomy, conservative management is advocated. The focus should be on elimination of symptoms rather than diaphragmatic repair.

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