Presentations of agenesis of the hemidiaphragm in an adult

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ABSTRACT

نصف مريض في العقد السادس من العمر جاء يشكو من ضيق في الصدر واستفراغ لمدة أسبوع عند الكشف كانت هنالك أصوات للأمعاء الدقيقة في أسفل الصدر في الجانب الأيسر.أظهرت صورة أشعة للصدر وصورة ملونة بحامض البيريم وأشعة مقطعية وضعت احتمال التواء في المعدة وفتاك في الحجاب الحاجز أجريت عملية جراحية للمريض أظهرت عدم وجود أي التواء فقط لا يوجد حجاب حاجز في الجانب الأيسر. وشوهدت الرئة صحيحة فوق الحاجز البلوري وتعتبر هذه من الحالات النادرة.

A 67-year-old man presented with chest tightness and vomiting of one-week duration. On physical examination bowel sounds were heard on the left chest. Plain chest x-ray, barium swallow, and CT scan suggested gastric volvulus with diaphragmatic hernia. At laparotomy there was complete agenesis of the left hemidiaphragm with no diaphragmatic remnants seen and no associated lung hypoplasia. This is an extremely rare condition and careful assessment is needed to differentiate between diaphragmatic hernia and agenesis of the diaphragm as more cases could be diagnosed in the future as a result of good perinatal care of agenesis of hemidiaphragm (AHD) and long term survival to adulthood.

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Congenital diaphragmatic hernia (CDH) occurs in approximately one out of every 2,200 to 7,000 births.[1,2] Males are more commonly affected than females.[3] Usually the infants are full-term and the defect occurs on the left side in the majority of cases (88%).[4] It is usually diagnosed within minutes or hours after birth due to respiratory distress[5] and carries a high mortality rate (62%).[2] Delayed presentation in adult life is an extremely rare occurrence with only 4 cases reported in English literature.[6-9] The vast majority of diaphragmatic hernias occurring in adults are either standard hiatal hernias or due to traumatic disruption of the diaphragm presenting acutely or with delayed manifestations. Cases of agenesis of hemidiaphragm (AHD) are very rare even in infants, and cases discovered in adulthood are extremely few. These cases were variable in presentation and there is controversy in the management of such patients. There is one case of true AHD reported previously with no intraoperative diaphragmatic remnants seen that presented with classical features of bowel obstruction, and had lung hypoplasia.[9] With better management of neonatal AHD, more adult cases will be seen in the future, hence, more careful work up and initial conservative approach may be adopted before embarking on surgery. We present this case to highlight the importance of the diagnostic work up when a case of AHD is encountered.

Case Report. A 67-year-old patient presented with vomiting, diarrhea, and chest tightness of one week duration. The vomiting got worse during the last 3 days. He was diagnosed as hypertensive 8 months ago and was on treatment (Stamb and Losartan). He had 2 previous operations; hemorrhoidectomy in 1972 and appendectomy in 2006. He denied any history of abdominal trauma. Examination revealed a healthy man with soft abdomen. Bowel sounds were heard on the left side of the chest. Electrocardiogram was normal. A chest x-ray showed a high left hemidiaphragm, which was reported by the radiologist as essentially normal with a possible partial gastric volvulus (**Figure 1**). A dilute barium study confirmed the presence of stomach in the left chest (**Figure 2**). All blood tests were normal. In view of the increased vomiting and the possibility of a diaphragmatic hernia or gastric volvulus, a laparotomy was carried out 3 days after presentation and one week from the onset of symptoms. There was complete absence of the left hemidiaphragm and the left parietal pleura could be seen with the left lung normally moving with respiration. The gastroesophageal junction was found

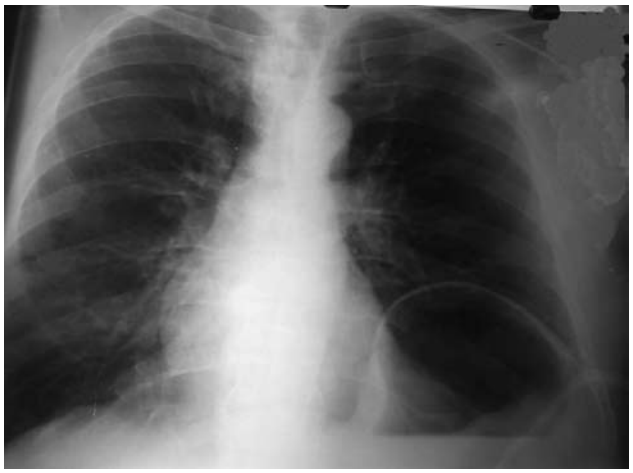

Figure 1 - Plain chest x-ray showing a high left hemidiaphragm.

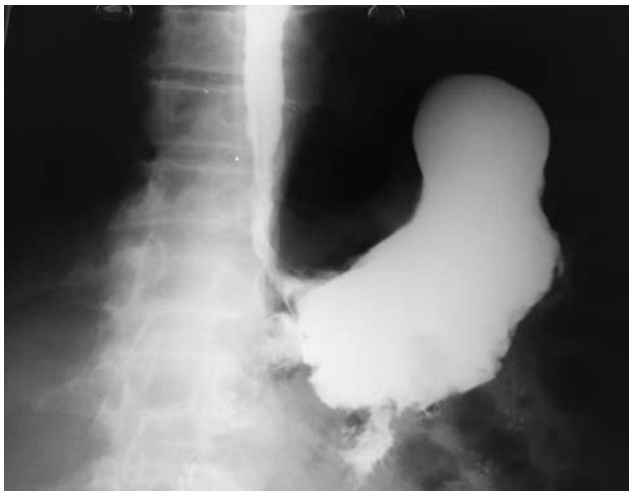

Figure 2 - Barium meal showing the stomach in the left chest.

to be in normal position. There were no adhesions. Primary repair of the defect was obviously not possible due to the complete absence of the left hemidiaphragm and its remnants. The abdomen was closed without any procedure. The patient had an uneventful recovery and was asymptomatic 12 months later.

Discussion. Congenital diaphragmatic hernia may be classified as complete or partial.[9] Partial defects of the diaphragm are more common, including posterolateral herniations at the foramen of Bochdalek, which present with the classical triad of respiratory distress, apparent dextrocardia, and scaphoid abdomen. Defects of the foramen of Morgagni, which occur less frequently are found anteriorly between the sternal and costal attachments of the diaphragm. Agenesis of hemidiaphragm is unusual and is now considered as a distinct entity rather than an extreme form of Bochdalek hernia.[3] It is usually associated with pulmonary hypoplasia, resulting in progressive respiratory failure and death of the neonate.[4] Late presentation of AHD is exceedingly rare. Agnesis of hemidiaphragm results from non-development of the embryologic origins of the diaphragm, including, septum transversum (the fused myotomes of the segments C3, C4 and C5), dorsal mesentery of the esophagus, innermost thoracic wall, and the pleuroperitoneal membranes. It is usually associated with lung hypoplasia and is generally thought to be a result of compression from herniated abdominal contents.[4] In spite of the associated lung hypoplasia, no previous reported case in adults presented with respiratory manifestations. It was postulated that the hypoplastic lung allows more favorable ventilation-perfusion matching.[4] Our case had normal lung appearance and function intraoperatively.

Tzelepis[6] et al reported absence of the left hemidiaphragm in an asymptomatic 22-year-old man. The asymptomatic presentation of AHD suggested that no surgical treatment is necessary as the defect is large enough to permit free movement of visceral structures, thus, lessening the chance of strangulation and incarceration. However, Sheehan[9] et al reported a 66-year-old woman presented with a 3-day history of classical features of large bowel obstruction. At emergency laparotomy, the transverse colon and splenic flexure were located in the left hemithorax. The entire left hemidiaphragm was absent, and there were no diaphragmatic remnants visible. This was the first reported case of true AHD in an adult and it was associated with lung hypoplasia. Travaline[8] et al reported a 39-year-old woman with asymptomatic AHD and lung hypoplasia discovered accidentally following a routine chest x-ray carried out for another reason. The patient was managed conservatively with no surgical intervention.

Singh and Bose[7] reported a case of partial left-sided defect that was repaired by suturing a synthetic mesh to the diaphragmatic remnants. No long-term follow-up data were available to assess success of treatment or possible complications. Abel Waki[10] et al reported a patient with AHD discovered during laparoscopy intended for cholecystectomy, which was abandoned, and the patient then managed conservatively. Our patient, a 67-year-old male who survived for 6 decades without symptoms. The benign nature of this disease is due to the wide space permitting free bowel movement. However, if adhesions develop then symptoms may arise. Our patient presented with acute symptoms that necessitated non-therapeutic laparotomy. No adhesions were encountered intraoperatively. The possibility

of transient gastritis or colitis or gaseous distension cannot be ruled out. Differentiation between AHD and diaphragmatic hernia may be difficult in acute situations, and laparoscopy may be helpful after a careful initial conservative period. The presence of normal left lung may be attributed to good perinatal and early childhood care and avoidance of repeated chest infection.

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