Case Report

Agenesis of Right Diaphragm in the Adults: A Diagnostic Dilemma

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Abstract

Diaphragmatic agenesis is the most extreme form of congenital diaphragmatic defect, and it may be unilateral or bilateral. Diaphragmatic agenesis is a rare diagnosis, typically made early in infancy and is generally associated with other genetic anomalies, especially anueploidy syndromes. It is associated with a high mortality, if not treated in infancy. However, a few patients have survived till adulthood. In this report, we describe the case of an adult female who presented with progressive shortness of breath during third trimester of pregnancy with complete agenesis of the right side of the diaphragm. A new diaphragm was created using polypropylene mesh after which she improved symptomatically and the lung showed good expansion. **[Indian J Chest Dis Allied Sci 2014;56:121-123]**

Key words: Diaphragmatic agenesis, Hernia, Chest, Computed tomography, Gastrointestinal.

Introduction

Development of the diaphragm starts at the fourth week of gestation and completes by ninth week. While experimental studies has been carried out, to understand the orgenogenesis of the diaphragm and its congenital defects, the cause of agenesis or complete absence of diaphragm is not established.[1] Diaphragmatic agenesis is the most severe form of diaphragmatic defects and is associated with a high mortality of 40% to 62%[2,3] and associated cardiac anomalies. We present an unusual case of adult onset progressive shortness of breath and peri-partum hepatic herniation into the chest, that on intra-operative exploration revealed diaphragmatic agenesis.

Case Report

A 25-year-old, primigravida presented in the obstetric outpatient clinic with a history of recent onset of shortness of breath during the second trimester of pregnancy. Physical examination showed decreased breath sounds on the right lower chest. In the next few days, her symptoms progressed and she developed dyspnoea even at rest in supine position. Chest radiograph (postero-anterior view) with abdominal shield (Figure 1) revealed a homogeneous opacity on the right side of the chest. She was diagnosed as a case of pleural effusion by the obstetrician. During the next few days patient's condition deteriorated further and aborted. The chest radiograph was repeated that showed similar findings. A contrast enhanced computed tomography (CECT) of thorax (Figure 2) revealed the entire liver and some loops of small intestine present on the right side of the chest. No fibres of the dome of diaphragm were seen. She had no history of thoracic trauma or any related problem in any of her family members.

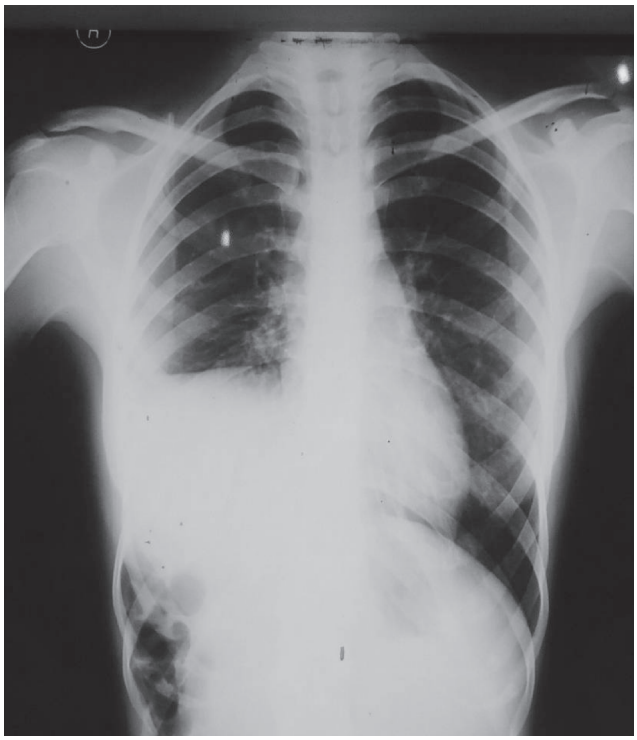

Figure 1. Chest radiograph (postero-anterior view) showing an apparently raised right diaphragm.

A right postero-lateral thoracotomy was performed through the 7th intercostal space. On exploration, the entire liver was on the right side of thorax with loops of small bowel along with an upward shift of the right kidney, without any evidence of diaphragmatic fibres (Figure 3). The right lung was completely collapsed

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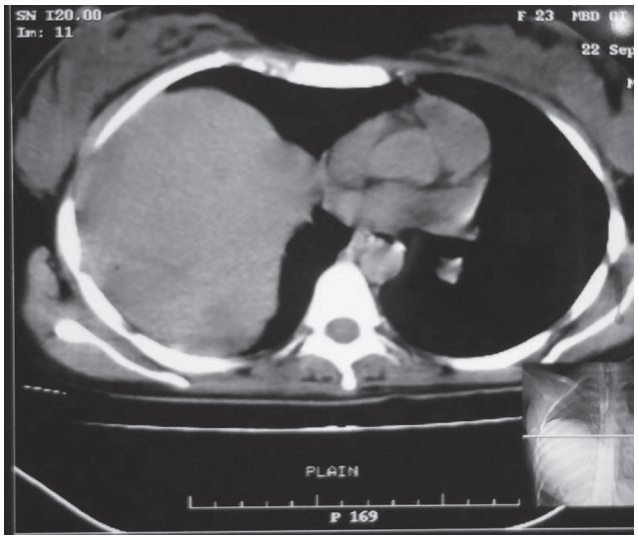

Figure 2. Computed tomography of thorax showing shifting of liver in the right hemithorax.

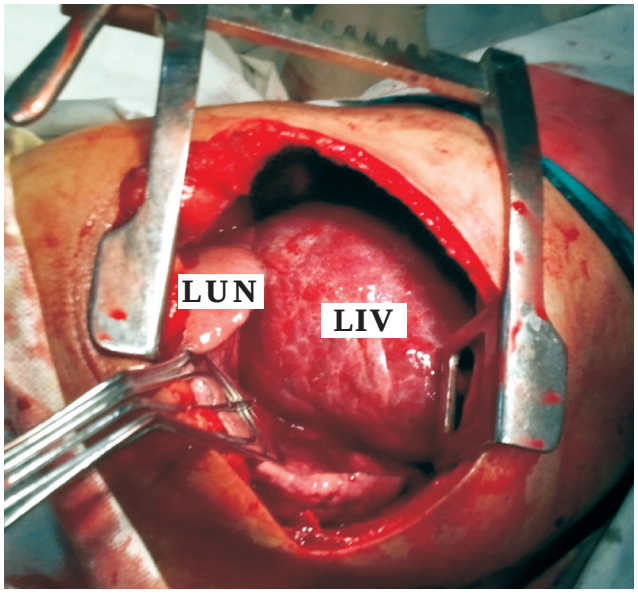

Figure 3. Intra-operative photograph showing no evidence of right diaphragm.

with some hypoplastic changes in lower part of the right lower lobe. After dissection and adhesiolysis, liver and other viscera were mobilised and reposed into the abdominal cavity. A new diaphragm was constructed between thorax and abdomen with the help of polypropylene mesh (15cm × 15cm) and fixed antero-laterally by anchoring sutures in 8th intercostal space with other lower ribs and posteriorly by posterior ribs and medially with mediastinal fascia (Figure 4). After fixation of the mesh, the right lung was inflated with high positive pressure and a chest drain was inserted. She was extubated next day after elective ventilation for 18 hours. Post-operatively, she had an uneventful course and was discharged on the 8th day. She has no complaints and on regular follow-up.

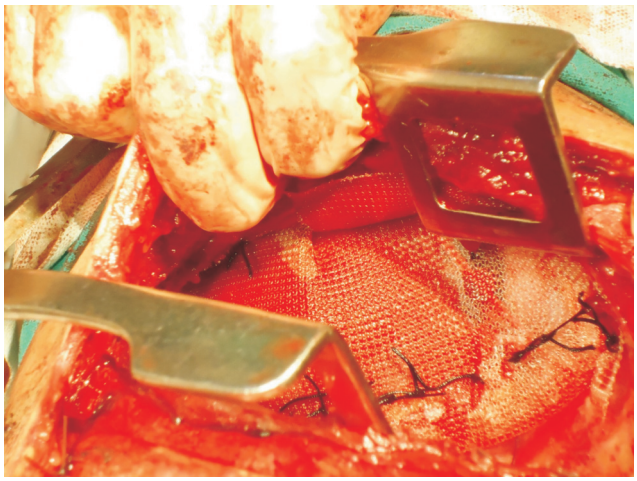

Figure 4. Construction of new diaphragm by polypropylene mesh with anchoring sutures.

Discussion

The development of the diaphragm occurs early in gestation via a fusion of the embryonic pleuraperitoneal membrane and the transverse septum. During the third week of gestation, the fusion of the transverse septum with the dorsal mesentery of the foregut creates two openings whereby the thoracic and abdominal contents meet.[2] During the ninth week of gestation, these openings close. Thus, any defect or arrest of this developmental process may lead to defects in the diaphragm, including congenital diaphragmatic hernia (i.e., hernia of Morgagni, or Bochdalek) and diaphragmatic agenesis. Left diaphragmatic defects are more common than the right side, probably because of earlier closure of the right pleura-peritoneal hiatus.[4] The Bochdalek's hernia accounts for about 90% of all cases of CDHs. It involves an opening on the left side of the diaphragm, and the stomach and intestines usually move up into the chest cavity. The Morgagni's hernia makes up about 2% of all cases, involves an opening on the right side of the diaphragm, and the liver and intestines usually move up into the chest cavity. Diaphragmatic agenesis is considered as one of the rare congenital malformations of the diaphragm and reported in 6% of all CDHs. A diagnosis of CDH can be made in the prenatal period and is more common on the left side. However, isolated diaphragmatic agenesis is an exceedingly rare entity.[6]

Diaphragmatic agenesis is typically diagnosed very early in infancy and carries a significant mortality of up to 38% to 62%, depending upon other congenital anomalies.[2,5] In complete hemidiaphragmatic agenesis, no diaphragmatic remnant is present but in partial agenesis a small rim of diaphragm may be present in the posterior aspect.[6] The first reported case of diaphragmatic agenesis in an adult was described in 1988 on the left side of hemidiaphragm.[7] No cases of CDH in humans have been unequivocally attributed to teratogenic or environmental exposures. Recently, a

potential association between one syndromic case of CDH (Fryns syndrome-like phenotype) and the immunosuppressive drug mycophenylate mofetil (MMF) has been proposed. Animal studies have also revealed that use of MMF during pregnancy is associated with diaphragmatic hernia. However, the mechanism by which MMF may cause diaphragmatic defects is unknown.[8]

The diaphragmatic hernia may or may not manifest any symptoms depending on the compensatory respiratory mechanisms, site and size of the hernia. While most adult patients with left-sided diaphragmatic defects become symptomatic because the of visceral herniation,[9] it has been seen that a right-sided agenesis may occur with few symptoms or may be asymptomatic due to the presence of the liver preventing other viscera from herniating through the diaphragmatic defect. Our patient was also asymptomatic prior to pregnancy. Her symptoms started only in the 3rd trimester of pregnancy when lung function became further compromised by the displaced gravid uterus.

Several imaging modalities are used to assess diaphragmatic hernias including routine chest radiographs after nasogastric tube placement, upper gestrointestinal contrast studies, ultrasonography, computed tomography of thorax and magnetic resonance imaging.[10] Despite all these, an early diagnosis is always challenging. Patients usually present with breathlessness or obstructive symptoms owing to change in thoracic respiratory mechanics and intra-thoracic visceral herniation. On routine chest radiography these have ill-defined opacity which can be confused with consolidation, pulmonary tumours, contusion or thickened pleura. Solid organ herniation may be more difficult to diagnose and may require computed tomography. Further imaging is crucial when symptoms are more understated or patients are asymptomatic.

The management of the right-sided diaphragmatic defects is controversial. In the trauma literature, patients who present with right-sided herniation can often be managed non-operatively because the liver prevents visceral herniation. However, patients with right-sided agenesis may require an operative intervention to prevent liver as well as other abdominal organ herniation.[11] Patients who are symptomatic with dyspnoea or asymptomatic should have an operative intervention to prevent further complications.[12] Our patient needed operative correction of diaphragm to prevent complications in future pregnancies. A thoracotomy, laparotomy or thoracoabdominal approach provides a good exposure and access to the abdomen depending upon expertise of operating surgeon. Several techniques of creating a new diaphragm with the use of different materials has been described in literature ranging from sutures, free grafts, abdominal muscle flaps and prosthetic material.[13] Reconstruction was performed in the present case with a polypropylene mesh with No. 1 silk anchoring sutures all around the chest wall making a compartment between chest and thorax in an interrupted fashion.[14,15]

In conclusion, congenital absence of diaphragm is a very rare diagnosis in adulthood, even more so on the right side. A possible differential diagnosis should always be kept in mind when there is an opacity in the right lower chest. Early diagnosis and management of a diaphragmatic hernia is extremely important to reduce further morbidity or mortality.

References

1. Clugston RD, Greer JJ. Diaphragm development and congenital diaphragmatic hernia. *Semin Pediatr Surg* 2007;16:94-100.
2. Travaline JM, Cordova F. Agenesis of the diaphragm. *Am J Med* 1996;100:585.
3. Available at URL: http://emedicine.medscape.com/article/978118-overview.
4. Komanapalli CB, Velamoor GR, Schipper PH, Sheppard B, Sukumar MS. Peripartum diagnosis and management of diaphragm agenesis 2006. Available at URL: http://www.ctsnet.org.
5. Gale ME. Bochdalek hernia: prevalence and CT characteristics. *Radiology* 1985;156:449-52.
6. Mirza B, Bashir Z, Sheikh A. Congenital right hemi diaphragmatic agenesis. *Lung India* 2012;39:53-5.
7. Gualandri V, Lalatta F, Orsini GB, Zorzoli A, Bertagnoli L, Gallicchio R. Prenatal diagnosis of a case of familial recurrence of unilateral diaphragmatic agenesis. *J Genet Hum* 1983;31:125-31.
8. Parisi MA, Zayed H, Slavotinek AM, Rutledge JC. Congenital diaphragmatic hernia and microtia in a newborn with mycophenolate mofetil (MMF) exposure: phenocopy for Fryns syndrome or broad spectrum of teratogenic effects?. *Am J Med Genet* 149:1237-40.
9. Tzelepis GE, Ettensohn DB, Shapiro B, McCool FD. Unilateral absence of the diaphragm in an asymptomatic adult. *Chest* 1988;94:1301-03.
10. Ghanem AN, Chankun TS, Brooks PL. Total gastric gangrene complicating adult Bochdalek hernia. *Br J Surg* 1987;74:779.
11. Kirkland JA. Congenital posterolateral diaphragmatic hernia in the adult. *Br J Surg* 1959;4:16-22.
12. Wakai A, Winter DC, O'Sullivan GC. Unilateral diaphragmatic agenesis precluding laparoscopic cholecystectomy. *JSLS (J Soc Laproendoscop Surg).* 2000;4:259-61.
13. Chen JC, Wilson SE. Diaphragmatic injuries: recognition and management in sixty-two patients. *Am Surg* 1991;57:810-5.
14. Shaffer JO. Prosthesis for agenesis of the diaphragm. *JAMA* 1964;188:1000-2.
15. Singh G, Bose SM. Agenesis of hemidiaphragm in adults. *Aust NZ J Surg* 1993;63:327-8.