Case Report

Agenesis of the Hemidiaphragm: A Rare Presentation in an Adult

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ABSTRACT

Unilateral diaphragmatic agenesis is a rare finding in adult patients. We report a case of unilateral agenesis of diaphragm in a 22-year-old male patient to highlight the fact that a rare entity of agenesis of diaphragm can have a misleading presentation in adulthood due to both pulmonary and abdominal symptoms.
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Key words: Diaphragm, Agenesis, Thoracotomy.

INTRODUCTION

Diaphragmatic agenesis, unilateral or bilateral, is a rare occurrence in adults. While traumatic diaphragmatic defects or congenital defects such as posterio-lateral Bochdaleck's or anterior Morgagni's hernia are well known, it is debatable whether agenesis of the diaphragm is a separate clinical entity rather than being a variant of diaphragmatic hernia.

Due to its rarity in adult patients, only anecdotal references are available in the literature.

We report a case of unilateral agenesis of diaphragm in a young male in whom the defect was diagnosed during surgical exploration.

CASE REPORT

A 22-year-old male patient was referred to our Institution with a diagnosis of suspected massive left pneumothorax for which an intercostal drainage had been inserted but not draining (Figure 1).

A detailed history of the patient revealed that he was treated for cough and vague abdominal discomfort for the last one month. When the symptoms did not respond to medications, his physician advised a chest radiograph that revealed air in the left chest, hemithorax, prompting insertion of an intercostal drain.

Clinical examination revealed him to be mildly dyspnoeic with normal pulse and blood pressure. On pulse oximetry, oxygen saturation was 92% on room air. Chest examination revealed absent left-sided chest movement, with absence of breath sounds.

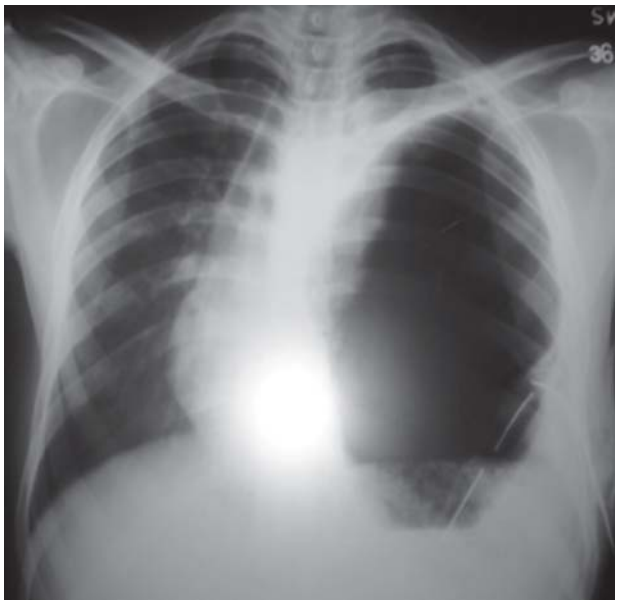

Figure 1. Chest radiograph (postero-anterior view) showing kinked intercostal chest tube *in-situ* with suspected left massive pneumothorax.

As the mediastinum was shifted to the right on chest radiography and tube was kinked, an attempt was made to manipulate the tube but abandoned after omentum was seen adhering to it.

A computed tomography of chest and upper abdomen (Figure 2) showed that the left lung was collapsed with a distended stomach filling the entire left chest cavity.

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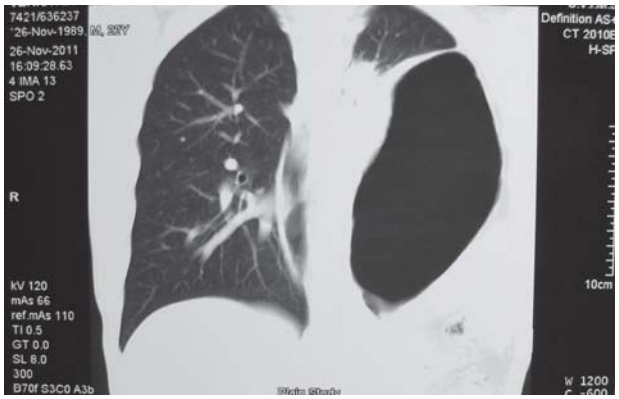

Figure 2. Computed tomography of chest showing distended stomach inside left pleural cavity with collapsed left lung with no apparent visible diaphragm on the left side.

The patient underwent a left posterio-lateral thoracotomy through the fifth intercostal space. Intra-operative findings included complete absence of the left hemidiaphragm with an open communication between the abdomen and the left chest. No vestige of left hemidiaphragm was seen. The stomach, spleen and part of the small and large bowel were found in the pleural cavity (Figure 3).

The viscera was replaced into the abdomen, and a repair was performed by reconstructing the left hemidiaphragm with a polypropylene mesh

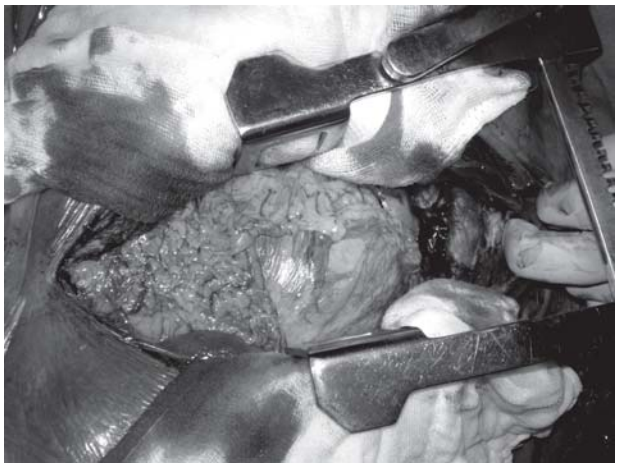

Figure 3. Operative photograph of left thoracotomy showing abdominal contents inside the left pleural cavity.

The mesh was circumferentially attached to the chest by taking interrupted Ethibond® (Johnson and Johnson) sutures from pericardium, left pulmonary ligament, endothoracic fascia and rib cage. The post-operative period was uneventful and the left lung expanded well (Figure 4).

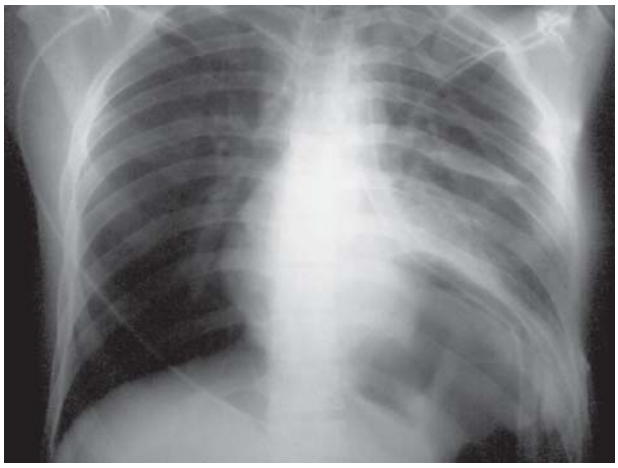

Figure 4. Post-operative chest radiograph (postero-anterior view) showing well expanded left lung.

DISCUSSION

Complete agenesis of a hemidiaphragm is a rare malformation in adulthood whose embryologic basis is unknown.[1,2] An abnormal organogenesis may lead to a diaphragmatic defect, that may be variable in site and size. The diaphragmatic agenesis is the largest diaphragmatic defect, and is associated with herniation of the abdominal contents into the thoracic cavity and pulmonary hypoplasia, that is usually responsible for a fatal cardio-pulmonary failure within the first hours after birth.[3,5] An adult presentation of the diaphragmatic agenesis is extremely rare.

Seven cases of adult patients with diaphragmatic agenesis have been reported in the literature with most of them presenting with severe dyspnoea or bowel obstruction, necessitating emergency thoracotomy or laparotomy.[1-5]

Tzelepis *et al*[6] who reported the first case of agenesis of the diaphragm and Travaline and Cordova[7], managed the patients conservatively because of the belief that bowel incarceration and strangulation were unlikely in the face of such a large defect.

In the present case, the patient was stable clinically with mild symptoms of a nagging cough and vague bowel discomfort. Chest radiograph initially suggested a tension pneumothorax leading to placement of an intercostal drain. The diagnosis of agenesis of the hemidiaphragm was suspected on CT of chest and upper abdomen.

The surgical treatment of diaphragmatic defects can be done by different techniques, but it must be performed with no stretching and minimal anatomical and functional alterations.[8] Among the different factors that determine the technical choice, the size of the defect is the most important.[2,9] In case of patients with a large defect or agenesis of

the hemidiaphragm, a direct suture may be impossible because of the absence of diaphragmatic tissue. We were able to successfully replace the diaphragm with a polypropylene mesh, although a few studies stress reconstruction by e-PTFE patch because of a lower risk of post-operative adhesions.[10]

In conclusion, complete diaphragmatic agenesis is rare and difficult to diagnose in an asymptomatic adult patient, with no history of trauma. Failure of response to intercostal drainage in an apparently complete pneumothorax should lead to a suspicion. A CT is suggestive and surgical reconstruction usually provides good results.

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