ISSN: 2395-6429

INTERNATIONAL JOURNAL OF CURRENT MEDICAL AND PHARMACEUTICAL RESEARCH

IJCMPR

Available Online at http://www.journalcmpr.com

DOI: http://dx.doi.org/10.24327/23956429.ijcmpr20170056

CASE REPORT

AN ESPY OF AGENESIS OF LEFT DIAPHRAGM IN AN ANC PATIENT: A CASE REPORT

Jagadish Hedawoo[1]., *Varsha Barai[2] and Kaustubh Vaidya[3]

Department of Surgery, Government Medical College & Hospital, Nagpur

ARTICLE INFO

Article History:

Received 15th February, 2017
Received in revised form 8th March, 2017
Accepted 27th April, 2017
Published online 28th May, 2017

Key words:

Diaphragmatic agenesis, Hernia, Pregnancy, Thoracotomy

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 aneuploidy 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 the second trimester of pregnancy with agenesis of the left side of the diaphragm. Open diaphragmatic hernia repair done with dual mesh closure after which patient improved symptomatically and lung showed good expansion.

INTRODUCTION

Development of the diaphragm starts at the fourth week of gestation and completes by ninth week. While experimental studies have been carried out, to understand the organogenesis of the diaphragm and its congenital defects, the cause of agenesis or complete absence of diaphragm is not established [1]. The anomaly, originating in organogenesis, may result in diaphragm imperfection and can occur in various locations and sizes. Diaphragmatic agenesis is the most severe form of diaphragmatic defects and is associated with a high mortality of 40% to 62% [2] and associated cardiac anomalies. While traumatic diaphragmatic defects or congenital defects such as postero-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.

Diagnosis of agenesis in asymptomatic adult patients with no history of trauma is quite complex. For such a diagnosis, clinical manifestations such as dullness to percussion and absence of vesicular murmur in a lung field must be taken into consideration [3]. Failure of intercostal drainage on suspicion of pleural effusion or hemothorax should also be considered. The greatest difficulty regarding the approach to large diaphragmatic defects centers around the issue of when, how, and whether to operate. Due to the danger of intestinal incarceration and strangulation, immediate repair is essential in cases of partial defects, including those in asymptomatic individuals. The requirement for surgical correction in cases of large diaphragmatic defects is discussed from another perspective, as the free space provides movement of the herniated viscera, offering a low risk of intestinal complications [4]. Due to its rarity in adult patients, only anecdotal references are available in the literature. Here we report a case of an adult female who presented with progressive shortness of breath during second trimester of pregnancy with agenesis of the left side of the diaphragm.

Case Description

A 21 year old pregnant female presented to us with history of breathlessness. She was previously treated at a private hospital as a case of pneumothorax with ICD insertion. There was no history of trauma. When no ICD output was observed, it was removed and patient was referred to us.

At presentation, patient was 5 months pregnant with breathlessness and associated vomiting. Respiratory rate was normal with SpO_2 of 92%. Chest X-Ray showed gastric bubble in thorax. On Ultrasonography of thorax bowel loops were visible in left hemithorax with underlying lung collapse. MRI was suggestive of non visualisation of lay aspect and left body of diaphragm with upper pole of spleen, stomach, large bowel with mesentery in left hemithorax with mediastinal shift to right (Figure 1). Obstetric opinion was taken and prophylaxis was given for premature labour and abortion (progesterone and duovadilol). In view of increasing respiratory distress, patient was planned for diaphragmatic hernia repair. Consent for surgery with risk of abortion was explained and taken. Intra-operatively, stomach, bowel loops along with spleen was reduced (Figure 2).

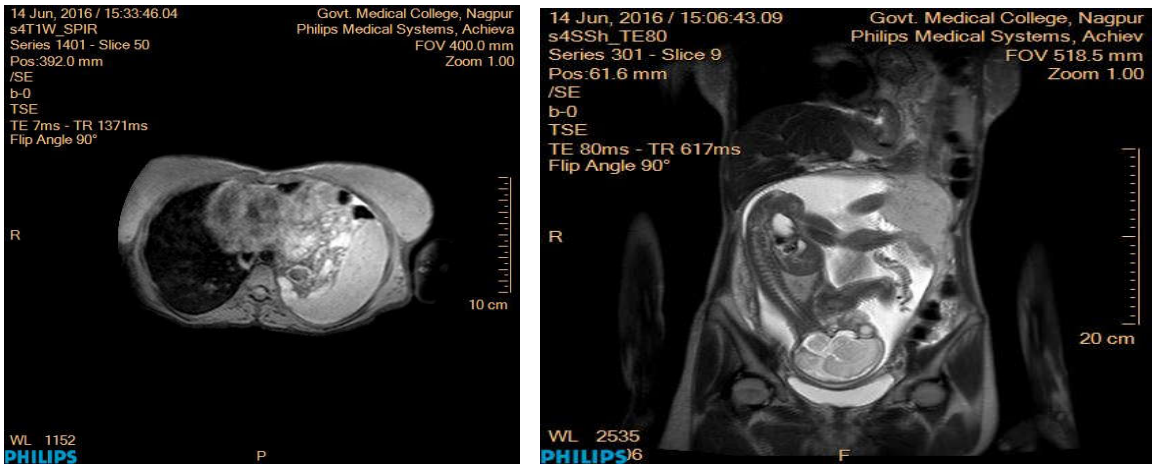

Figure 1 MRI images

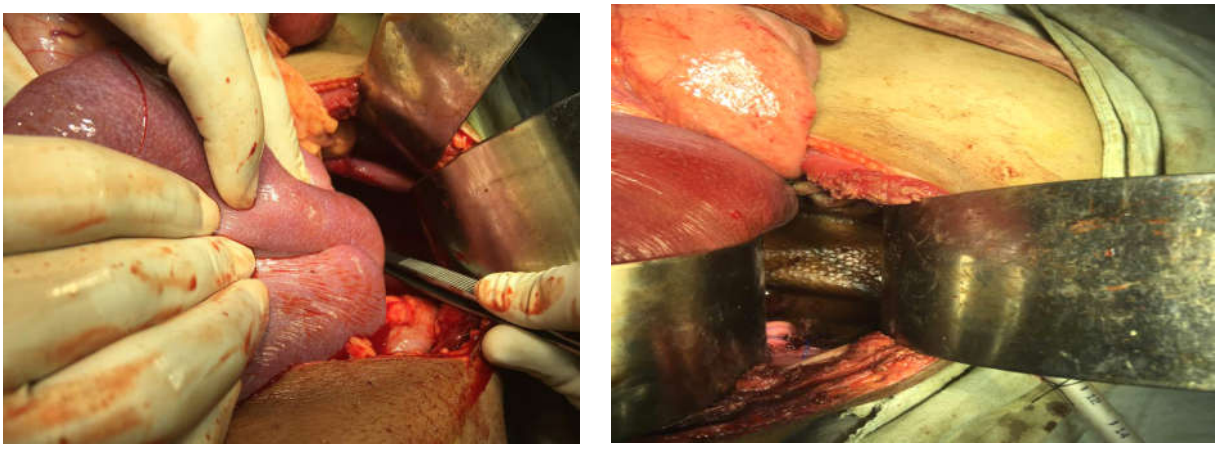

Figure 2 Intra-operative Findings

Open diaphragmatic hernia repair was done with dual mesh closure (figure 2.) of only skin (incisional hernia) to accommodate fetus and abdominal contents. Post-operative surgical and obstetric assessment was done daily. Patient aborted on day 4 and was discharged on post-op day 8 with regular follow ups and mesh herniorrhaphy for incisional hernia was done after 6 months. Patient is still on follow up and in good condition.

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 (CDH) (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 [5]. 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 [2,7]. 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 [8]. 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 mycophenolate 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 [9]. 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 [10], 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 surprisingly asymptomatic prior to pregnancy. Her symptoms started only in the 2nd trimester of pregnancy when lung function became further compromised by the displaced gravid uterus. A similar case, but of right sided diaphragmatic agenesis was reported by Ali SA *et al*. [11] where the patient was asymptomatic till 3rdtrimester of pregnancy.

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 [12]. 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 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 [3, 13]. 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 [14].

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. Anderson CD, Eliason J, Roberts JR, Sharp KW. Bilateral diaphragmatic agenesis in an adult complicated by a paraduodenal hernia. *Am Surg* 2003; 69:523-5.
4. Singh G, Bose SM. Agenesis of hemidiaphragm in adults. *Aust N Z J Surg* 1993; 63:327-8.
5. 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
6. Mirza B, Bashir Z, Sheikh A. Congenital right hemi diaphragmatic agenesis. Lung India 2012; 39:53-5.
7. Gale ME. Bochdalek hernia: prevalence and CT characteristics. *Radiology* 1985; 156:449-52.
8. 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.
9. 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.
10. Tzelepis GE, Ettensohn DB, Shapiro B, McCool FD. Unilateral absence of the diaphragm in an asymptomatic adult. *Chest* 1988; 94:1301-03.
11. Ali SA, Haseen MA, Beg MH. Agenesis of right diaphragm in the adults a diagnostic dilemma. The *Indian Journal of Chest Diseases & Allied Sciences*. 2016; 56: 121-5.
12. Ghanem AN, Chankun TS, Brooks PL. Total gastric gangrene complicating adult Bochdalek hernia. *Br J Surg* 1987;74:77
13. Atkinson JB, Poon MW. ECMO and the management of congenital diaphragmatic hernia with large diaphragmatic defects requiring a prosthetic patch. *J Pediatr Surg* 1992; 27:754-6.
14. Fei L, Saviano C, Moccia F, del Genio G, Trapani V, Nunziale A, *et al*. ePTFE soft tissue patch reconstruction of hemidiaphragmatic agenesis with late clinical presentation. *Hernia* 2008; 12:103-6.

****