Case Report

# Diaphragmatic agenesis in a young adult

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## ABSTRACT

Most neonates delivered with diaphragmatic agenesis (DA) do not live more than hours to days. However, very few affected cases survive the childhood period and even their adulthood if treated surgically or conservatively. 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. We have hereby reported a case of neglected left hemi-DA for more than three decades.

**Key words:** Adulthood, Diaphragmatic agenesis, Dyspnea

The absence of diaphragmatic development in any portion of the diaphragm is known as diaphragmatic agenesis (DA). Because congenital diaphragmatic hernia permits intra-abdominal viscera to be forced into the thoracic cavity, respiratory failure progresses, causing activity-induced and even resting dyspnea, pulmonary hypertension, and adult gastrointestinal complaints. This ultimately results in the death of the neonate [1]. The biggest diaphragmatic defect is complete diaphragmatic dome agenesis. This is an extremely rare condition, and careful assessment is needed to differentiate between diaphragmatic hernia and agenesis of the diaphragm [2]. DA is considered one of the rare congenital malformations of the diaphragm and is reported in 6% of all congenital diaphragmatic herniation (CDHs) [2]. It is an uncommon deformity in adulthood because the cause of mortality usually happens in the first few hours of existence. In asymptomatic adult patients without a history of trauma, the diagnosis of agenesis can be highly intricate [3]. It is very rare to see an adult with complete agenesis of the lung in adult life.

Hence, we hereby report a case of complete agenesis of the diaphragm presenting in the fourth decade of life.

## CASE REPORT

A 38-year-old female presented with a history of breathlessness on exertion for 2–3 years, which was aggravated for 4 months. Breathlessness was Modified Medical Research Council (MMRC) grade 1 for 2–3 years which progressed over the past 4 months to MMRC grade 2. Breathlessness aggravated on strenuous activity and in the supine position and was relieved in a sitting position or taking rest. There was no history suggestive of seasonal or diurnal variation. Her symptoms persisted despite taking antibiotics and bronchodilators over the previous months. There was no history of cough, expectoration, wheezing, fever, or chest pain. There was no history suggestive of cardiac ailments. There were no complaints of nausea, vomiting, dyspepsia, or bloating sensation. She has a known case of hypothyroidism for 12 years, hypertension for 2 years, and a known case of type 2 diabetes mellitus for 1 month. There was no past history of trauma or surgery.

On examination, the patient was obese, with a body mass index of 27. Vital parameters were within normal limits with $SpO_2$ of 94%. Respiratory system examination revealed decreased thoracic movements on the left side. Tactile vocal fremitus and vocal resonance were decreased on the left side anteriorly and posteriorly. Auscultation revealed peristaltic bowel sounds in the left basal areas.

Chest X-ray revealed elevation of the left dome of diaphragm and silhouetting of the left heart border (Fig. 1). Contrast-enhanced computed tomography abdomen was advised for confirmation of diagnosis which revealed elevated left hemi diaphragm and thinned out with superior migration of the left lobe of liver, stomach, spleen, bowel loops, and splenic flexure into thoracic cavity on the left side with significant compression on left lower lobe resulting in atelectasis and mediastinal shift. All these features were suggestive of eventration of the left

| Access this article online | |
|---|---|
| Received - 31 December 2024<br>Initial Review - 14 January 2025<br>Accepted - 18 April 2025 | Quick Response code |
| **DOI:** 10.32677/ijcr.v11i5.4979 | |

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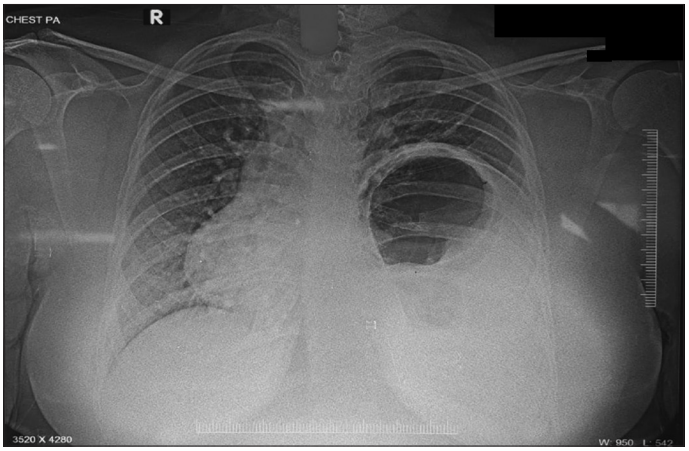


**Figure 1: Elevated left dome of the diaphragm**

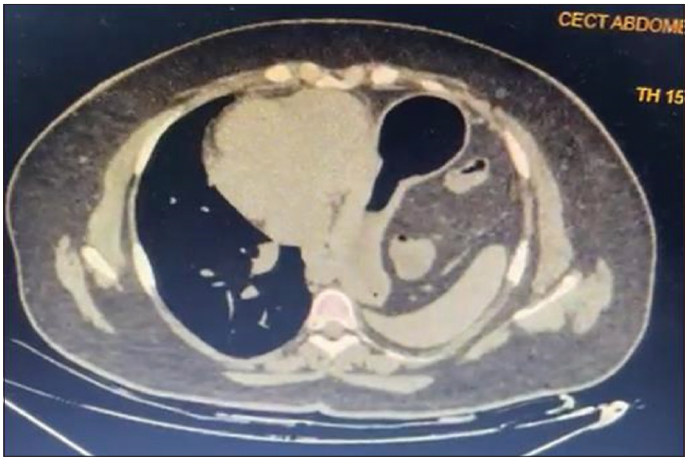


**Figure 2: Herniation of abdominal contents in the left hemithorax**

diaphragm (Fig. 2). However, agenesis of the left diaphragm was also considered for differential diagnosis.

The patient was referred for surgical correction of the diaphragm to department surgery. A diagnostic laparotomy was performed to confirm the diaphragmatic defect. Diagnostic laparotomy showed complete agenesis of the diaphragm in this case (Fig. 3). All the abdominal contents from the left side had gone into the left thoracic cavity. Mesh repair was not attempted since there were no remnants of the diaphragm.

Postoperatively, the course was uneventful, and the patient had a saturation of 93% in room air. During the 1-year follow-up of the patient, there was grade I breathlessness, and she is able to perform daily activities.

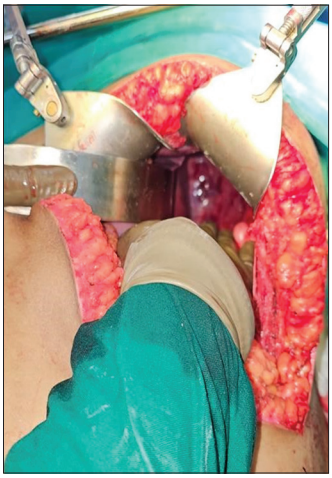

**Figure 3: On laparotomy, a complete absence of the left diaphragm is observed**

## DISCUSSION

The diaphragm is an important muscle of respiration and keeps the abdominal viscera from herniating into the chest and keeps the thorax and abdomen apart [1]. The four major embryologic fountains – the septum transversum, pleuroperitoneal membrane, dorsal esophageal mesentery, and the body wall-join in developing this dome-shaped septum that separates the abdominal and thoracic cavities around the 3–4$^{th}$ month of gestation [2]. During the 3$^{rd}$ 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. Because of an earlier closure of the right pleura-peritoneal hiatus, left defects are more common than right defects [1]. Maldevelopment of the diaphragm from birth can result in lung hypoplasia, pulmonary hypertension, and symptoms of digestive tract obstruction [2].

Causes for pulmonary hypertension in congenital DA or hernias include a reduction in the total pulmonary vascular bed for a decreased number of vessels per unit of the lung, along with pulmonary vascular remodeling. The muscle precursors of the diaphragm are cervical somites. Migrating to the pleuroperitoneal folds, they start to proliferate and differentiate [3]. The genes responsible for this process interact with one another in the embryonal pathway for the development of the brain, heart, lungs, diaphragm, kidneys, and pancreas. Two hypotheses are proposed to explain this phenomenon: One suggests maldevelopment during organogenesis leading to bilateral hypoplasia, whereas the other posits that later-stage compression of the ipsilateral lung occurs due to herniation of abdominal viscera [4].

In 1988, Tzelepis *et al*. [5] reported the first case of DA in an adult, and the patient had a prolonged lower lobe infiltration; however, it was eventually established that the patient had a complete absence of the left hemi-diaphragm. CDH is a rare condition, manifesting in approximately one out of every 4000 live births. The mortality rates for infants with CDH are notably high, with only two-thirds of affected live-born infants surviving to reach their first birthday [6].

DA is considered one of the rare congenital malformations of the diaphragm and is reported in 6% of all CDHs [2]. A diagnosis of CDH can be established during the prenatal period, whereas isolated DA is deemed a rare occurrence. DA is usually identified very early in infancy and is associated with a notable mortality rate ranging from 38% to 62%, depending on the presence of other congenital anomalies [2]. In nearly all cases, agenesis results in the demise of patients within their 1$^{st}$ h or days of life. However, individuals who manage to survive into adulthood typically remain asymptomatic until their third decade [3]. The manifestation of symptoms associated with a diaphragmatic

hernia can vary, contingent upon the compensatory respiratory mechanisms, as well as the site and size of the hernia [7]. In adults, left-sided diaphragmatic defects commonly lead to symptoms due to visceral herniation. Conversely, right-sided agenesis may present with few symptoms or even remain asymptomatic, often attributed to the liver's presence, which acts as a barrier preventing other viscera from herniating through the diaphragmatic defect [1]. Lung hypoplasia and immaturity occur in cases of the ipsilateral side of the defect [2].

Digestive symptoms, including nausea, vomiting, flatulence, abdominal pain, epigastralgia, constipation, anorexia, difficulty gaining weight, and varying levels of gastroesophageal reflux, may often coexist [3]. In instances of partial or complete agenesis of a hemidiaphragm, patients typically experience either no complications or only subtle ones [8]. It is only when symptoms such as worsening dyspnea or the obstruction of bowel loops into the thorax arise that they are prompted to seek care at a surgical center. Dyspnea, coughing, chest pain, and acute respiratory distress syndrome are common respiratory symptoms seen. The differential diagnosis to be considered in this condition includes diaphragmatic hernia through the diaphragmatic defects, eventration of the diaphragm, which may be congenital or acquired due to trauma, or diaphragmatic paralysis due to phrenic nerve palsy [9]. Izeldi *et al*. [9] also reported a case of DA in an adult presenting with acute onset of breathlessness due to a secondary infection in the lungs.

Although primary closure is the mainstay treatment method for many diaphragmatic hernias, in the cases of larger defects or agenesis, direct suturing is difficult because of the absence of adequate muscular tissue or due to the presence of the adhesions [2]. Avoid absorbable meshes due to their insufficient long-term support, as they are quickly absorbed by the body [1]. Prosthetic materials have been introduced, employing various literary techniques to create a new diaphragm [1]. Several methods have been suggested for repairing these defects, including direct suturing to the liver or prerenal fascia, utilizing free grafts or abdominal/intercostal muscle flaps, employing prosthetic materials, and utilizing bovine pericardium [1]. In the present case, mesh repair could not be done due to the complete absence of the diaphragm on the left side.

## CONCLUSION

The majority of DA instances pass away in the neonatal population; these cases are either undiagnosed or discovered during an autopsy. Hence, rare in the adult population, respiratory and digestive disorders are the most prevalent symptoms they present in adults. In our case, breathlessness was the main symptom, and it was diagnosed in the fourth decade of life.

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*Funding: Nil; Conflicts of interest: Nil.*

**How to cite this article:** Prabha SS, Gaude GS, Togale M. Diaphragmatic agenesis in a young adult. Indian J Case Reports. 2025; 11(5):214-216.