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Case Report

Anatomy Section

Eventration of Diaphragm: A Rare Cadaveric Case Report

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ABSTRACT

The respiratory diaphragm is a barrier between the thoracic and abdominal cavities. It is a chief skeletal muscle of respiration that plays a critical role in the process of inspiration. The defective diaphragm may be clinically present with or without obvious symptoms. Depending on the severity of its defects, it can lead to mild to severe fatal consequences. Protrusion of abdominal contents into the thoracic cavity through the weakened or defective part of the diaphragm is known as a diaphragmatic hernia. Such herniations will exist either in the form of congenital birth defects or acquired defects in the diaphragm. An acquired hernia may be due to spontaneous or iatrogenic causative factors. Congenital Diaphragmatic Hernia (CDH) can occur due to the disruption of various cellular mechanisms involved in organogenesis during the gestation period. Such herniations may exist with or without content protrusions into the cavity of the thorax, later referred to as Eventration of the Diaphragm (ED). In the Department of Anatomy at JSS Medical College, Mysuru, Karnataka, India, a rare case of diaphragmatic eventration was noticed in a male cadaver aged about 70 years, during the routine dissection class of preclinical medical students. In this rare case report, diaphragmatic eventration along with various factors involved in its presentation would be considered holistically.

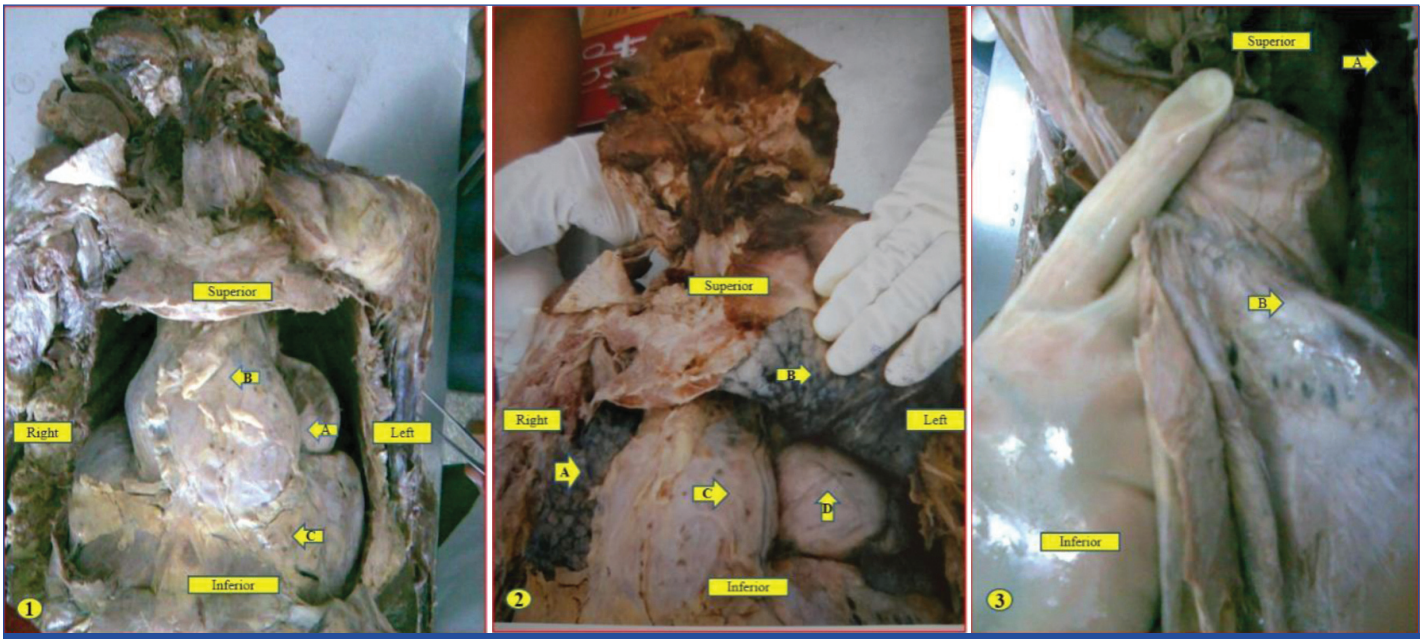
Keywords: Congenital, Gestation, Organogenesis

CASE REPORT

In the Department of Anatomy at JSS Medical College, Mysuru, Karnataka, India a rare case of eventration of the diaphragm of the left-side was noticed during the dissection of the thorax in a male cadaver aged about 70 years [Table/Fig-1]. It was initially noticed as a protruding blind pouch from the posterolateral aspect of the superior surface of the diaphragm facing toward the thoracic cavity. It was lying close to the mediastinal surface of the left lung and pericardium of the heart [Table/Fig-2]. The development of both lungs was found normal. Later, on opening the abdominal cavity the same herniated diaphragmatic pouch was followed and it was found to be an empty sac [Table/Fig-3] without protrusion of any contents of the abdomen into the thoracic cavity. Later the condition was identified as an "Eventration of the Diaphragm".

DISCUSSION

The diaphragm is a musculoaponeurotic organ which acts as a barrier between the thoracic and abdominal cavity; plays an important role in respiration. Its normal development is essential for infant breathing after birth. Organogenesis is a crucial part; it starts during the embryonic period. The first trimester of gestation is a critical phase; disruption of the molecular signalling mechanism may result in teratogenic effects causing foetal malformations. Morphogenesis is further driven through intercellular signalling mechanisms between groups of cells that are derived from different germ cell lines. During this phase molecular events are tightly regulated; their subtle disruptions can result in a wide range of defects in an organ or system formation [1].



[Table/Fig-1]: Showing eventration of the diaphragm after removing lungs. A) Showing eventration of diaphragm; B) Pericardium; C) Diaphragm.
[Table/Fig-2]: Showing posterolateral eventration of the diaphragm in-situ. A) and B: Right and Left Lungs; C: Pericardium; D: Diaphragm eventration.
[Table/Fig-3]: A) Lungs; B) Diaphragm eventration showing blind pouch without any contents. (Images from left to right)

At the end of the gastrulation process, the germ layers are formed. Later the actual phase of organogenesis begins, which will continue till the end of 2nd month of the gestation period. The cells of each germ layer will differentiate into specialised and super-specialised groups which in turn form a basis for specific tissue or organ formation [2]. The diversified embryological components are involved in the development of the human diaphragm which occurs during the 4th-12th weeks of the gestation period [3]. They are including, the septum transversum-which gives rise to the central tendon of the diaphragm, the pleuroperitoneal membrane-which divides the coelomic cavity into the thoracic and abdominal cavities and somites which are derived from paraxial mesoderm contributing the musculature of the diaphragm [4]. By birth, if the diaphragm wall is defective, the herniation of visceral contents protruding into the thoracic cavity is referred to as CDH [5].

The CDH occurs in about 1 of 2500 live births. The left-side of the diaphragm defect is involved more often than the right i.e., in 80% of cases. Posterolateral CDH is known as Bochdalek hernia; it may possess protruded contents of the abdomen into the thorax, which is a relatively common clinical anomaly. The probable reason being the left pericardioperitoneal canal is larger and closes much later than the right [6]. Sometimes such diaphragmatic herniations are clinically presented as a hallow protrusion of part of the diaphragm or the entire diaphragm without any protrusion of contents into the cavity of the thorax called "Eventration of Diaphragm" (ED). It may be a congenital or acquired anomaly [7]. Eventration is a rare presentation seen in 1 in 10,000 live births [8]. This is usually seen as secondary to defective diaphragm formation with weak musculature or defective nerve innervations [5].

A study has underscored the importance of molecular signalling interactions between the diaphragmatic muscle fibres and its innervating phrenic nerve fibres in establishing proper neuromuscular junctions. This is crucial in the existence of a fully functional diaphragm; its failure can cause undue protrusions of the weakened diaphragmatic wall leading to eventration [4]. The aetiology of CDH largely remains unclear and is currently thought to be multifactorial. CDH can be associated with cardiac, gastrointestinal, and genitourinary anomalies or with chromosomal aneuploidy such as trisomy 18,13, and 21 [9]. The present case report highlighted one such rare case of ED herniation which could be caused due to neurogenic or muscular aplasia.

Pregnancy is a critical period and adequate dietary supplementation is vital during this period to avoid congenital birth defects. Micronutrient deficiency may result in the death of the foetus or it can lead to congenital malformations. Vitamin A (Retinoic acid) is one such micronutrient that plays an important role in achieving the uninterrupted growth and development of the foetus. Basic experiments have shown that vitamin A deficiency can disrupt the subtle molecular signalling patterns resulting in CDH [10]. Altered values of vitamin A or Retinoic Acid (RA) can influence the neurectoderm and endodermal germ layers which are involved in the development of several organs like the spinal cord, brain, limbs, pancreas, lungs, somites, etc. RA-targeted genes could be a cause behind developmental organ malformations [11].

The diaphragm is a chief inspiratory skeletal muscle that plays an important and active role in the process of respiration. The central tendon of the diaphragm is a "crux" point where different myofibres will come for their insertion around it. Diaphragm muscle progenitors are derived from C3-5 cervical somites which are influenced by different genes for their formation. Mutations in *Pax3* genes are dominantly expressed in somitomeres which can lead to defective or weak musculature resulting in ED. The abnormal migration of myoblast cells may result in the improper or incomplete formation of the musculotendinous wall [12].

There are different types of congenital diaphragmatic hernias, the most commonly reported one is Bochdalek type hernia which constitutes about 70%, Morgagni type hernia 27%, and central type hernia 2-3% [13]. Posterior ED without any visceral contents may resemble the 'Bochdalek hernia of the diaphragm" where radiological investigations play an important role in the differential diagnosis [14]. Developmental weakness in diaphragmatic muscle fibres or their improper formation may lead to herniation. Such herniations may contain visceral (abdominal) organs that lead to the incidences of CDH. Sometimes such herniations may remain as a point of protrusion of the diaphragmatic wall toward the thoracic cavity without any contents referred to as ED usually it is presented without pulmonary hypoplasia [8]. Based on the clinical findings, the severity effect of ED can be noticed in infants; it may be associated with symptoms of severe respiratory distress. But in the case of adults, it may remain asymptomatic, hence often ED is found as an incidental finding in radiological investigations. In a symptomatic patient, it is presented with dyspnoea on physical exertion, or with signs of orthopnoea due to elevated movements of the diaphragm [7].

The ED may be seen as unilateral or bilateral wall protrusion; it shows paradoxical movements in breathing. Being an important inspiratory muscle, the tone of the diaphragm musculature is crucial for effective breathing. Paralysed or weakness in the diaphragm musculature can affect optimum lung functionality, showing reduced lung function parameters like Vital Capacity (VC), Total Lung Capacity (TLC), and Residual Capacity (RC) with hypoxia. The surgical repair to strengthen the weak diaphragmatic musculature can be attempted through thoracotomy or laparotomy. The surgical repairing through strengthening the musculature of the diaphragm called "Plication" has shown considerable improvement in establishing pulmonary functions. A surgically strengthened diaphragm will help to accommodate good air space for the proper expansion of the lungs [15].

The clinical manifestations of CDH and ED are often overlapping. Hence to differentiate between these conditions' surgeons need a keen clinical evaluation. The CDH contains protruded abdominal contents like intestines (viscera) into the thoracic cavity through the malformed or disrupted structure of the diaphragm. Often such events will cause mechanical compression of the lung, which may be associated with the underdevelopment of lungs called congenital pulmonary hypoplasia [16]. The CDH associated with pulmonary hypoplasia is seen with reduced formation of airway branching pathways caused due to the compression of developing lung buds because of undue protrusion of abdominal contents into the thoracic cavity on developing foetal lung tissue [13].

Explicit manifestation of Bochdalek hernia is relatively rare in adults most often it will clinically manifest during the very early life of an individual [17]. Such incidences are likely to be associated with respiratory symptoms due to pulmonary hypoplasia. Persistent pulmonary hypertension is likely to manifest as a tertiary symptom associated with an underlining congenital diaphragmatic defect followed by developmental lung hypoplasia. If Bochdalek hernia is found in adults it is likely to present with complications related to the gastrointestinal tract probably due to compression of viscera-like intestines [5].

The prognosis of isolated CDH is generally better than CDH complicated by other associated multiple anomalies. Despite the unclear aetiology of CDH over the past few decades, reports have suggested increasing trends of survival in infants with CDH with the medical and surgical advances in the management of CDH,

with the reported overall 70-90% of survival [18]. The symptoms of diaphragmatic hernias may range from an asymptomatic condition to a life-threatening clinical manifestation. In the present case, the development and morphology of the lungs were seen as normal. Any visceral organ protrusion or displacement into the thoracic cavity had not been noticed which may indicate the probability of weak phrenic nerve innervations or injury to the phrenic nerve itself or weak muscle tone. Hence these cadaveric findings may be suggestive of probable weakness in the diaphragm causing the eventration-like manifestation.

Some cases of eventrations were found with a previous history of infections coincidentally associated with ED [19,20]. Some cases were reported with eventration of the solid organ like the liver on the right-side [21]. Two separate rare cases were noticed, one with multiple organ protrusion along with the displacement of the right kidney into the thorax [22]. A rare case was reported with organoaxial gastric volvulus, both were associated with the incidental eventration of diaphragm findings [23]. But most of the incidental radiological findings were frequently associated with the protrusion of coils of intestines as a common entity towards the chest cavity from the point of diaphragm wall weakness [24,25].

In the present case study, the unilateral ED could be due to the weak tone of its musculature rather than the explicit lack of muscle fibres. But in the case of herniation, the muscle fibre continuity may be affected, it may involve the whole or a part of the diaphragm musculature, probably due to myoblast cell migration failure. The lungs were well formed without any grossly identifiable defect. Any traumatic injury had not been noticed on the body or injury to the diaphragmatic musculature hence any accidental injury can be ruled out because donated cadavers were having a history of natural death. The eventration may facilitate just protrusion of abdominal contents to some extent without affecting the functionality of lungs or other mediastinal contents because the existing pouch of musculature can act as a physical barrier to some extent. In the present case, the cadaver was devoid of any abdominal contents in the pouch space (eventration), and no physical sign of compression of contents in the thorax cavity was found. [Table/Fig-4] shows the previous studies on the ED [19-25].

S. No.	Author's name	Place of study	Findings reported
1.	Paudel K and Dahal S, 2021 [19]	Nepal	A 72-year-old lady presented right-side ED with the protruded right lobe of the liver associated with a history of chronic tuberculosis infection.
2.	Pradhan P et al., 2020 [20]	Nepal	A 47-year-old lady presented left ED with underlying bowel associated with a previous history of infection.
3.	Andrew O et al., 2018 [21]	New York	A 43-year-old female presented right-side ED with protrusion of the right lobe of the liver and omental vascular structures.
4.	Carrasco A and Castro R, 2018 [22]	Peru	A 17-year-old female patient presented right ED with loops of the intestine, right kidney, and liver noticed.
5.	Shah NN et al., 2008 [24]	Mexico	60-year-old male presented left ED with organoaxial gastric volvulus.
6.	Souza-Gallardo LM et al., 2016 [23]	Aligarh	A 21-year-old female presented left ED with bowel loops.
7.	Mantoo SK and Mak K, 2007 [25]	Singapore	A 51-year-old male presented left ED with high infiltration of bowel loops on the left-side of the chest.
8.	Present study 2022	Mysuru, India	Left-sided ED without any contents.

[Table/Fig-4]: Previous studies/case reports on the eventration of diaphragm [19-25].

CONCLUSION(S)

In the present case, a report of left-sided eventration of the diaphragm was noticed in a male cadaver during the routine dissection class of preclinical medical students. The ED is a rare clinical presentation, its explicit clinical manifestations are rare and often they are incidental radiological findings. Its symptoms may mimic or overlap with other types of diaphragmatic hernias, need to elucidate the exact pathology for its presentation. An incidental radiological finding of ED may require a detailed evaluation of the previous surgical history of the patients. In the event of preclinical medical teaching and learning anomalous cadaveric findings with ED act as a real trigger that can create synchronisation of preclinical subject understanding in the direction of their clinical applications.

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