Heart on the left, diaphragm on the right: A case of congenital diaphragmatic eventration

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1 | INTRODUCTION

Diaphragmatic eventration (DE) is a rare disease entity defined as a partial or complete elevation of one or both the hemidiaphragm, due to muscular or nervous dysfunction.1 DE can present as a congenital defect due to paucity or absence of varying degrees of muscle fibers or, acquired due to phrenic nerve injury.2 This report highlights the case of a right-sided DE in a 5-month-old male infant presenting with recurrent lower respiratory tract infections and ongoing pneumonia.

2 | CASE REPORT

A 5-month-old male infant presented to the University of Health Science, Pakistan, with recurrent episodes of fever and non-productive cough which had increased in frequency in the last month. The infant was full-term and delivered via spontaneous vaginal delivery at home to a 30-year-old mother. The mother was screened and found negative for HIV and hepatitis panel and denied any use of alcohol, tobacco, and illicit substances throughout the course of her pregnancy. The infant weighed 2500 g at
birth; cried immediately after birth and has been breastfeeding without signs of distress. The infant had been meeting the required motor and language milestones along with good social maturation. Family history was significant for a consanguineous marriage along with the uneventful spontaneous vaginal delivery of two male babies who are currently healthy. The patient was immunized as per the national immunization schedule.

On admission, the infant was found to be irritable and inconsolable. General physical examination was significant for a fever (102°F). Chest examination revealed decreased movements and breath sounds in the right infra-mammary, infra-axillary, and infra-scapular areas. Upon auscultation, crepitations were present in the right lung field. Initial laboratory investigations displayed an elevated total leukocyte count of 15,000/mm³. Inflammatory markers such as C-reactive protein (CRP) and erythrocyte sedimentation rate (ESR) were negative. Hemoglobin and metabolic panel were also within normal limits. Chest X-ray (CXR) was performed and showed that the dome of the diaphragm was raised on the right hand side as seen in Figure 1A. CT scan of the chest confirmed the diagnosis of a right-sided diaphragmatic eventration with consolidative changes visualized in Figure 1B.

To treat ongoing pneumonia, the infant was empirically started on injection ampicillin, cloxacillin, and cefotaxime. He was also treated with albuterol and ipratropium bromide nebulization, dexamethasone, antipyretics, and multivitamin supplementation. Thoracoscopic plication of the diaphragm of the right side was planned. Under general anesthesia, the infant was placed on the left lateral decubitus position and a 3mm port was inserted in the 4th intercostal space at the posterior axillary line. A second 3mm incision in the 6th intercostal space of the right anterior axillary line was used to insert the Maryland grasper. Upon visualization of the diaphragm, right-sided eventration was confirmed. There were no adhesions with abdominal viscerae present. The diaphragm was plicated using 2–0 silk. Chest tube of appropriate size was placed in the right 5th intercostal space and closure was done in layers.

The immediate postoperative period was uneventful and the infant was able to maintain normal oxygen saturation on room air. Repeat CXR displayed a flattened hemidiaphragm on the right side. After successful management the infant was discharged and was symptom-free at his 3-month follow-up appointment. CXR was repeated and was unremarkable showing adequate inflation of bilateral lungs and flattened diaphragm.

3 | DISCUSSION

Diaphragmatic eventration, often described as the neu-rogenic muscular aplasia of the diaphragm, has been found to occur in varying age groups of the population.1 Congenital diaphragmatic eventration can be attributed to the inability of myoblasts to properly migrate to the septum transversum, thus leading to the substitution of muscle fibers by fibroelastic fibers.3 This histopathological picture of the diaphragm can be used to differentiate it from acquired causes of DE including birth trauma, phrenic nerve injury, nerve compression, pneumonia, and multiple sclerosis.1,4

On top of the challenging presentation of DE, barriers to health care such as poor socioeconomic status, add to the complications. Home-deliveries carried out by midwives have become a cultural norm in Pakistan in both rural and urban settings and have been correlated with a higher risk of maternal mortality.5 These practices have been attributed to the low socioeconomic status and deep-rooted cultural beliefs prevailing in these regions. The delayed diagnosis of DE in the present case, similarly, may be due to the family’s psychosocial views and inaccessibility of the medical healthcare. Recognizing cultural limitations is necessary as DE has widely been underreported due to its largely asymptomatic presentation. A summary table of our patient Physical exam findings has been presented in Table 1.

The non-contracting, higher position of the diaphragm results in lung collapse followed by atelectasis.
predisposing the individual to bronchial or parenchymal infections. Presenting symptoms often include cough, dyspnea, chest pain, and cyanosis, and at times, gastrointestinal symptoms such as nausea, vomiting, abdominal pain, and acid reflux. In infants, these symptoms can be extremely debilitating due to the underdeveloped thoracic cage and intercostal muscle weakness resulting in paradoxical respiration ultimately necessitating the use of mechanical ventilation.

The workup of DE primarily includes imaging modalities including the use of ultrasonography, chest x-ray, and computed tomography (CT) scan. It is vital to exclude intrathoracic, mediastinal, or abdominal masses such as diaphragmatic hernia before concluding the diagnosis of DE. Fluoroscopic sniff test has been used to differentiate diaphragmatic eventration and paralysis in cases of unilateral diaphragmatic paralysis, however, this test was not used in our case. As recurrent lower respiratory tract infections are a common complexity arising from DE, it is of primary interest to treat the underlying infection and provide respiratory support with oxygen supplementation in patients presenting with hypoxia. A summary of previously published cases has been presented in Table 2. Nutritional supplementation is vital in infants presenting with DE as undernourishment due to poor feeding often accompanies the condition. Symptomatic DE presenting with respiratory distress, failure to thrive, recurrent pneumonia, and failure to wean of ventilator support have been shown to benefit greatly with diaphragmatic plication with a good prognosis and improvement of quality of life.

Diaphragmatic plication has been achieved through thoracotomy, thorascopic, and laparoscopic surgeries. Thoracoscopic surgeries have been proven to be more advantageous with a lower length of hospital stay, lower rate of complications, and better prognosis. Laparoscopic surgeries are also associated with lesser pain as intercostal nerve damage is avoided. The placement of a prosthetic mesh has been successful along with plication in cases of extreme amyotrophy, however at the cost of increased cost and chance of infection. Possible complications of diaphragmatic plication include pneumonia, dyspnea, pulmonary edema, and pleural effusion. These complications, however, were not observed in our case.

Symptomatic diaphragmatic eventration is associated with high morbidity and failure to thrive if not treated promptly. Adequate workup to exclude underlying conditions, nutritional status, associated abnormalities, and treatment of the same is vital to ensure better survival rates in infancy. Minimally invasive procedures like laparoscopic plication of the diaphragm are found to be very effective in the treatment of diaphragmatic eventration with a low incidence of complications and a good prognosis.

### CONCLUSION

Congenital diaphragmatic eventration (DE) is a rare pathology that can be fatal if left untreated. DE’s are difficult to diagnose as they can present without symptoms thus requiring intricate management. Infants dealing with DE are at an increased risk of morbidity as their thoracic cage is underdeveloped leading to life-threatening complications including failure to thrive. This case demonstrates the successful outcome of the patient due to accurate diagnosing of congenital DE, and the performance of minimally invasive procedures such as laparoscopic plication.
| First author et al. | Year of publication | Country       | Age                          | Gender | Comorbid                                      | Symptoms                                                   |
|---------------------|--------------------|---------------|------------------------------|--------|-----------------------------------------------|-----------------------------------------------------------|
| Mouroux J. et al.   | 2005               | France        | Mean age = 57.7 ± 14.8 years | 4 males & 8 females | Trauma (n = 9), Charcot-Marie disease (n = 1), calcified para-aortic nodes (n = 1) | Dyspnea (n = 12), palpitations (n = 4), chest pain (n = 3), dyspepsia (n = 2) & recurrent pneumonia (n = 1) |
| Shwaartz C., et al. | 2017               | USA           | 31                           | male   | Hypertension                                  | Palpitations, shortness of breath & chest pain              |
| Zhao S., et al.     | 2020               | China         | Median age: 12.2 months      | 90 male, 35 female | 19 with congenital heart disease, 16 with congenital pulmonary dysplasia, 8 with pectus excavatum, 4 with hiatal hernia, 3 with pectoral malformations | Cough, asthma, dyspnea, recurrent respiratory tract infections, milk refusal, vomiting & arrhythmia |
| Omenai SA., et al.  | 2020               | Nigeria       | 69                           | male   | Intestinal malrotation, renal agenesis, thoracoabdominal compartment syndrome, dilated cardiomyopathy | Easy fatigability, orthopnea, paroxysmal nocturnal dyspnea, pedal swelling, worsening breathlessness, early satiety and abdominal pain |
| Shaher A et al.     | 2019               | Saudi Arabia  | Early 20                      | male   | No known comorbid                              | Shortness of breath, abdominal distention                   |
| Wu S. et al.        | 2015               | China         | 10.28 ± 2.35 months          | 128 male, 49 female | Hypoplastic lung, congenital heart disease, cryptorchidism | asymptomatic, Rapid breathing, vomiting, recurrent respiratory infections |
| Carrasco A. et al.  | 2018               | Peru          | 17                           | female | Thoracic renal ectopia                         | Dry cough, chest pain, respiratory distress, bronchial spasms, repetitive episodes of bronchial asthma |
| Kang H., et al.     | 2019               | Korea         | 28 months                    | male   | osteochondroma, premature,                    | Asymptomatic                                               |
| Boufidou A., et al. | 2011               | Greece        | 70 years old                 | female | N/A                                           | Retrosternal, stabbing pain with radiation to precordial area |
| Guzman JPS., et al. | 2017               | Philippines   | 32                           | female | None                                         | Intermittent dyspnea, epigastric discomfort                |
| Deveer M., et al.   | 2013               | Turkey        | 64                           | male   | None                                         | Sudden onset severe dyspnea after strong cough             |
| Gunadi., et al.     | 2020               | Indonesia     | 16 days old                  | male   | None                                         | Respiratory distress, decreased breath sounds              |
| Chowdhury S., et al.| 2018               | Saudi Arabia  | 48 yo                        | Male   | None                                         | Spontaneous breathing, decreased air entry, increased respiratory rate |
| Kasdallah N., et al.| 2017               | Tunis         | 4 days old                   | male   | Neonatal gastric perforation                  | Bilious vomiting, refusing feeds, jaundice, respiratory distress |
| Diagnostic criteria | Final Diagnosis | Surgical Management | Medical Management | Sepsis | Outcome (Dead/ Survived) |
|---------------------|----------------|---------------------|--------------------|--------|-------------------------|
| CT upper abdomen, MRI and/or phrenic electromyography | Diaphragmatic Eventration | VATS with 2 thoracoports & 4cm mini- thoracotomy- Diaphragmatic plication | Post-op follow-up included physical exams, chest roentgenogram & spirometry at 3, 6 & 12 months | None | Survived (n = 12) |
| Chest Radiograph, CT Abdomen, Barium enema | Left Diaphragmatic Eventration | Laparoscopic exploration and mesh removal with left thoracotomy, diaphragmatic plication | Post op radiograph, Follow-up at 1 and 3 weeks | None | Survived (n = 1) |
| CXR, CT or GI radiography | Congenital Diaphragmatic Eventration | Thoracotomy on R. diaphragmatic eventration & laparotomy for Left Diaphragmatic Eventration. Transthoracic diaphragm plication & transabdominal diaphragm plication | Yearly radiological exams | None | Survived (n = 124) Died (n =1) |
| ECHO, Autopsy | Thoracoabdominal compartment syndrome due to right hemidiaphragm eventration | N/A | N/A | N/A | Died |
| Chest radiograph, acidic pH, lactate 8 mg/dl | Right- sided Diaphragmatic Eventration | The patient was shifted to OR, norepinephrine infusion was started. Midline laparotomy was done and a huge colon was encountered and eviscerated | Patient was resuscitated with 4L of 0.9% normal saline with no urine output, and intubated with minimal dose sedation (25 mcg fentanyl) | Septic Shock | Died |
| CT Abdomen, ECHO, CXR | Congenital Diaphragmatic Eventration | Diaphragmatic Plication | No recurrence at annual follow-up | None | Survived (n = 177) |
| CT, CXR | Diaphragmatic Eventration | Laparoscopy, posterolateral thoracotomy, hemidiaphragm plication | None | Survived |
| CXR, Fluoroscopy and ultrasonography | Congenital diaphragmatic eventration | N/A | None | Survived |
| CXR, thoracic CT | Diaphragmatic eventation | ? | No recurrence of symptoms | none | Survived |
| CXR, CT chest | Congenital left diaphragmatic eventration | Diaphragmatic eventration via abdominal approach | Incentive spirometry, deep breathing exercises, 2 year follow-up no symptoms | None | Survived |
| CT Thorax | Diaphragmatic eventration | Laparoscopy | N/A | None | Survived |
| CXR, CT | Congenital diaphragmatic eventration | Hemidiaphragm plication | Mild cough at 6 month follow-up | None | Survived |
| CXR, FAST, chest CT | Diaphragmatic eventation | Patient refused | No complaints on 11th day post admission | None | Survived |
| CXR, ultrasound | Congenital diaphragmatic eventration | Laparotomy & diaphragmatic plication | Infant well at 15 months old | None | Survived |

(Continues)
TABLE 2 (Continued)

| First author et al. | Year of publication | Country | Age | Gender | Comorbid | Symptoms |
|---------------------|---------------------|---------|-----|--------|----------|----------|
| Joshi A et al.²²     | 2018                | India   | 4 days old | female | None     | Breathing difficulty since birth |
| Rajkumar JS., et al.²³ | 2017                | India   | 28 yo | female | 30 weeks gestation, | Acute respiratory distress, decreased breath sounds |
| Makwana K., et al.²⁴ | 2017                | India   | 58   | female | None     | Fever, cough, yellowish expectoration for 1 week |
| Pradhan P., et al.²⁵ | 2020                | Nepal   | 47   | female | Typhoid fever at 17 | 1 year of abdominal pain, bloating & fullness after meals |
| Dontukurthy S., et al.²⁶ | 2020                | USA     | 46   | female | None     | Shortness of breath, food intolerance, inability to sleep supine for 1.5 years |
| Rajkumar JS., et al.²³ | 2017                | India   | 28   | female | 30 weeks pregnancy | Acute onset respiratory distress |
| Al-Zayer F., et al.²⁷ | 2019                | Saudi Arabia | 27 | female | NKCM | Respiratory distress post elective cesarean section |
| Pradhan P., et al.²⁸ | 2020                | Nepal   | 47   | Female  | Bilateral foot drop since 30 years | Abdominal distension, pain and bloating after meals |
| Vinod Kumar MS., et al.²⁹ | 2018                | India   | 5    | Male    | NKCM | Abdominal pain, vomiting, constipation, fever |
| Stamenovic D., et al.³⁰ | 2017                | Germany | 60   | female | Right leg amputation secondary to arterial embolism | Chronic assisted ventilation |
| Li XS., et al.³¹     | 2021                | China   | 24   | male    | Neurofibromatosis type 1 | Spontaneous pain and swelling of left upper abdomen |
| Manson HJ., et al.³² | 2017                | UK      | 30   | female | Dyspepsia secondary to gastric herniation | Worsening abdominal pain |
| Fujii T., et al.³³   | 2019                | Japan   | 72   | male    | Gastric cancer of antrum | Abdominal pain |
| Glasberg T., et al.³⁴ | 2017                | USA     | 1 day | female | Preterm baby, Polyhydroamnios 1 week prior to delivery | Respiratory insufficiency |
| Sharan KV., et al.³⁵ | 2021                | India   | 47   | male    | Breathlessness, left sided chest pain and fever | None |
| DiChiaccio L., et al.³⁶ | 2018                | USA     | 3 days | male    | Recurrent, chest infections | Preterm, recurrent pneumonias |
| Diagnostic criteria | Final Diagnosis | Surgical Management | Medical Management | Sepsis | Outcome (Dead/Survived) |
|---------------------|-----------------|---------------------|--------------------|--------|------------------------|
| CXR, ECHO           | Left congenital diaphragmatic eventration. Followed by right-sided eventration | Laparotomy & diaphragmatic plication, followed by right thoracotomy | (Synchronized Intermittent Positive Pressure Ventilation, followed by CPAP and PEEP) | After recurrence but on left side with GBS | Survived |
| MRI                | Diaphragmatic eventration | 4 port technique with thoracoscopic diaphragmatic plication | Mom and baby well 2 months after surgery | None | Survived |
| CXR, CT chest, PET/CT, | Diaphragmatic Eventration | Diaphragmatic plication | n/a | none | Survived |
| MRI chest           | Huge eventration of the right dome of diaphragm | Thoracoscopic diaphragmatic plication | None | None | Survived |
| ECG, CXR, Abdominal CT | Right diaphragmatic herniation | Right posterolateral thoracotomy | None | None | Survived |
| MRI chest           | Eventration of left hemidiaphragm | Left mini thoracotomy | None | None | Survived |
| CT chest, abdomen and pelvis | Left sided diaphragmatic hernia | Laprotomy with left subcostal incision | None | None | Survived |
| MRI                | Diaphragmatic eventration | Double-lined diaphragmatic plication by means of uniportal video-assisted thoracic surgery technique | None | None | Survived |
| CXR, CT chest, biopsy of diaphragm | Left hemidiaphragm hernia caused by spontaneous diaphragmatic rupture | Diaphragmatic folding | None | Just fever | Survived |
| CT chest, abdomen and pelvis | Congenital diaphragmatic hernia | Needle thoracostomy, laparotomy, total gastrectomy with Roux-en-Y reconstruction and splenectomy, sutured repair of the defect in the left hemidiaphragm | Analgesia, antiemetics | None | Survived |
| CXR, upper GI endoscopy | Left sided diaphragmatic eventration | Laparoscopic distal gastrectomy followed by diaphragmatic plication | None | None | Survived |
| CXR, ECHO, abdominal US | Pulmonary hypoplasia, hepatomegaly | - | Multiple pressors, fluid resuscitation, optimizing ventilator | None | Died |
| CXR, CT thorax | Hepatodiaphragmatic interposition | Thoracoscopic diaphragmatic plication | Antibiotics and nebulization | None | Survived |
| CXR, CT angiogram chest | Extrapulmonary versus intrapulmonary sequestration with a systemic feeding vessel from the left internal mammary artery | Video assisted thoracoscopic resection | None | None | Survived |
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CONFLICT OF INTEREST
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Written informed consent was obtained from the patient for publication of this report and any accompanying images. A copy of the consent is available for review by the Editor in Chief of the journal.

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DATA AVAILABILITY STATEMENT
Data sharing is not applicable to this article as no new data were created or analyzed in this study.

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