Clinical Curiosity Double Trouble: Diagnosis and Management of Bilateral Congenital Diaphragmatic Eventration in a Preterm Infant

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1- Introduction

Congenital diaphragmatic eventration (DE) is an uncommon disorder that may result from abnormal development where diaphragmatic muscle is replaced with fibroelastic tissue or damage to the phrenic nerve. Presentation is variable from asymptomatic incidental findings on X-ray to respiratory distress. Bilateral DE is a rarer diagnosis with a guarded prognosis. We present a preterm infant born at 32 weeks with bilateral DE who presented with persistent respiratory insufficiency, resulting in a management dilemma.

2- Case Presentation

A male infant was delivered at 32 weeks completed gestation at a level 2 rural neonatal intensive care unit (NICU) center via emergency cesarean section due to fetal heart rate abnormalities. Maternal serologies and antenatal ultrasounds were unremarkable. One dose of betamethasone was given before delivery. Apgar scores at birth were 5, 6 and 9 at 1, 5 and 10 minutes. His birth weight was 2108g. He was admitted to the NICU for Continuous Positive Airway Pressure (CPAP) respiratory support and ongoing care related

to prematurity. He appeared non-dysmorphic and appropriately developed for his gestation age with an unremarkable physical examination. A partial septic work up (PSWU) was completed at birth and antibiotics were given for 4 days then discontinued based on negative cultures and lack of infectious symptoms. An initial X-ray at birth for umbilical venous catheter (UVC) line placement showed mild bilateral hazy opacities and no consolidation within the lungs (Figure 1). He was gradually weaned from CPAP to room air by day 5 of life. He tolerated feeds well. By day 12 of life, tachycardia and tachypnea requiring respiratory support recurred. His chest X-ray showed an elevated left hemidiaphragm and worsening bilateral lower hazy opacities with an area of lucency in the lower left hemithorax (Figure 2). A PSWU was completed with a normal white cell count, low C-reactive protein (CRP) and a negative blood culture and respiratory pathogens panel (RPP).

Figure 1: AP Chest X-ray following UVC line insertion on day 1 of life showing no consolidation identified within lungs, slightly hyperinflated right lung. Lung fields show diffuse mild bilateral hazy opacities suggestive of transient tachypnea of the newborn (TTN) or respiratory distress syndrome (RDS). The left hemidiaphragm was notably elevated compared to the right.

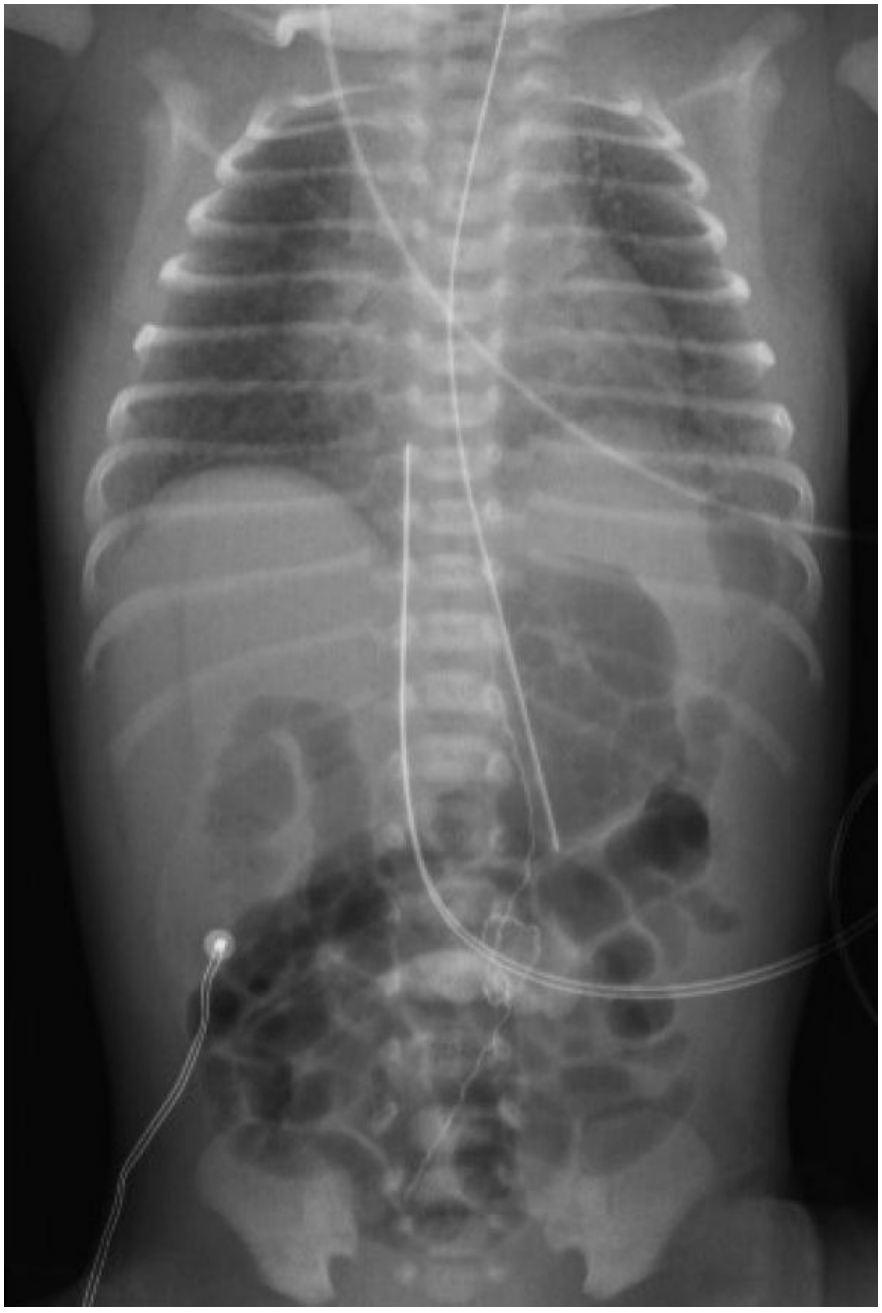
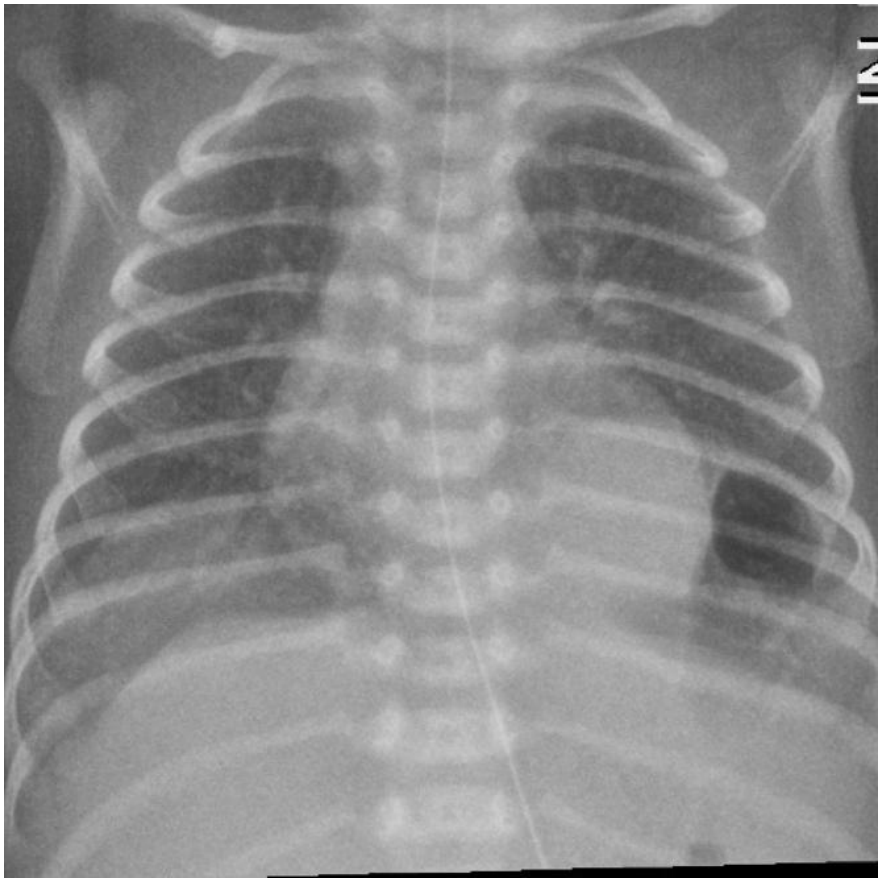
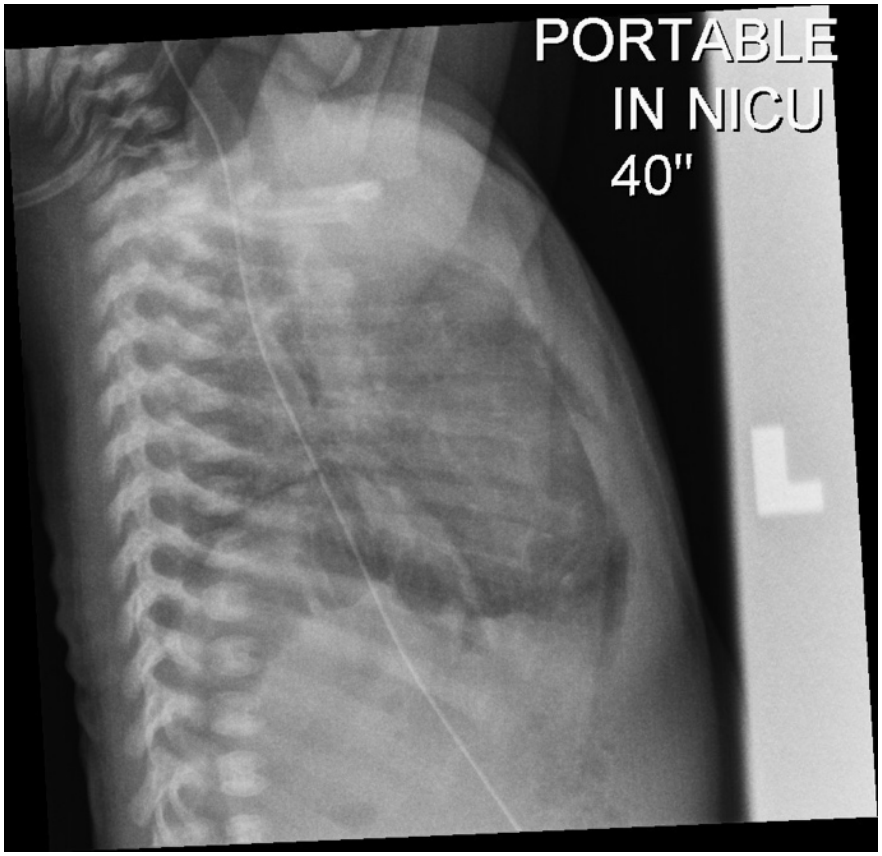


Figure 2: AP and lateral chest X-ray on day 12 of life following new onset tachypnea and tachycardia shows bilateral hazy opacities in the lower lung fields with an elevated left hemidiaphragm. A lucency is noted on the left side at the cardiac margin. Lateral X-ray shows that the lucency is bowel lying below the left diaphragm. The right lower zone opacity is either elevated right hemidiaphragm or atelectasis. The location and shape of the opacities are suggestive of diaphragmatic eventration, with abdominal contents herniating into the thorax on the left, still under the diaphragm.





2.1 -Challenge Point

An infant born at 32 weeks gestation developed respiratory distress at day 12 of life after being weaned off CPAP several days prior. A repeat PSWU was unremarkable. A chest X-ray showed bilateral lower lung field opacities with an area of lucency in the left lower lung field.

2.2 - Learner reflection

What is most likely going on with this patient? (leading and differential diagnosis)

What would you do next and why?

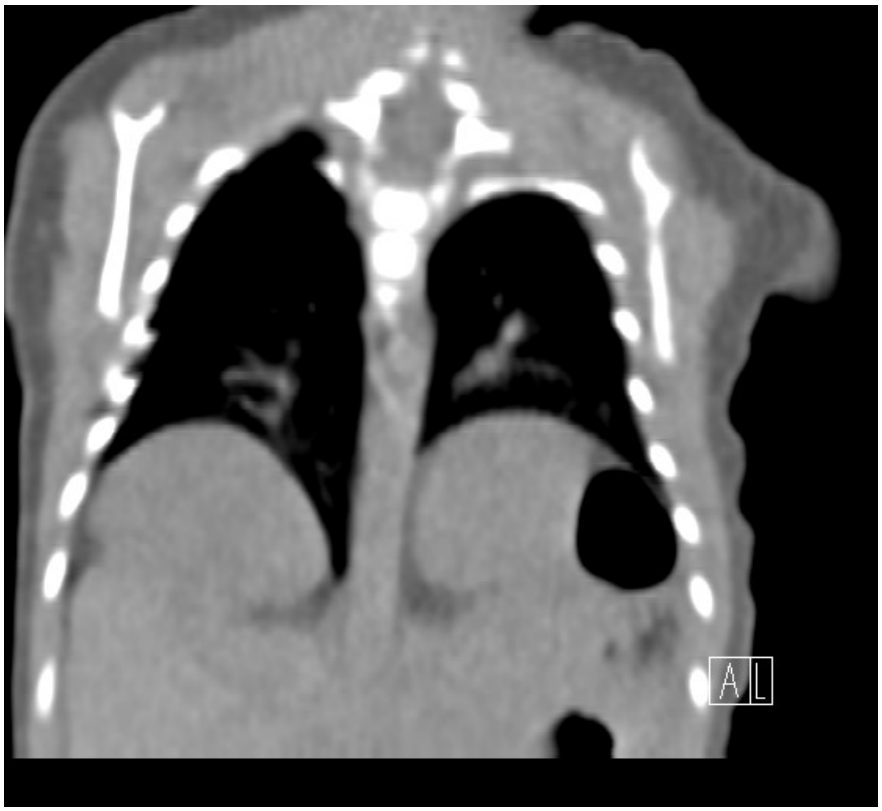
Listen to the Supporting Information Podcast 1 to hear the consortium's decision-making process.

2.3 - Case progression

Recurrent tachypnea and respiratory distress on Day 12 of life was treated with CPAP after which the tachypnea and tachycardia improved. A follow up chest X-ray 12 hours after CPAP initiation showed persistent elevation of left diaphragm with a round lucency in the lung. Two days after restarting CPAP, the team attempted to wean pressures but the baby did not tolerate this, with increased tachypnea. After another 2 weeks of support he was successfully weaned to room air. During this time, he had a normal head ultrasound and echocardiogram. A chest ultrasound was non-diagnostic for eventration while on CPAP. A chest CT (Figure 3), conducted at the peripheral hospital, was reported to show a possible left posterior diaphragm eventration or hernia and a right posterior hemithorax lesion separate from the liver.

Figure 3: Unenhanced CT chest and abdomen coronal reconstruction: Elevated hemidiaphragms bilaterally. Sigmoid colon and left kidney are in the left hemithorax, with a posterior diaphragmatic eventration or hernia.

The right kidney is in the right hemithorax, with a posterior diaphragmatic eventration or hernia.



2.4 -Challenge Point

An infant born at 32 weeks gestation presented with respiratory distress on day 12 of life, with a history of requiring CPAP support from birth to day 5 of life. A repeat PSWU was unremarkable. A chest X-ray showed persistent elevation of left hemidiaphragm and a CT showed possible left eventration and a right posterior hemithorax lesion.

2.5 - Learner reflection

What is the best modality for diagnosis of diaphragmatic eventration?

What would you do next and why?

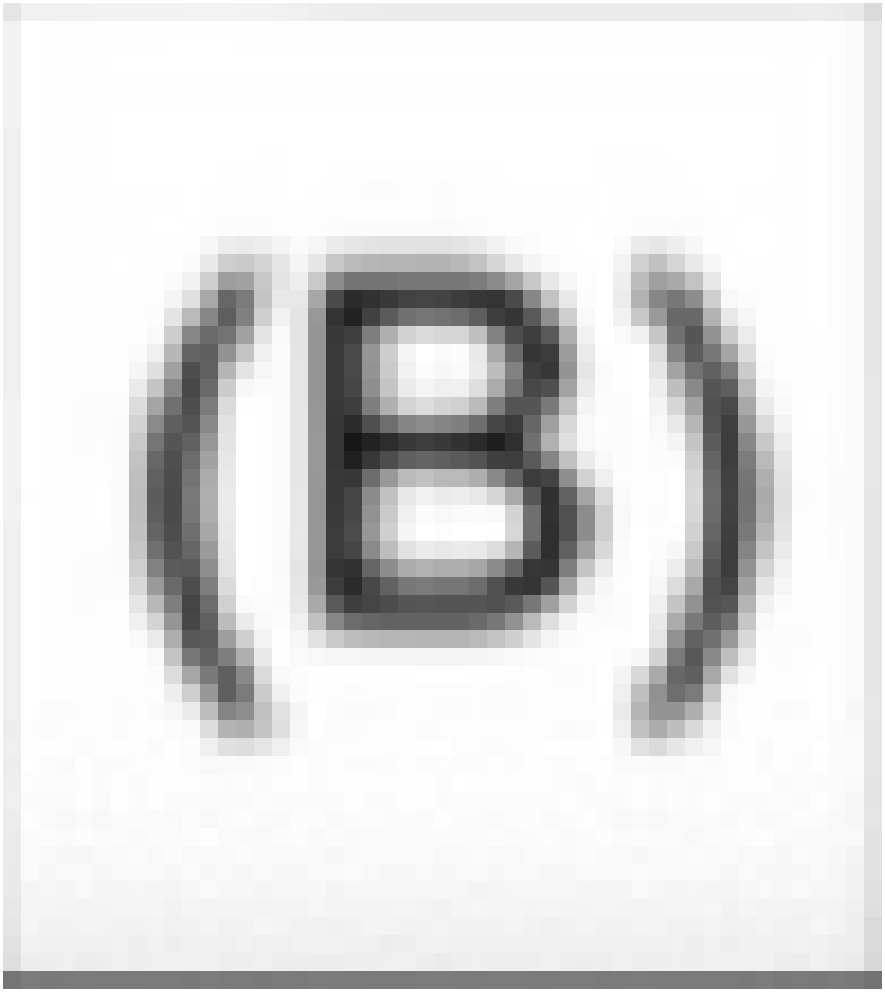
Listen to the Supporting Information Podcast 2 to hear the consortium's decision-making process.

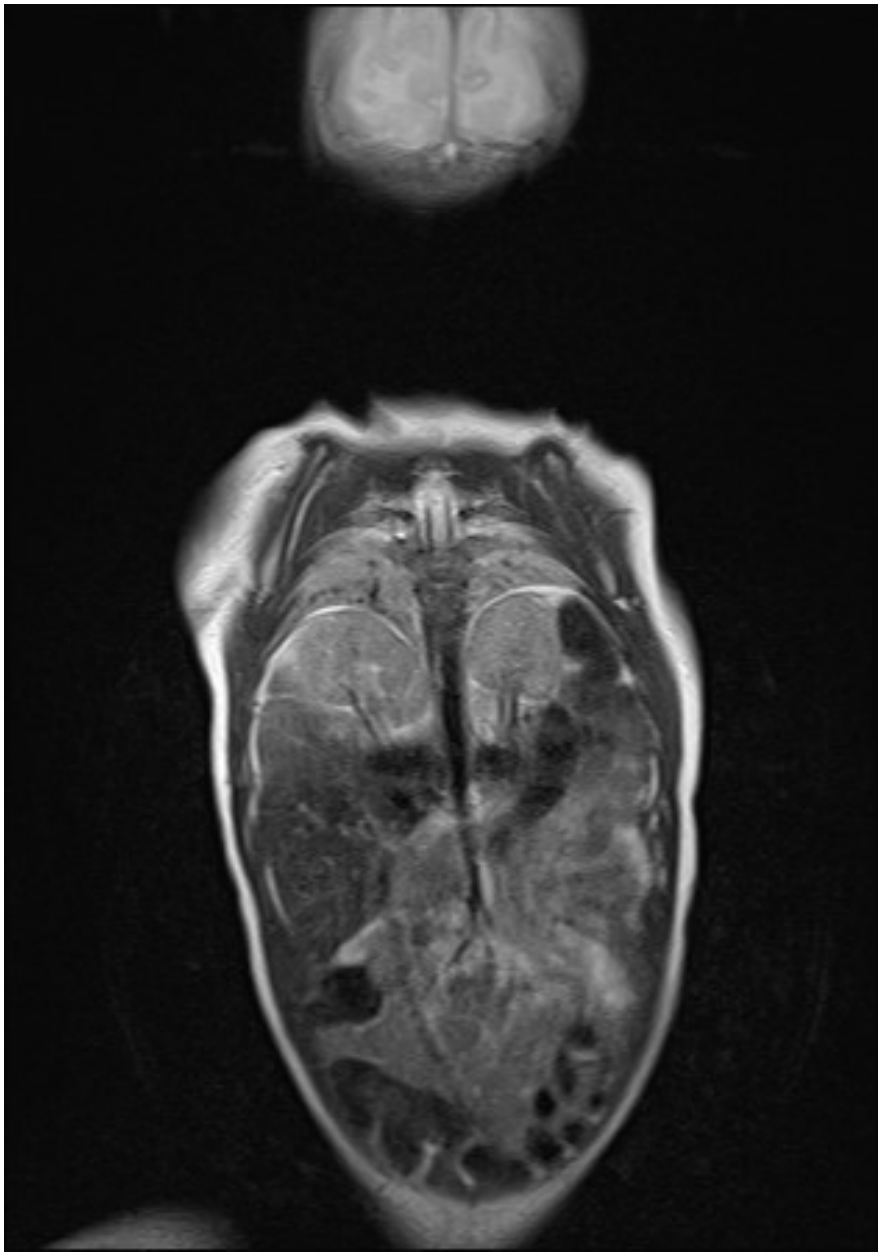
2.6 - Case progression

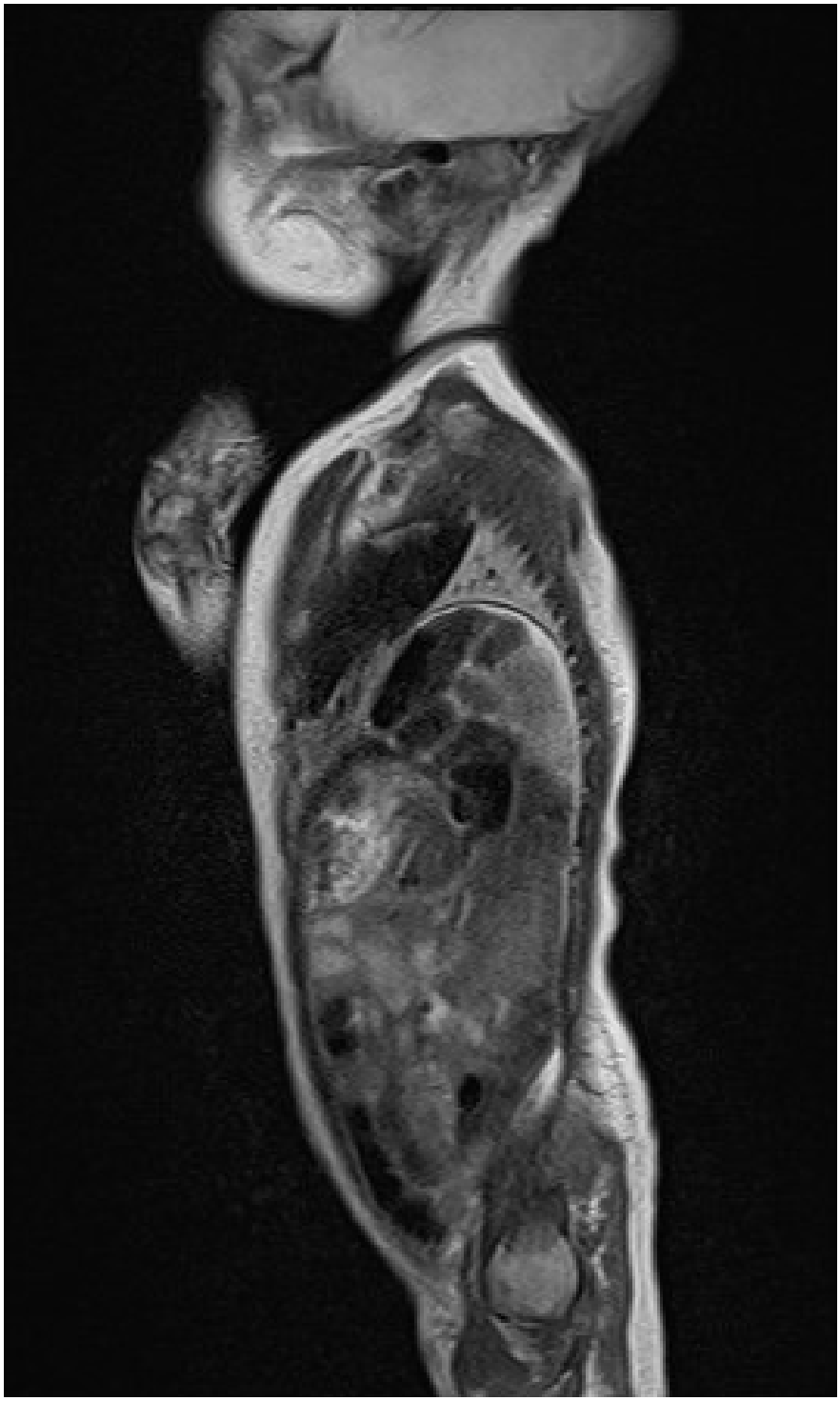
The baby was seen by General Surgery, Genetics and Pediatric Pulmonology at 1 month old (36 completed weeks corrected gestational age). At that time he had received no respiratory support for several days. An MRI of his thorax and abdomen (Figure 4) showed bilateral posterior DE with associated near-complete collapse of bilateral lower lobes of the lungs. Of note, there was elevation of his abdominal contents including kidneys, stomach, and bowel being present in his posterior thorax. The right chest mass seen on CT was confirmed to be a malpositioned right kidney. There was no evidence of a diaphragmatic hernia on MRI. Respiratory support was re-initiated based on intermittent respiratory distress, worsening respiratory acidosis on blood gas analysis and the MRI findings suggesting persistent bilateral lower lobe atelectasis. He was switched from oral to continuous nasojejunal (NJ) feeds given his higher risk of aspiration from

the combination of elevated bowel and persistent work of breathing. Other investigations included Upper Gastrointestinal Series (UGI) which ruled out malrotation. Genetic investigations were also sent including comparative genomic hybridization and Whole Exome Sequencing (WES), which were unremarkable.

Figure 4: MRI of Thorax and abdomen A) Coronal respiratory gated HASTE image of the posterior diaphragm: Elevated right and left posterior hemidiaphragms with a thin membrane above subdiaphrenic fat, characteristic of eventrations. The left kidney and sigmoid colon are elevated but lie below the diaphragmatic membrane. The right kidney is elevated but below the diaphragmatic membrane.







B) Sagittal respiratory gated HASTE image of the left hemidiaphragm: Elevated left posterior hemidiaphragm with a thin membrane and normal subphrenic fat. Left kidney and bowel are elevated into the left chest but lie below the diaphragm. The left lower lobe is collapsed.

2.7 -Challenge Point

This Ex-32-week infant is now approaching term corrected gestational age. His symptoms have now been diagnosed as bilateral eventration with persistent atelectasis of the lower lobes of the lungs bilaterally. He has ongoing intermittent respiratory support needs and is at risk of aspiration.

2.8 - Learner reflection

What intervention(s) should be considered?

What are the considerations for the timing of surgical repair and associated risks?

Listen to the Supporting Information Podcast 3 to hear the consortium's decision-making process.

2.9 - Case progression

Based on his respiratory insufficiency and risk of long-term complications secondary to persistent atelectasis and aspiration, the team decided for surgical repair of his diaphragmatic eventration. He was intubated pre-operatively and underwent bilateral plication via laparotomy and abdominal wall closure with mesh insertion at 39+6 weeks corrected gestation age. A chevron incision was made to access the abdominal cavity, revealing a left hemidiaphragm with a muscular anterior two-thirds and a thin membranous posterior third, along with the left kidney and large bowel occupying the thoraco-abdominal area underneath the intact diaphragm. The diaphragm was flattened by a series of U-stitches to plicate it, and the kidney was repositioned down further in the abdomen. The right side revealed a similar issue with the right kidney and liver in the thoraco-abdomen below intact diaphragm; both were reduced to the proper abdominal cavity. Both sides of the diaphragm were successfully plicated, resulting in a stable, flat position. Loss of abdominal domain was noted after repositioning the liver, kidneys, and bowel. A Gore-Tex DualMesh was used for abdominal wall reconstruction to prevent abdominal compartment syndrome. The infant tolerated the procedure well and was sent back to the NICU intubated with minimal blood loss. His postoperative course was complicated by suspected Necrotizing enterocolitis (NEC) on Post-operative Day (POD) 6, which presented as abdominal distention and suspected pneumatosis on X-ray. During that time, he received parenteral nutrition and treated with antibiotics for 7 days. His abdominal distention improved and he successfully transitioned to full enteral feeds without difficulty following treatment.

He was extubated to non-invasive respiratory support on POD 10 and gradually weaned to room air by POD 18. He was discharged from the NICU on room air and tolerating full oral feeds at 73 days of age. Since being discharged he presented once to an emergency department for bloody stools, suspected to be a cow's milk protein allergy (CMPA) after his breastfeeding mother introduced milk products into her diet. This issue was resolved by switching to a hydrolyzed formula. He continues to grow well and meet his developmental milestones.

3 - Discussion

The diaphragm is the primary muscle used during inspiration. It acts as a barrier between the thoracic and abdominal cavity and is innervated by the phrenic nerve. DE is a condition where there is an abnormal elevation of the diaphragm with maintained continuity and attachments to the costal margin. The diaphragm can be partially or fully replaced with fibroelastic tissue. (1, 2) DE can cause respiratory issues due to reduced lung volume and impaired breathing mechanics. This could lead to atelectasis, pneumonia as well as aspiration and respiratory insufficiency requiring support. (2) Abdominal contents can be displaced into the thoracic cavity, further compromising breathing by compressing the lungs. Complications of eventration can also occur later in life with case reports of volvulus and other issues with malpositioned abdominal contents being the highest risk of morbidity.(3, 4) In infants, DE can be congenital due to abnormal development or acquired due to damage to the phrenic nerve. (5) In congenital diaphragmatic eventration, there is inadequate development of the diaphragmatic muscle or absence of the phrenic nerve. In acquired diaphragmatic eventration, the most common cause is an injury to the phrenic nerve leading to diaphragmatic paralysis. Damage to the phrenic nerve can be caused by either trauma at the time of delivery or thoracic surgery.(5) In our case, there was no history or exam findings suggestive of birth trauma at the time of delivery suggesting a congenital etiology is the more likely cause. Congenital DE constitutes only 5% of all diaphragmatic defects

including congenital diaphragmatic hernias (CDH) and is usually unilateral with the left side more commonly affected than the right. (6) It also has a higher male predominance. (7) The incidence and prevalence of congenital DE in infants is unknown due to some lesions being clinically silent. Some reports estimate this to be as low as 1 in 10,000 but the overall incidence is likely higher due to the number of unreported cases. (8) DE can also be associated with many disorders and infections. These include Spondylocostal dysostosis, Kabuki syndrome, Beckwith-Wiedemann syndrome, Poland syndrome, chromosomal abnormalities (trisomy 18), pulmonary hypoplasia, spinal muscular atrophy, malrotation, and congenital heart disease. Infectious associations include fetal rubella and cytomegalovirus infections.(9-12) In our case, the infant appeared non-dysmorphic and there were no associated findings on any investigation to suggest an associated syndrome or disorder.

The diagnosis of DE is usually made postnatally and can be delayed due to the absence of symptoms.(13, 14). Chest X-ray can suggest the diagnosis by showing the elevation of the hemi-diaphragm. Still, it can be challenging to differentiate between DE and CDH due to the similarities in imaging findings. Ultrasound can be used to confirm the findings by demonstrating minimal or paradoxical diaphragmatic movement although this is operator dependent. In our case, the final diagnosis was made using MRI which was able to identify diaphragmatic eventration on both sides of the diaphragm. With regards to management, the choice between conservative management or surgical repair depends on several factors including the presence or absence of symptoms, adequacy of nutritional intake and growth, the need for respiratory support and the presence of complications such as volvulus or recurrent pneumonia. Surgical management is recommended for symptomatic cases, particularly for bilateral eventration although there is no standardized approach regarding the timing of surgical repair. Stabilization on non-invasive ventilation before surgery is suggested, although with limited literature support. Earlier intervention may also improve growth and prevent lung injury (2) In this case, surgery was done closer to term as the infant continued to be symptomatic with ongoing tachypnea and bibasilar atelectasis despite interval growth. Complications after abdominal plication of a diaphragmatic eventration commonly include atelectasis and rarely, abdominal compartment syndrome and splenic injury after mobilization of the left upper quadrant abdominal structures. (15, 16) In our case, he developed findings suggestive of NEC which was managed with bowel rest and antibiotics. Following discharge, he will require routine monitoring and follow-up with a multidisciplinary team including a Pediatrician, Pediatric Pulmonologist and a Pediatric Surgeon as there is a possibility of relapse of eventration. Overall, the prognosis of infants who undergo surgical plication for DE is good. (14) The infant is currently doing well, not requiring any respiratory support and growing appropriately. This case is an excellent example of a multidisciplinary collaboration between Neonatology, Pediatric Pulmonology, Radiology, and Pediatric Surgery teams toward the management of a rare condition.

4- Conclusion

Bilateral DE has a variable presentation in the neonatal period, making diagnosis challenging. It should be considered in any infant with respiratory distress and lower lobe opacities on X-ray. MRI is the best modality to confirm the diagnosis. The timing of repair is not well defined but dependent on the severity of symptoms as well as the size of the baby. Surgical repair should be considered early in symptomatic cases to prevent complications and sequelae of prolonged respiratory insufficiency. Risks of early intervention must be weighed against the potential benefits of delaying surgery to allow the baby time to grow. Bilateral diaphragmatic eventration can present as an isolated finding or in association with other anomalies and conditions which should be investigated. Follow-up of patients with diaphragmatic eventration is necessary for possible complications or recurrence of eventration.

Author contributions

Case report conception and design: Abdulaziz Abul, Anne Hicks, Matthew Hicks

Assembly for case: All authors

Manuscript preparation: All authors

All authors reviewed the results and approved the final version of the manuscript.

Conflict of interest statement

The authors declare no conflict of interest.

Data availability statement:

Data sharing is not applicable to this article as no data sets were generated or analyzed during the current study.

Ethics statement:

Case presentation and images used with written consent from patient's family. Project was reviewed by the Health Research Ethics Board (REB) and was exempt from review. Ethical approval was not required.

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