CASE REPORT

Left-sided Congenital Diaphragmatic Hernia Coexisting with Anorectal Malformation and Recto-vesical Fistula

Ahmadu MS[1], Farouk AG[2], Ahidjo A[1], Tahir A[1]

ABSTRACT

Background: Although congenital diaphragmatic hernia (CDH) is a common congenital anomaly with incidence ranging from 1 in 2,500 to 1 in 5,000 live births, its coexistence with anorectal malformation (ARM), and recto-vesical fistula is rare. The aetiopathogenesis of CDH remains unclear, and its coexistence with other congenital anomalies suggests that CDH occurs as a result of multiple, complex, and varied developmental abnormalities. The most common associations of CDH are with cardiac, gastrointestinal tract, genitourinary tract, central nervous system, as well as musculoskeletal anomalies. This report described a case of congenital diaphragmatic hernia associated with anorectal malformation (ARM) and recto-vesical fistula (RVF) in a 2-day-old neonate.

Keywords: Congenital Diaphragmatic Hernia, Anorectal Malformation, Recto-vesical Fistula.

[1]Department of Radiology, University of Maiduguri Teaching Hospital, Maiduguri, Nigeria
[2]Department of Paediatrics, University of Maiduguri Teaching Hospital, Maiduguri, Nigeria

Corresponding Author
Dr. Ahmadu Muhammad Sani
Department of Radiology, University of Maiduguri Teaching Hospital
PMB 1414, Maiduguri, Borno State, Nigeria.
Phone: +234 705 588 2425
Email: drahmadums@unimaid.edu.ng

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Introduction

Congenital diaphragmatic hernia (CDH) is a developmental malformation characterized by a defect in the posterolateral aspect of the diaphragm, the foramen of Bochdalek, through which the abdominal viscera migrate into the chest during fetal life.[1] This defect which permits the herniation may occur on the right, left, both sides, anterior-medial or through oesophageal hiatus of the diaphragm.[2] The incidence of CDH ranges from 1 in 2,500–5,000 live births in the United States. The hernia is twice more common in males than females. Approximately 80% of posterolateral diaphragmatic hernias occur on the left side and 20% on the right side. Bilateral CDH is rare. The size of the defect varies from small (2 or 3 cm) to very large, involving most of the hemidiaphragm.[1-3] Presence of associated anomalies has been reported to be about 40% in most cases. The commonest abnormalities associated with CDH are cardiovascular anomalies, skeletal, central nervous system, genitourinary,

gastrointestinal, craniofacial, and chromosomal and syndromic defects.[1, 2, 4]

The aetiology of CDH is still unknown, but certain drugs such as thalidomide and nitrofuratoin have been implicated. Although CDH is generally considered to be sporadic, some familial cases have been reported.[4, 5]

Patients with CDH usually present in the early neonatal period as cases with varying degrees of respiratory distress, tachypnoea, grunting respiration, and cyanosis. But in some cases, these presentations may be delayed.[5] Presence of associated anomalies has consistently been associated with decreased survival.[6] In the evaluation of patients with CDH with or without associated congenital anomalies, radiology plays a crucial role in diagnosis and management.[3]

This report presents a case of left-sided congenital diaphragmatic hernia in a 2-day-old neonate with associated anorectal malformation and recto-vesical fistula.

Case Presentation: B.Y.G. was a 2-day-old male baby who was brought to the special care baby unit (SCBU) of the University of Maiduguri Teaching Hospital by his mother with history of difficulty in breathing, progressive abdominal distension, passage of greenish urine, and on/off bluish discolouration of the lips and skin since birth. The mother also complained of the child refusing to breastfeed and crying excessively. The child was delivered to a 28-year-old para 5 mother (all alive and well). The mother had not attended antenatal clinic (ANC) throughout the pregnancy and was delivered by a traditional birth attendant at home. No history of consumption of drugs or traditional concoctions during the pregnancy. However, the mother had noticed that her abdomen was unusually larger, while she was carrying this pregnancy, than the previous pregnancies she had. The index pregnancy was carried to term. There was no family history of congenital anomaly. The mother was a full-time housewife while the father is a petty trader.

On general examination, the neonate was acutely ill-looking evident by respiratory distress and crying, afebrile, not pale, not jaundiced, centrally cyanosed with no dysmorphic facie. He weighted 3kg and had occipitofrontal circumference (OFC) of 37cm.

Respiratory system examination revealed a dyspnoeic child with subcostal recession more severe on the left. Respiratory rate was 60 cycles/minute. Bronchovesicular breath sounds were heard on the right hemithorax while bowel sound was picked on the left hemithorax. Apex beat could not be palpated but there were normal 1st and 2nd heart sounds and no other abnormal auscultatory findings.

The abdomen was uniformly distended, non-tender.

There was no evidence of anal opening on examination of the perineum. Greenish fluid (meconium) was noted to be oozing per-urethra.

The full blood count, electrolytes, urea, and creatinine were all within normal ranges.

Plain radiograph of the chest, anteroposterior view (Fig. 1) showed multiple lucencies and gas filled bowel loop extending through a diaphragmatic defect from the abdomen into the left hemi-thorax with associated shift of the mediastinum to the right side.

Barium meal and follow-through study (Figs. 2 and 3) showed the entire opacified stomach and part of the small intestine within the left hemithorax passing through a posterolateral diaphragmatic defect.

Transfontanelle and abdominal ultrasonography were normal.

A provisional diagnosis of congenital diaphragmatic (Bochdalek) hernia with

suspected anorectal malformation (ARM) and recto-vesical fistula (RVF) was made based on the plain chest radiographic findings, barium meal/follow-through, and abdominal distension, absence of anal opening and passage of meconium per-urethra.

The patient was rushed into the theatre for an emergency surgery in order to reduce the herniated stomach and part of the small intestine from the left hemithorax and subsequent repair of the diaphragmatic defect. A transverse colostomy was also done to divert faeces.

Patient's respiratory symptoms improved after the surgery and a distal colostography (Figs. 4 and 5) was requested 10 days post-operative, which showed the distal segment of the bowel ending as a pouch at the recto-sigmoid junction above the pubococcygeal line (demarcating the puborectalis sling of the levator ani muscle) approximately 5cm from the anal dimple (metallic marker).

The urinary bladder was noted to be filled with contrast via a fistulous communication and evidence of contrast was also noted in a micturating phase within the urethra (Fig. 5). The patient was also noted to have passed barium mixed with urine through the urethra while micturating in the course of the procedure.

The diagnosis of anorectal malformation (high type) associated with recto-vesical fistula was further confirmed by the distal colostography.

The patient made a remarkable improvement in his symptoms and was discharged home two weeks post-operative.

The repair of anorectal malformation and recto-vesical fistula was planned for when the patient reaches nine months of age.

Follow-up at Paediatric Surgical Outpatient Clinic showed the patient to be doing well.

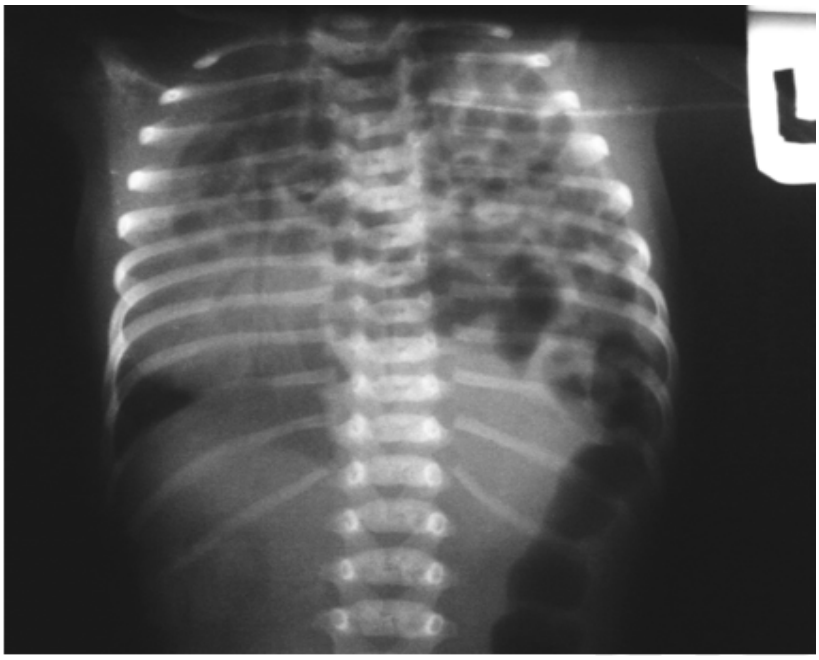

Figure 1: Anteroposterior chest radiograph showing multiple lucencies and gas filled bowel loop extending through a diaphragmatic defect from the abdomen into the left hemi-thorax with associated shift of the mediastinum (white arrow) to the right side

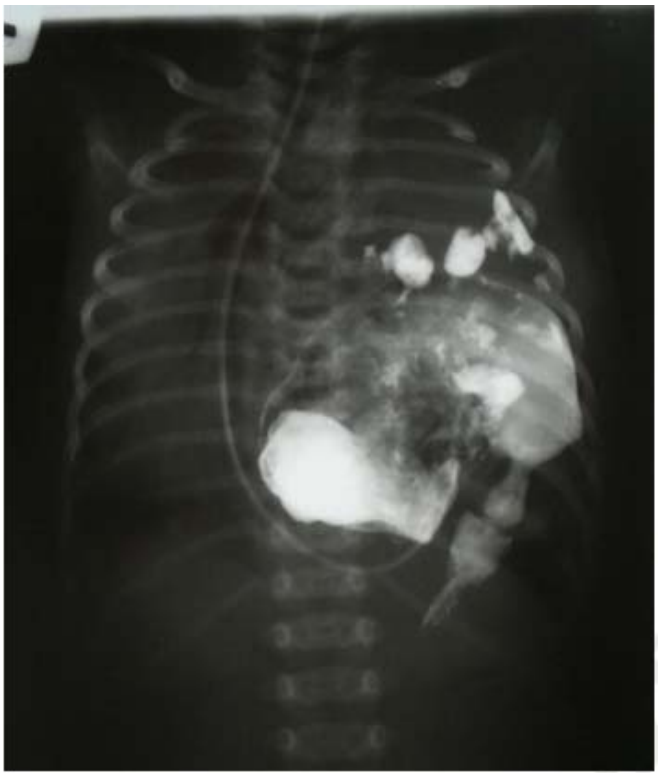

Figure 2: Barium meal showing the entire opacified stomach and part of the small intestine within the left hemithorax. Note the position of the duodenal cap (white arrow) laterally

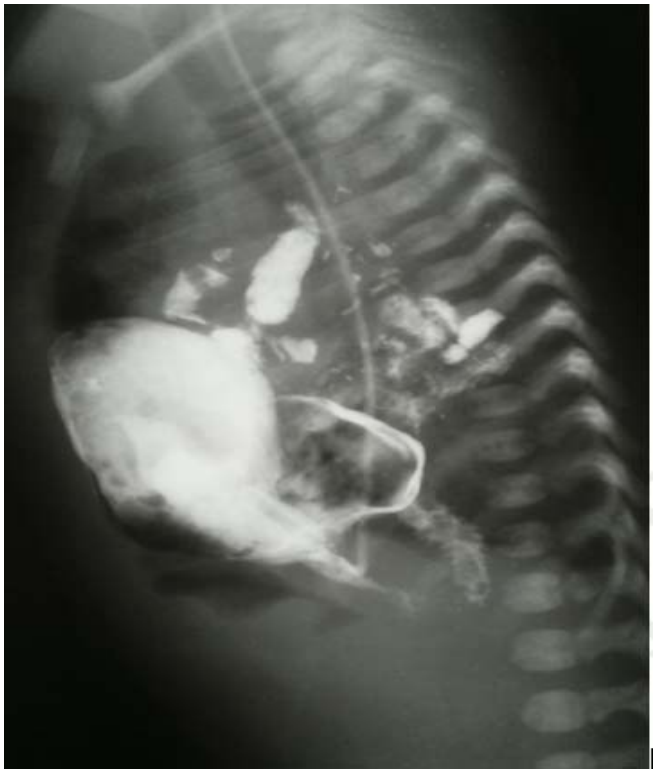

Figure 3: Barium meal/follow-through, lateral projection, showing the opacified stomach and part of the small intestine within the left hemithorax passing through a posterior diaphragmatic defect (white arrow)

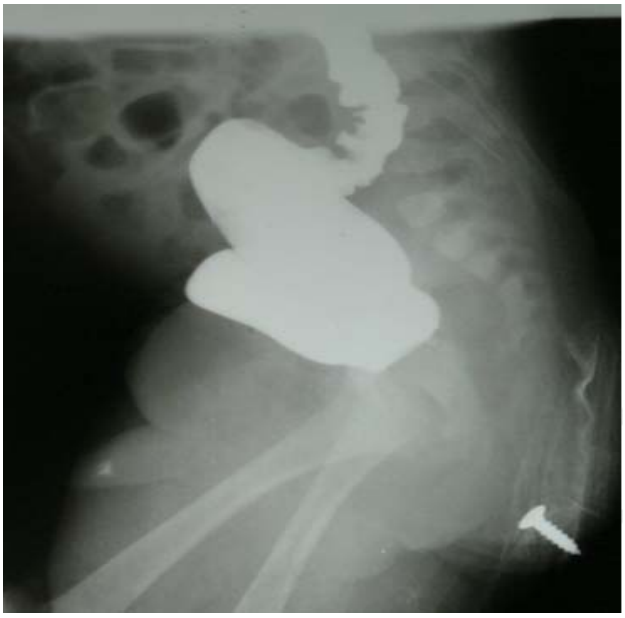

Figure 4: Lateral projection, distal colostogram, showing the distal segment of the bowel ending as a pouch at the recto-sigmoid junction approximately 5cm from the anal dimple (metallic marker)

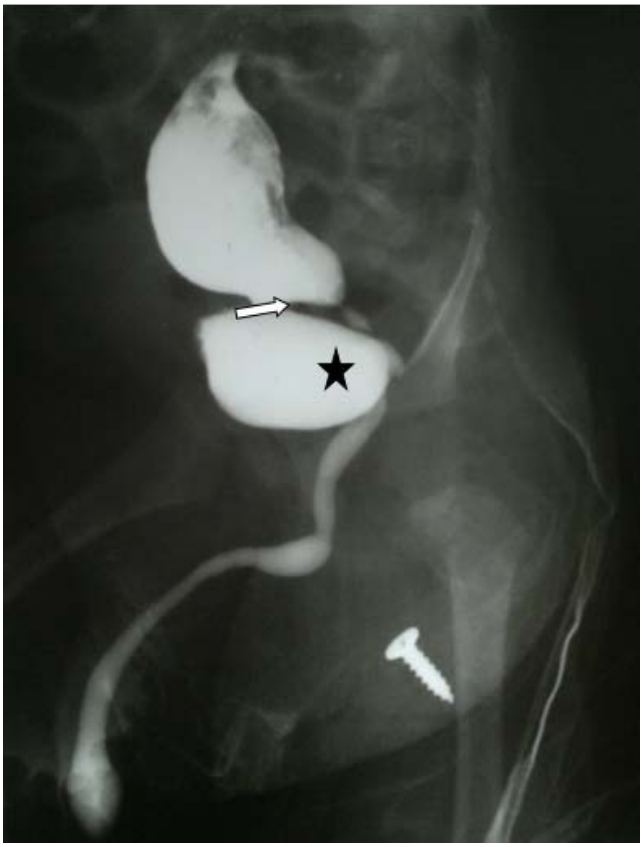

Figure 5: Colostogram, oblique projection, showing the urinary bladder (black star) filled with contrast (barium) via a fistulous communication (white arrow) and evidence of contrast was also noted in a micturating phase within the urethra

Discussion

Congenital diaphragmatic hernia (CDH) is classified into three types: the posterolateral (Bochdalek) which occurs through the posterior pleuroperitoneal communication of Bochdalek, the Morgagni hernia which occurs through the sternocostal hiatus of the diaphragm, and the hiatus hernia which occurs through the oesophageal hiatus.[1,3] Among all these types, Bochdalek is the commonest of the CDH, occurring in approximately 85% of cases, and is also more common on the left.[5] The case presented in this report had a left Bochdalek CDH. The pathogenesis of CDH remains unclear, and its association with various concurrent congenital anomalies and variable pathological patterns and clinical presentation suggests that CDH is a result of multiple, complex developmental abnormalities.[7] It is estimated that associated congenital anomalies present in 30 to 40% of the cases of CDH of which the most common are those of cardiac, urinary tract, gastrointestinal tract and central nervous system defects, as well as skeletal and neural tube defects.[1,2,4]

Although the cause of the CDH in the patient presented could not be found as obtained in other studies, he was found to have associated congenital anomalies involving the gastrointestinal tract

and the genito-urinary system in form of anorectal malformation and recto-vesical fistula. The clinical presentation of patients with CDH depends on the size of the defect and associated anomalies.[1-3]

The patients most often present early in the neonatal period with respiratory distress, cyanosis, and scaphoid abdomen if the defect is large.[8] The case presented in this report had respiratory distress as well as cyanosis, but did not present with scaphoid abdomen possibly because of the associated ARM which led to the distension of the abdomen.

Congenital diaphragmatic hernia is associated with wide range of complications, apart from it being associated with other congenital anomalies. These complications include pulmonary hypoplasia, gastric volvulus, and rotational abnormalities in the gut.[9] The patient presented in this case had left pulmonary hypoplasia, pulmonary hypertension, and gastric volvulus. In the evaluation of the patient with CDH with or without associated congenital anomalies, radiology plays a crucial role.[3,9] The timing of these imaging could be in the prenatal or postnatal period.[9] Prenatal ultrasonography as early as the second trimester in pregnant women carrying a fetus with CDH relies on detecting abdominal organs in the chest of the fetus and mediastinal shift. But the sonographic hallmark is a fluid filled mass behind the left ventricle representing an intrathoracic bowel loop or spleen. Other sonographic features include polyhydramnios, a small abdominal circumference and the detection of stomach in the fetal chest.[9] These sonographic features were not seen in the case presented because the mother did not attend antenatal clinic during the pregnancy. However, she admitted to having observed an unusual increase in her abdomen during the index pregnancy which may represent polyhydramnios. Other prenatal radiological investigations include fetal echocardiogram to detect associated cardiac anomalies. Ultrafast fetal magnetic resonance imaging may also be done.[4]

In the postnatal evaluation of CDH, plain chest x-ray is valuable.8 In the early neonatal period, before air enters the bowel, the fluid-filled diaphragmatic hernia produces a large opaque hemithorax. After air is swallowed and is passed through the gastrointestinal tract, loops of bowel in the hernia become gas-filled.[2-4] The plain chest x-ray of the patient presented in

this case showed multiple lucencies and gas filled bowel loop extending through a diaphragmatic defect from the abdomen into the left hemithorax with associated shift of the mediastinum and airway to the right side confirming the presence of a herniated gastrointestinal content into the left hemithorax.

Other imaging modalities that could be used to evaluate CDH and its associated congenital anomalies during the postnatal period include barium studies, computed tomography (CT), and magnetic resonance imaging (MRI) scan.[1-4] The barium meal/follow-through and distal colostography done in the case presented were pathognomonic of CDH with associated ARM and RVF. The role of radiology in demonstrating the type of ARM in association with CDH is also important.10 The case presented had a high type of ARM.

Management of patients with isolated CDH is usually surgical and depending on the associated congenital anomalies, other management therapies may be employed. Classical timing of the surgical management of CDH depends on the patient's condition.8 Often, the repair is delayed until the infant has stabilized.[4] If the infant's condition deteriorates, extra-corporal membrane oxygenation (ECMO) can be instituted before the surgery, and in some cases, repair can be done while the patient is on ECMO.[2] ECMO is directed towards optimizing oxygenation, especially in patients with pulmonary hypoplasia. The patient presented in this case report had left pulmonary hypoplasia but he was stable enough to undergo the repair for the CDH.

Conclusion

Congenital diaphragmatic hernia (CDH) is a common congenital anomaly occurring 1 in 2,500 to 1 in 5,000 live births. However, its coexistence with other congenital anomalies like anorectal malformation (ARM), and recto-vesical fistula is rare as presented in this case report.

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