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CASE REPORT

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Right-sided congenital diaphragmatic hernia in a term neonate seen at a tertiary health facility in Southern Nigeria: A case report

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Abstract: Congenital diaphragmatic hernia (CDH) results from abnormal development of the diaphragm in utero. Symptoms such as tachypnoea, respiratory distress and cyanosis may be noticed after birth especially when the diaphragmatic defect is large. A high index of suspicion is required for early diagnosis.

Keywords: Right-sided congenital diaphragmatic hernia, neonate, respiratory distress.

Resume: La hernie diaphragmatique congénitale (HDC) résulte d'un développement anormal du diaphragme in utero. Des symptômes tels que la tachypnée, la détresse respiratoire et la cyanose peuvent être observés après la naissance, en particulier lorsque le défect diaphragmatique est important. Le diagnostic précoce repose sur une forte suspicion Clinique.

Mots clés: Hernie diaphragmatique congénitale droite, nouveau-né, détresse respiratoire.

Introduction

Congenital diaphragmatic hernia (CDH) is a defect in the diaphragm resulting in the communication between the thoracic and abdominal cavities with or without the protrusion of the contents of the abdominal cavity into the thorax of the foetus.[1,2] Though associated with anomalies in other systems, its aetiology is unclear.[3] It is a cause of respiratory distress in the newborn.[4] The incidence rate is 1 - 4/ 10,000 live births.[5] The most common lesion is through a posterolateral herniation via the foramen of Bochdalek in 70 – 75% of cases and the incidence is commoner on the left side (85%) compared to the right (13% to 17%).[5-10] Morbidity and mortality are high in the presence of associated lung hypoplasia, pulmonary hypertension and other life-threatening congenital anomalies.[11,12] Right sided lesions are associated with a higher morbidity and mortality compared to lesions on the left side of the diaphragm.[13] This report aims to highlight the importance of having a high index of suspicion when evaluating neonates with tachypnoea and respiratory distress from birth. It is also important to document the challenges encountered in the management of neonates with rare abnormalities, such as discharge against medical advise (DAMA) because many of such neonates are denied a chance to proper medical care.

Case presentation

The patient was a 6-day-old neonate who was brought to the facility on account of fast breathing from birth. He was delivered at a peripheral hospital at term via emergency caesarean section on account of prolonged labour of 48 hours. He did not cry well at birth and the referring facility did not document what his APGAR scores were. There was an associated history of prolonged rupture of membranes. No peripartum pyrexia.
Pregnancy was spontaneously conceived but there was a history of failed medical termination of pregnancy within the first trimester. Although the parents of the child were unmarried but co-habiting, the mother did not disclose to us why she initially attempted to terminate the pregnancy. Pregnancy was then booked for antenatal care at a private hospital at 22 weeks gestational age. Booking parameters were essentially normal. An obstetric ultrasound scan detected no abnormalities. The identified teratogens obtained from maternal history were the consumption of alcohol and alcohol-based herbal mixtures throughout the pregnancy.

Examination of the newborn at presentation revealed a conscious, dyspnoeic child with flaring of the alae nasi, intercostal and subcostal recessions, cyanosed with SPO_2 of 49% on intranasal oxygen at 2L/ minute, afebrile with a temperature of 37^0C, not pale, anicteric.
The chest wall was symmetrical. Respiratory rate was 88 cycles per minute and the breadth sounds were reduced in the right lung fields.
The heart rate was 164 beats per minute with normoactive praecordium. Apex beat was heard at the fourth intercostal space, lateral to the mid-clavicular line on the left hemithorax. First and second heart sounds with Grade 3/6 systolic murmur maximal at the left lower sternal border were heard.

The abdomen was full and moved with respiration. Liver and spleen were not palpable per abdomen and bowel sounds were normoactive. Anus was patent.
The child had ambiguous genitalia with the presence of a short penile shaft, 11mm stretched length. Glans penis and labia majora were present. The urethral opening was located on the ventral surface, towards the base of the phallus.

The neonate was conscious with patent and normotensive anterior fontanelle. Occipitofrontal circumference was 36cm. No significant head lag. Good primitive reflexes were present, with normal tone globally.
Anteroposterior chest radiograph (Fig 1) showed cystic structures in the right hemithorax. The mediastinum was shifted to the left and the abdomen was relatively devoid of gas. A diagnosis of right-sided congenital diaphragmatic hernia was made, also considering cystic adenomatoid malformation as a differential.
A confirmatory upper gastrointestinal contrast study (Fig 2) further done, showed multiple ill-defined soft tissue opacities seen in the region of the right lung field with its components having bowel signature.
The child was admitted to the neonatal intensive care unit and was placed on intranasal oxygen, intravenous broad-spectrum antibiotics, intravenous fluids and oral sildenafil. Serum electrolytes, urea and creatinine were essentially normal.

The result of abdominopelvic ultrasound scan to visualize the reproductive organs was inconclusive. A multidisciplinary approach to the management of this neonate was to be commenced with the cardiothoracic and paediatric surgeons invited to review and co-manage. However, the parents left against medical advice on account of financial constraints, despite several counselling sessions.

Fig 1: Anteroposterior chest radiograph

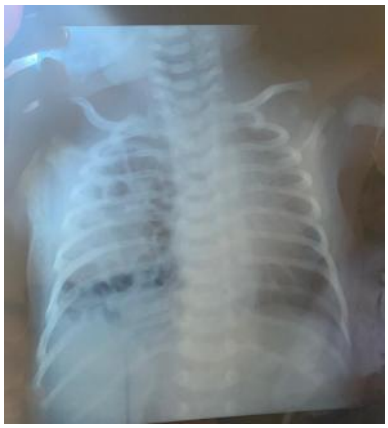
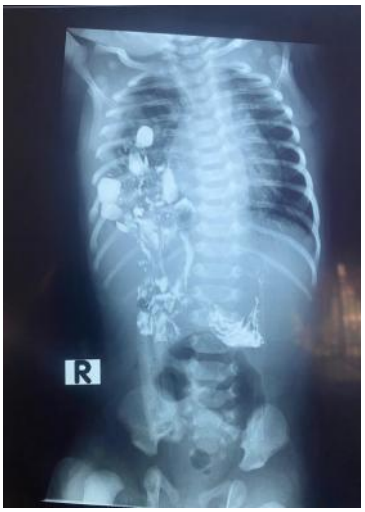


Fig 2: Barium swallow and follow through



Discussion

The development of the foetal diaphragm is usually completed by the 12th week of gestation and any interruption in its formation can result in congenital diaphragmatic hernia.[5,14] The herniation of the abdominal contents into the chest region will cause the developing lung on the affected side to be hypoplastic and with aberrant pulmonary vasculature.[5,14] At birth, patients with large congenital diaphragmatic hernia present with respiratory distress, cyanosis, decreased breath sounds on the affected side and displaced heart sounds.[5] These findings were consistent with those of our index case.
Prenatal exposure to alcohol during pregnancy could affect retinol metabolism, which plays a role in the formation of the diaphragm, leading to its abnormal development.[14,15] Alcohol intake during pregnancy and an attempt at terminating the pregnancy may have increased the risk of abnormal development of the diaphragm of our index patient.

The posterolateral defect (Bochdalek hernia) occurs in majority of cases and the left-sided type is the most

common.[14] The condition is higher among males[16] and the presence of congenital diaphragmatic hernia may be associated with other congenital abnormalies.[14] An early sign seen in patients with congenital diaphragmatic hernia is respiratory distress with tachypnoea, cyanosis and use of accessory muscles for respiration.[17] The gender of the index patient was not yet assigned because the child had ambiguous genitalia. Although a cardiac murmur was heard while auscultating the child's praecordium, an echocardiography could not be done to confirm the co-existence of a cardiac abnormality due to parental financial constraints.

Diagnosis of the condition can be made in the antenatal period during an anomaly scan usually performed by the 18th week of pregnancy.[5] Our patient's mother did not undergo an anomaly scan during pregnancy and this could have assisted in the early diagnosis of the condition and management of the patient post-delivery. A chest radiograph with upper gastrointestinal contrast study done revealed herniation of the gut into the right hemithorax with shifting of the mediastinum, confirming the diagnosis of right-sided congenital diaphragmatic hernia.

The management of CDH entails ventilatory support, use of pulmonary vasodilators in cases of poor oxygenation and pulmonary hypertension, and surgical repair of the defect on the diaphragm.[9,17] The prognosis of patients with CDH depends on the size of the diaphragmatic defect, the degree of lung hypoplasia and pulmonary hypertension. Also, gestational age at birth and the presence of other congenital anomalies are important parameters.[17]

Discharge against medical advice, is a recurring narrative in many neonatal units in Sub-Saharan Africa, including Nigeria.[18,19] This is especially so, in neonates with conditions that may require a wide variety of investigations and surgical interventions. Likewise, unmarried mothers without adequate social and financial support are particularly vulnerable.[20] This was the case in our index patient. DAMA denies many of such children a chance to access medical care and consequent improved chances for survival and optimal growth.

Conclusion

The right-sided diaphragmatic hernias are rare, but does occur, and clinicians must be on the look-out for them. Early diagnosis and prompt intervention results in better outcome. Health insurance coverage from the government would help ensure that affected children have an increased chance at receiving needed intervention and care, for better prognosis.

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