

Congenital Diaphragmatic Hernia

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Abstract

Congenital diaphragmatic hernia (CDH) refers to a diaphragmatic defect that allows herniation of some abdominal viscera into the thoracic cavity. We present a male neonate with CDH and bilateral cryptorchidism managed in our facility with limited expertise for cardiothoracic surgery. We highlight some of the challenges in the management.

Key words: Congenital diaphragmatic hernia, respiratory distress, pulmonary hypoplasia, Mediastinal shift

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Introduction

The diaphragm is a dome shaped tissue that separates the thoracic cavity from the abdominal cavity and serves important functions in respiration. Congenital diaphragmatic hernia [CDH] refers to the presence of a defect in the diaphragm that allows herniation of some abdominal viscera into the thoracic cavity causing respiratory distress at birth with or without pulmonary hypoplasia/hypertension¹. CDH can be classified into three namely: the posterolateral defect [Bochdalek hernia] seen in about 70%, the anterior defect [Morgagni hernia] in 23% and the central defect [hiatus hernia] seen in about 7% of cases². CDH usually presents in the new-born period with respiratory distress which may worsen progressively. Delay in diagnosis and management can result in significant morbidity and mortality rate up to 80%³.

We report a male neonate with CDH and bilateral cryptorchidism managed at the Ekiti State University Teaching Hospital (EKSUTH), Ado-Ekiti.

The Case

A male neonate weighing 3.5kg admitted to the Special Care Baby Unit of EKSUTH at the 9th hour of life with respiratory distress. He was referred because of 'mild asphyxia' and respiratory distress from a secondary health care facility following vagina delivery at term. Mother was a 26 year old P3+1 who booked late at 28 weeks gestation, had one obstetric scan done at 36 weeks which was reported normal. Apart from her being managed for hypertension in pregnancy with α -methyl dopa, she had no other medical abnormality. She denied using herbal preparations, ingestion of alcohol, cigarette smoking or substance abuse.

Baby had depressed primitive reflexes, respiratory distress, cyanosis, hypothermia (temp- 35.9°C) and low oxygen saturation

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range; 52%-57% in room air, which improved to between 88%-91% following oxygen administration via nasal cannula. Heart rates and sounds were normal but heard only on the right hemi-thorax. The breath sounds on the left hemi-thorax were absent and replaced with bowel sounds which were absent in the abdomen. The scrotum was hypoplastic and the testes could not be palpated in both the scrotal sac and the groins.

Chest x-ray revealed right-sided mediastinal shift and bowel loops in the left hemi-thorax (Figure 1). Abdominal ultrasound scan revealed absence of the spleen and bowel loops in the abdominal cavity, normal liver and kidneys; the testes were also not visualised in the scrotum, groin or abdominal cavity. Echocardiography was normal.

An exploratory laparotomy was done on the fifth day of life to repair the defect following inability of the parents to honour cardiothoracic surgical repair referral on financial grounds. At surgery, the diaphragmatic defect was on the left side with herniation of the spleen, transverse colon, descending colon and the small intestine into the thoracic cavity. There was also pulmonary hypoplasia and malrotation of the gut. The testes were seen attached to the mesentery but could not be mobilized to the scrotum. Baby made good recovery postoperatively and was discharged home seventeen days after surgery. Chest x-ray done after surgery showed that the lung on the left side has re-expanded and there was no evidence of mediastinal shift (Figure 2). He is currently doing well on follow up. He is being planned for orchidopexy later.



Fig 1: Chest x-ray of the neonate with congenital diaphragmatic hernia



Fig 2: Post-operative Chest x-ray showing normally placed heart and expanded right lung tissue

Discussion

Most CDH cases (60%) in the developed world are diagnosed prenatally thus allowing proper antenatal counselling about the severity of the CDH, method of delivery, expected outcomes and long-term morbidities^{2,4}. Prenatal ultrasound diagnosis [PUD] helps in making informed decision in planning for the delivery of the baby at the appropriate facility with the necessary skills and personnel. PUD has high yield, aids surveillance for in-utero complications and also useful in prognostication^{4,5}. Our patient failed to enjoy these benefits due to inadequate antenatal care (ANC). The hallmark of PUD is the detection of abdominal organs within the thoracic cavity. Other indirect signs are presence of polyhydramnios, abnormal cardiac axis or mediastinal shift⁴.

However, some CDH cases may be missed prenatally thus presenting with progressively worsening respiratory distress after delivery⁴ as seen in this patient. Examination may reveal a barrel-shaped chest, scaphoid abdomen, absence of breath sounds on the ipsilateral side, shifted cardiac sounds and bowel sounds in the chest⁴. Chest and abdominal X rays are usually diagnostic showing contralateral mediastinal shift with the stomach and gas filled bowels within the chest⁶. Although most of these features were present in our patient, the diagnosis was initially missed at the referring centre as he was being managed for congenital pneumonia which is a close differential of CDH. The report of the ultrasound done at 36 weeks of gestation was not helpful and might have contributed to the misdiagnosis at the referral centre. The prompt referral of the patients for further care deserves commendation and typifies the envisaged positive collaborations expected among

different care levels in delivering quality health care to Nigerian patients.

The aetio-pathogenesis of CDH is poorly understood. Some authors have demonstrated reduced plasma level of retinol and retinol binding proteins in the cord blood of babies with CDH.⁷ Others have implicated maternal exposure to alcohol, smoking, periconceptional low intake of retinol, obesity and other substances^{7,8}. Association with some chromosomal or genetic abnormalities as parts of syndromic entities such as Trisomies 13, 18 and 21, Beckwith-Wiedemann, Turner, Denys-Drash, Cornelia -de- lange, Fryns, Donnai-Barrow, Pallister-killian and thoracoabdominal syndromes have also been proposed^{2,9}. There was no identifiable risk factor for CDH in this patient except for the α -methyl dopa that the mother took for the hypertension in pregnancy and this was commenced late in pregnancy by which time the defect would have formed. The lack of identifiable risk factor is not surprising as the aetiology of CDH is unknown in about 80% of cases⁴.

Definitive repair of the diaphragmatic defect is by surgery. This was successfully offered to the baby and he made satisfactory recovery afterwards necessitating his discharge. He is currently on follow up for long-term CDH sequelae like pulmonary diseases, gastro-intestinal morbidity, poor growth, neurological impairment, hearing loss, musculoskeletal abnormalities and poor life quality^{10,11}. Other future plans for the patient include a repeat surgery to close the abdominal wall fascia and also to attempt relocating the testes into the scrotal sac in addition to growth and nutritional monitoring.

Conclusion

This case emphasizes the need for thorough evaluation of neonates with respiratory distress using both clinical and simple investigational tools to exclude differentials. The case also highlights the need for appropriate referral and linkages between health care facilities in addition to good ANC as there was no prenatal suspicion of CDH in the patient. It also highlights the need for appropriate training of health care workers at the primary or secondary care levels.

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