Death Due to Late Onset Diaphragmatic Hernia

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Abstract

Diaphragmatic hernia is usually congenital, nevertheless it can also be acquired, particularly following trauma. Most of late onset hernias are acquired and reported cases of congenital type are rare. This case explicates a late onset congenital diaphragmatic hernia (CDH) where the diagnosis was made at the time of autopsy. Autopsy revealed the stomach and the proximal small intestines to be present in the left hemi thorax through a 4.5cm X 5cm defect in left hemi-diaphragm. Left lung was found to be collapsed and 60 g in weight. Histology revealed pulmonary hypoplasia. The Cause of Death was declared as CDH complicated with lung hypoplasia.

Keywords

- Diaphragmatic hernia
- Congenital defect
- Lung atelectasis

Introduction

In 1679, Riverius registered the first noted case of a congenital diaphragmatic hernia (CDH). It was diagnosed after death of a 24-year-old man.[1] The first effort to operate on a CDH was by Nauman in 1888. Laparotomy was accomplished in a 19-year-old man with acute respiratory distress and acute abdomen. In 1889, O'Dwyer attempted the first repair of CDH during infancy.

Diaphragmatic hernia is a congenital defect of the diaphragm which results

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from incomplete closure of the pleuroperitoneal canal or septum transversum during fetal development. Four to ten percent of all infant deaths are due to congenital anomalies caused by congenital diaphragmatic hernia (CDH).[1] It is usually diagnosed immediately after birth or within the neonatal period [2,3] with the presence of severe respiratory distress. The term 'Late onset CDH' refers when the symptoms appear after the neonatal period and they account for 10%-13% of all CDH cases.[4, 5]

This case explicates where the diagnosis was made at the autopsy of an eleven-year-old boy.

Case Presentation

The deceased patient was an 11-year-old boy who was living in a children's home since two months of age. According to the matron's statement he was not diagnosed to have any major medical ailment, except for intermittent mild to moderate respiratory infections for one year. He has had complained of abdominal pain and nausea for 3 days and he had been taken twice to a General Practitioner. He was treated for gastritis and gastroenteritis on both occasions. Despite the treatment given, the symptoms did not subside completely. He developed vomiting and diarrhea night before his death.

However, he had fallen asleep after dinner, after having two biscuits as the discomfort subsided to a certain extent with vomiting. He was found unresponsive on the following morning and the body was cold at that time. The matron had noticed the presence of vomitus around his pillow and on the floor. He was immediately brought to the hospital. He was pronounced dead on admission at the outpatient department. The patient did not give any history of blunt or penetrating trauma to chest or abdomen. An inquest was ordered. A post mortem autopsy was ordered by the inquirer into sudden deaths. The autopsy was done on 17th April 2017, after about six hours of confirmation of death.

Autopsy Findings

The body was that of a slim child with a brown skin complexion. There were no signs of any dysmorphic features or congenital abnormalities. There were no external injuries. On opening the thoracic cavity, it was noted, that the stomach and the proximal part of the small intestines to be present in the left hemithorax **Figure 1**. On further dissection a 4.5X5cm defect was noted in the posterior part of the left hemi-diaphragm **Figure 2**. The stomach contained whitish undigested rice meal. Bowels did not show any necrosis, malrotation or malfixation throughout its entire length.

Left lung was found to be collapsed **Figure 3** and was 60 g in weight. The right lung was normal in size and weighing 110g. Features of pulmonary hypertension were not seen. Histological examination of the lung revealed pulmonary hypoplasia of the left lung whereas all other organs showed normal histology. Cause of death was given as late onset diaphragmatic hernia, complicated with lung hyperplasia.

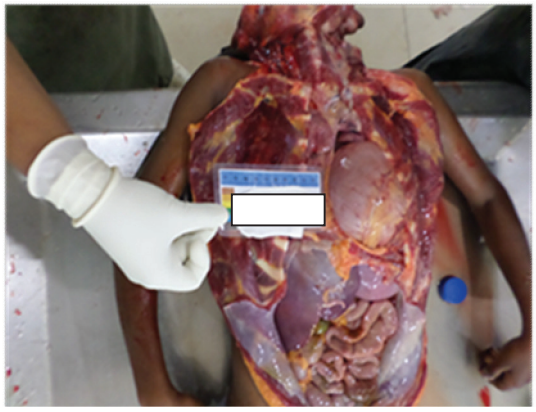

Figure 1: Position of the organs in the chest and abdominal cavity

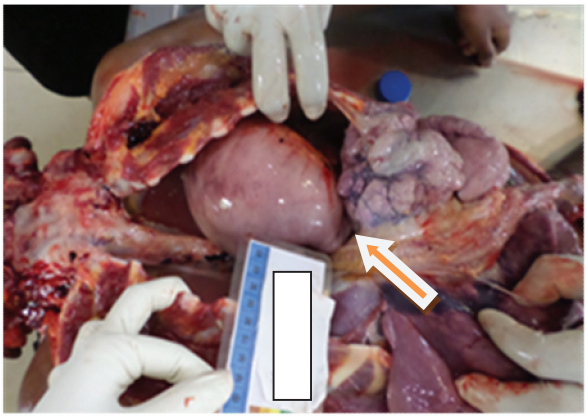

Figure 2: Defect in the diaphragm

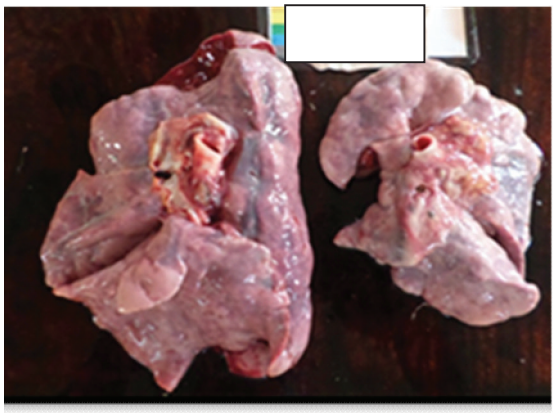

Figure 3: Normal right lung with hypoplastic left lung

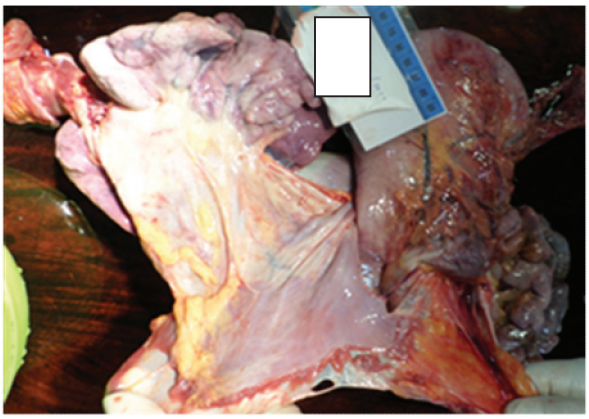

Figure 4: Hernation through the diaphragm

Discussion

Diaphragmatic hernia is usually congenital. Nevertheless, it can be of acquired type, particularly following trauma. Most of the time, late onset hernia are acquired in nature [6,7,8] which occurs following blunt trauma as it happens in falls or blows, or penetrating injuries (stab or firearm injuries). Rarely, it could be an iatrogenic due to an inadvertent injury during surgery.[9]

In this case the patient happened to be a previously healthy child. There was a history of only intermittent attack of chest infections. A plain radiograph which had taken four years ago is said to be apparently normal. There was no history of trauma or any surgery. Postmortem features showed herniation of bowels into the left hemi thorax through a diaphragmatic defect with pulmonary

hypoplasia. This indicates a long-standing problem rather than an acute event. Recurrent lung infections as given in the history also indicate a chronic event. It is probable that interference with the lung movements and function particularly on the left side could have been the reason for these recurrent episodes of lung infections. In this case the history together with the post-mortem features indicates a diagnosis of congenital diaphragmatic hernia excluding acquired defect of the diaphragm.CDH occurs in about 1 in 3300 live births[10] and it is usually diagnosed before or just after birth. Prenatal diagnosis of CDH is possible in majority of cases. Earlier, 80% of patients with CDH used to die in their neonatal period in spite of optimal treatment.[11] Recent studies have shown that there is an immense impact on the outcome of the child with CDH with advent of prenatal diagnosis and successful treatment has reached to 80 %.[12] Recurrence rate of CDH is 2% in subsequent pregnancies on a rough estimate.[12]

Three types of CDH are identified. 'Bochdalek' Hernia is the most common type. The defect is confined to the posterior or lateral aspect of the diaphragm. It is more common in left than right side of the diaphragm and carries a high rate of mortality. 'Morgagni' Hernia is rare, and the defect of the diaphragm is located in the anterior part of the diaphragm. 'Diaphragm event ration' the third type is used to describe when there is elevation in an intact diaphragm. In this patient the defect was found in the posterior part of the left dome of the diaphragm which goes along with the classical picture of 'Bochdalek' Hernia.

Incomplete closure of the diaphragm during fetal development, herniation of abdominal viscera to the chest and lung hypoplasia are the three main pathologies which are identified in CDH. If hernia formation precedes the development of lung, pulmonary hypoplasia sometimes occurs with severe respiratory symptoms at birth. In adulthood, it is not common, the development of lungs in most instances is normal and therefore symptoms are rare. [13] In this case histology revealed lung hypoplasia which indicates that the herniation of abdominal contents could have been there since birth. It is difficult to explain the absence of any significant symptoms and signs for such a long period. Both Lurie and Enns et al. have published remarkable papers of chromosomal anomalies in association with CDH.[14, 15] There are many other congenital chromosomal abnormalities believed to be associated with pathogenesis of congenital hernia i.e., genes COUP-TFII, FOG2, GATA4, WT1, and SLIT3.[16]

In the older age groups there are two clinical scenarios are common: an incidental finding on plain radiographs performed for reasons other than the hernia [17] or when symptoms develop due to incarceration, strangulation and visceral rupture inside the chest cavity. In this case neither of them was present but in author's opinion the history is a little unreliable as he was inmate of a children's home and possibly absence of close supervision could have missed certain features of

CDH. The diagnosis is made by a simple chest radiograph, computerized axial tomography or an MRI. Although it was revealed that an X-Ray was taken four years prior to his death, it was not available for observation. This could have been an important piece of evidence to come to a conclusion whether this to be an acute or chronic event.

In this case most of the features present were common with some other pathologies like gastritis and gastroenteritis. In a case of late presenting type there is usually a lesser degree of herniation of small part of bowel since birth, which could be misdiagnosed due to non-specific symptoms of vague nature.

It is said that the presentation, management and prognosis are somewhat different in congenital and acquired forms of diaphragmatic hernia. In neonatal type, presentation is mainly with the respiratory distress compared to late onset CDH. In the latter type those respiratory symptoms together with recurrent respiratory infections are common mainly with right sided CDH. Left sided CDH are mainly presented with gastrointestinal symptoms which matches the findings in this case. The deceased here also had abdominal pain, vomiting and loose stools which again mimics the common clinical picture. According to the history he had not had any significant history of those symptoms before.

Prognosis of late onset CDH is excellent with early diagnosis and surgical repair than the neonatal type, but the morbidity is high. [2, 3] Another important fact which has direct impact on the prognosis is the association of other malformations with late onset diaphragmatic hernia. Main co-existing anomalies identified are gut malrotation or malfixation, pulmonary hypoplasia, pulmonary sequestration, umbilical hernia, atrial septal defect, ventricular septal defect, polysplenia, type 1 diabetes mellitus. Gut malrotation and the pulmonary hypoplasia are the most two common associations.[2] However, 50 to 60 percent of cases of congenital diaphragmatic hernia are isolated, which means that patients have no other major anomalies. In this case, the presence of pulmonary hypoplasia was confirmed by the histology.

Conclusion

Congenital diaphragmatic hernia is relatively uncommon in the older age group. Severe defects usually present soon after birth. Nevertheless, less severe ones can present later as in this case. As children find it difficult to explain their problems, it might lead to missed or wrong diagnoses in some instances. The insight to this anatomic defect is crucial to identify and manage patients of older age groups, as it should be surgically corrected to avoid complications or to deal with them if they are already present.

This case highlights the importance of higher clinical vigilance when an early teenage patient presents with the vague gastro-intestinal and respiratory symptoms as timely medical intervention could save his life.

Ethical Considerations

The material contained in this article refers to coronial autopsy report which is publicly available according to Srilankan law. However, authors have observed anonymity and other general guidelines biomedical research in preparation of the paper.

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Conflict of interests

There are no conflicts of interest.

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