

Late-presenting congenital diaphragmatic hernia: a potentially life threatening case

遲顯現的先天性膈疝：一個有潛在生命威脅的個案

HY Chan 陳浩然, CC Wong 黃哲峻, F Ng 吳奎

Congenital diaphragmatic hernia (CDH) generally presents with respiratory distress in the neonatal period. Late onset CDH is less common and is associated with a wide range of clinical symptoms. We report a case of a 4-year-old child presenting with sudden onset of dizziness, abdominal pain and vomiting after swimming. Radiological investigations showed a left CDH with mediastinal shift. She gradually developed respiratory distress after admission. Urgent operation showed that the contents of the hernia included stomach, spleen, small and large bowels. This case highlights the importance of suspicion of CDH, proper clinical examination and investigation of children with acute non-specific gastrointestinal complaints. (*Hong Kong j.emerg.med.* 2009;16:102-105)

先天性膈疝一般在新生期呈現呼吸窘迫。遲開始顯現的先天性膈疝較少見，及有廣泛的臨床症狀。本文報告一名4歲的兒童於游泳後突然開始呈現頭暈、肚痛及嘔吐的個案。放射檢查顯示有左側先天性膈疝及縱膈移位。入院後她呼吸愈來愈困難。緊急手術顯示疝內載物包括胃、脾及大小腸臟。這個案突顯了懷疑先天性膈疝，正確臨床檢查及調查兒童急性非特異性腸胃申訴的重要性。

Keywords: Congenital abnormalities, diaphragmatic hernia

關鍵詞：先天性異常，膈疝

Introduction

Patients with congenital diaphragmatic hernia (CDH) commonly present in the neonatal period with respiratory distress. Late-presenting CDH is less common and the majority present with non-specific gastrointestinal or respiratory symptoms later in childhood or even in adult life. CDH with mediastinal shift by hernial contents beyond the neonatal period is a rare but potentially life threatening emergency.

The purpose of this case report is to familiarise emergency physicians with the clinical presentations that can occur in children with CDH, in order to facilitate the management of this potentially dangerous condition.

Case report

A 4-year-old girl presented with sudden onset of dizziness, upper abdominal pain and vomiting after swimming in July 2008. She was brought by ambulance to our emergency department. This child had a normal birth history and enjoyed good past health all along. There was no associated history of trauma, fever or shortness of breath. Her vital signs were within normal range and SpO₂ was 97% on room air. Clinically, she was not in respiratory distress. On physical examination, the neurological and abdominal

Correspondence to:

Chan Ho Yin, MRCP(Irel), MRCEd, FHKAM(Emergency Medicine)
Caritas Medical Centre, Accident & Emergency Department
111, Wing Hong Street, Shamshuipo, Kowloon, Hong Kong
Email: henrychanhy@yahoo.com

Wong Chit Chun, MBChB(CUHK)
Ng Fu, MRCP(UK), FRCSEd, FHKAM(Emergency Medicine)

examinations were unremarkable. However, she was found to have decreased left chest expansion. On auscultation, breath sounds were decreased on the left side with dullness on percussion over the lower two-thirds of the left hemithorax. Plain chest radiograph showed the presence of the gastric bubble and bowel loops in the left hemithorax with mediastinal shift to the right side (Figure 1). Left CDH was diagnosed and the patient was admitted to the surgical unit. She only complained of epigastric discomfort with no respiratory symptoms after admission. Her clinical condition was stable with pulse oximetry reading of SpO₂ 100% on room air. She was then transferred to the paediatric surgical unit of another hospital for further management.

About seven hours after the initial presentation, the patient developed increasing shortness of breath with respiratory rate of 36 per minute. A nasogastric tube was inserted for decompression of the hernial content. Urgent water soluble contrast follow through study was performed and showed the fundus of stomach was located inferiorly while the gastric outlet was located superiorly and in the left hemithorax (Figure 2). On laparotomy, a left CDH was identified with defect at the posterolateral aspect of the diaphragm (Bochdalek hernia), with herniation of the stomach, all small bowel, large bowel and spleen into the left hemithorax. The herniated contents were replaced into the abdominal cavity and repair of the diaphragmatic hernia was performed.

The patient complained of persistent shortness of breath after operation. Clinically, there was decreased breath sounds and increased resonance on percussion over the left chest. Chest radiograph showed the left lung had not yet expanded, with a left pneumothorax. The mediastinum was deviated to the right side with the right upper lobe collapsed (Figure 3). The patient was admitted to the intensive care unit and a chest drain was inserted. The patient's condition was then stabilised and serial chest radiographs showed left lung expansion with resolving pneumothorax (Figure 4). The patient made a good recovery and was discharged on the tenth post-operative day.

Discussion

Most CDHs are detected antenatally by ultrasound with mean gestational age at diagnosis of 24 weeks. The defect is usually posterolateral (Bochdalek hernia),

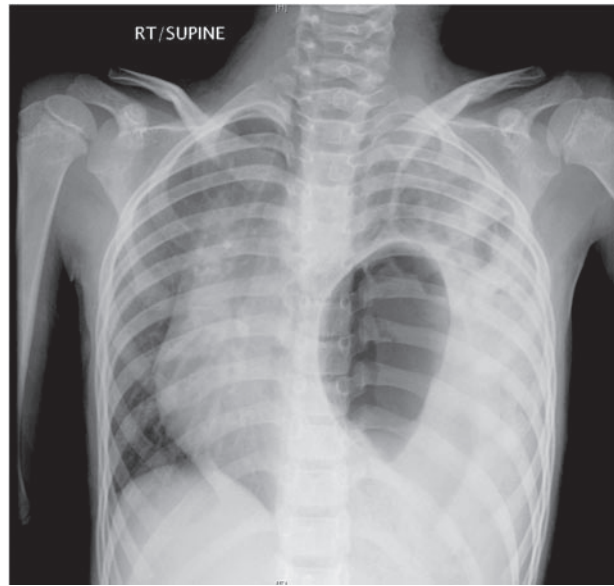


Figure 1. Chest radiograph showing the gastric bubble and bowel loops in the left hemithorax with mediastinal shift to the right side.

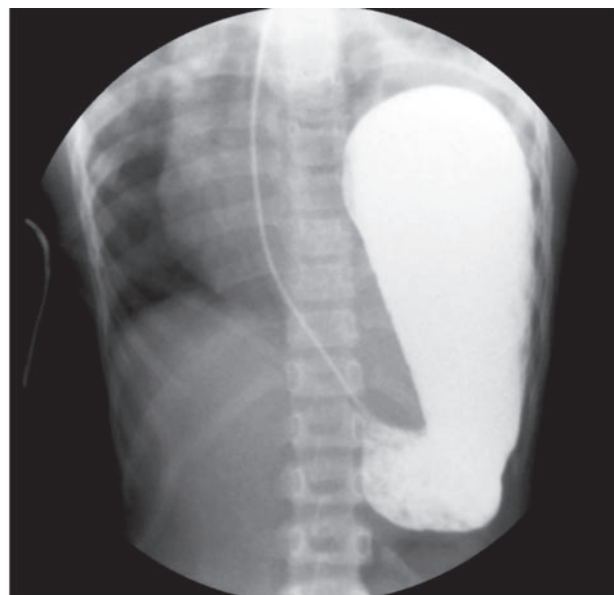


Figure 2. Water soluble contrast follow through study showing the fundus of stomach is located inferiorly while the gastric outlet is located superiorly and in the left hemithorax.

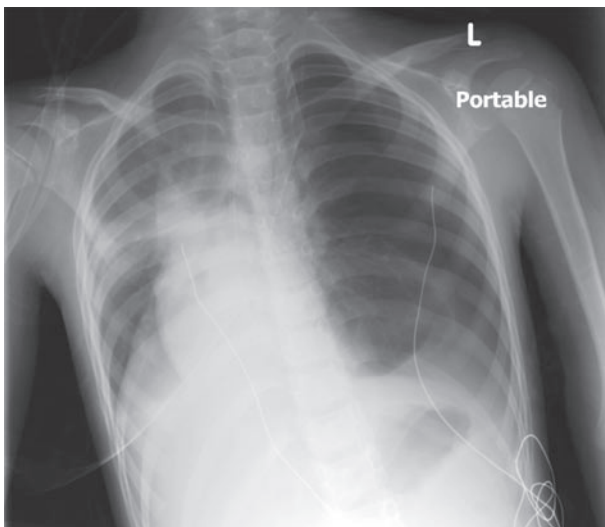


Figure 3. Chest radiograph showing left pneumothorax with mediastinal shift after operation.

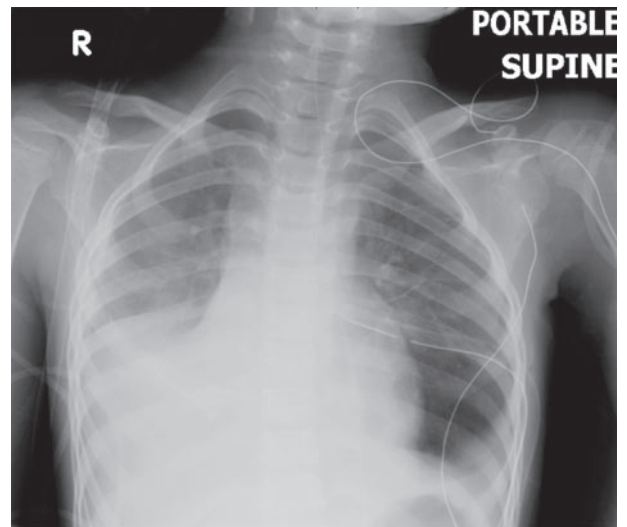


Figure 4. Chest radiograph showing resolving left pneumothorax after chest drain insertion.

but may be anterior (Morgagni hernia), or rarely central. However, 10-20% of cases are recognised beyond the neonatal period¹ and they most commonly present within one year of age.² Some series reported a higher incidence of late presenting CDH in males^{3,4} but no conclusion has been made about gender association. The presentation can either be acute or insidious onset with respiratory or gastrointestinal symptoms while acute gastrointestinal obstruction or respiratory distress is rare.⁵ CDH should be considered in the differential diagnosis of any child with unusual gastrointestinal or respiratory symptoms and abnormal chest X-ray film findings.

CDH is generally considered to result from defective formation and fusion of the pleuroperitoneal membranes which provide a potential passage for abdominal contents to herniate into the thorax. The relative rarity of right CDH is due to the earlier closure of the right pleuroperitoneal opening and to the protective effect of the liver developing in the septum transversum. The explanations of delayed presentation are that the herniation of bowel loops are protected by the right and left lobes of the liver or by a 'sac' associated with the abdominal contents to the thoracic cavity. CDH increases in size with

somatic growth and herniation may be provoked by mechanical factors that increase intra-abdominal pressure such as swimming and sneezing after exercise as in our case.

The outcomes of late-presenting CDH are usually favourable,⁶ which are related to the absence of accompanying pulmonary hypoplasia and low incidence of other congenital malformations. However, it can result in many complications e.g. gastric volvulus, gastric perforation, bowel necrosis, splenic torsion.² Deaths have been reported as a result of mediastinal shift with kinking of the vena cava and cardiovascular collapse by the herniated abdominal viscera.⁷

A previously normal chest radiograph does not exclude late onset CDH. The radiological findings may sometimes be misleading; it may resemble pneumothorax, pleural effusion and pneumonia. A chest radiograph following the passage of a nasogastric tube is useful in facilitating the diagnosis of CDH, since there is a risk of misinterpretation with the potential hazard of inserting a chest drain into the herniated viscus. In one series, 18% of the cases of CDH were subjected to insertion of chest drain.¹ The

diagnosis can be confirmed by contrast follow through study⁸ but it may underestimate the extent of herniation due to spontaneous reduction in the erect posture by the hydrostatic pressure of the barium. Computed tomography and liver-spleen scintigraphy seem to be the best diagnostic options in children with right-sided CDH.³

Among the patients with left-sided CDH, more than 30% had the spleen herniated into the chest² and this was also present in our patient. One case of late CDH with splenic infarction presenting as pleural effusion has been reported.⁹ Herniation of a solid organ e.g. liver, spleen, is recognised as a risk factor for radiological misdiagnosis of late presenting CDH in children because the typical appearance of bowel herniation may be sparse or obscured leading to a diagnosis of lung disease instead.¹⁰

Respiratory support is most important in the emergency management for the patient with CDH. Blow-by oxygen or bag-masking can lead to gastric and abdominal distension and compression of the lung and should therefore be avoided. Patients presenting with severe respiratory distress should be immediately intubated and ventilated with low peak inspiratory pressure to minimise lung injury. A nasogastric tube connected to continuous suction should be placed in the stomach for decompression of the abdominal contents and decreasing respiratory compromise. Extracorporeal membrane oxygenation is generally reserved for patients who continue to have hypoxia and hypercarbia refractory to conventional ventilation.

Surgery was performed via laparotomy in our patient. Chest drain insertion was usually performed at the end of the operation for drainage to help the affected lung to re-expand. The thoroscopic or laparoscopic approach for the repair of late-presenting CDH has been described recently.¹¹ However, it is not recommended in unstable children with mediastinal shift or acute incarceration of CDH because the insufflation pressure can contribute to circulatory collapse and cardiac arrest.

Conclusion

We report a case of delayed presentation of a potentially life threatening CDH. The variable clinical features of CDH presenting beyond the neonatal period may result in clinical and radiological misdiagnosis. CDH with complicating mediastinal shift and respiratory distress requires urgent gastrointestinal decompression and respiratory support. The most significant factor in achieving diagnostic success is to consider it early in the differential diagnosis to avoid misguided or delayed therapy.

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