

Diagnosing Boerhaave Syndrome in a 16-year-old girl in a primary care centre

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Received: June 2022 Accepted: July 2022; Published: August 1, 2022.

Citation: Javed Khan, Abdul Muhid, Farah Mushtaq. Diagnosing Boerhaave Syndrome in a 16-year-old girl in a primary care centre. World Family Medicine. 2022; 20(8): 93-97 DOI: 10.5742/MEWFM.2022.9525125

Summary

Boerhaave syndrome is the spontaneous rupture of the oesophagus due to raised intra-oesophageal pressure and is a potentially fatal condition. The patient often presents with retrosternal chest pain and upper abdominal pain associated with significant history of retching and vomiting. The condition has a mortality rate of up to 40% hence early diagnosis and timely intervention is crucial. This emergency is rarely seen in children and young adolescents. We report a case of Boerhaave syndrome in a 16-year-old girl who presented to a primary care setting with vomiting, a abdominal pain, and subcutaneous emphysema. The diagnosis of oesophageal rupture was made clinically. Thus, immediate referral to secondary care was vital in providing urgent management. This discussion highlights that this emergency can present in younger age adolescents, therefore high index of suspicion is imperative for good prognosis.

Key words: Boerhaave syndrome, oesophageal perforation

Background

Boerhaave syndrome is rare in children under the age of 18 years however it should be considered if there is a history of neck, chest or abdominal pain associated with preceding vomiting episodes. Focused clinical evaluation especially to look for subcutaneous emphysema, pneumothorax and pleural effusion is important. There should be a high index of clinical suspicion and awareness of this condition by front-line clinicians. These are the most important factors especially in a primary care setting where there is no access to immediate imaging like a chest X-ray. This case highlights a positive outcome after a timely referral to specialist services in hospital was made with a clinical diagnosis of oesophageal rupture in primary care. Absence of classic features can cause a diagnostic challenge.

Case Report

In a general practice surgery, a telephone call was triaged by a general practitioner (GP). The call was from a mother of a 16-year-old Caucasian girl who was previously fit and well with no significant past medical history. The mother mentioned that her daughter has recently been complaining of constipation and had vomited several times overnight. The patient reported passing small amount of stool the day before. The mother was advised to bring her daughter to the surgery for a medical consultation. The patient was reluctant to visit the surgery and declined a face-to-face appointment. Therefore, telephonic advice was given about managing the constipation and offering anti-sickness medication along with safety netting. The mother rang an hour later as she was concerned about the appearance of her daughter's chest and there was also complaint of mild chest and abdominal pain. An urgent review was arranged in the primary care. The patient appeared alert but unwell. Her blood pressure was 126/84 mmHg, heart rate was 133 beats/ minute, respiratory rate was 18 breaths/ minute, temperature 37.7° with oxygen saturation being 98% in

air. Chest examination revealed a palpable subcutaneous emphysema over the anterior chest wall. There was no respiratory distress. Her abdomen was generally tender, and her urine dipstick showed presence of ketones.

The provisional diagnosis made by the GP was oesophageal rupture due to excessive vomiting, leading to subcutaneous emphysema. The patient was admitted urgently to the local hospital for further management under the care of surgical team. In the hospital, the patient had a chest x-ray showing a pneumomediastinum and subcutaneous emphysema as seen in Figure 1. This was also seen on CT thorax confirming the rupture of mid oesophagus (Figure 2). The patient was managed conservatively with nil oral intake along with nasogastric tube drainage, intravenous antibiotics, fluids, parenteral nutrition through a central line. She was an inpatient for nine days. The patient made a full recovery as no leak from the oesophagus was seen in the water-soluble contrast swallow study (Figure 3), on day 8 of admission at which point oral intake was commenced

Figure 1: Chest X-ray showing pneumomediastinum (red arrow) and subcutaneous emphysema (yellow arrow).

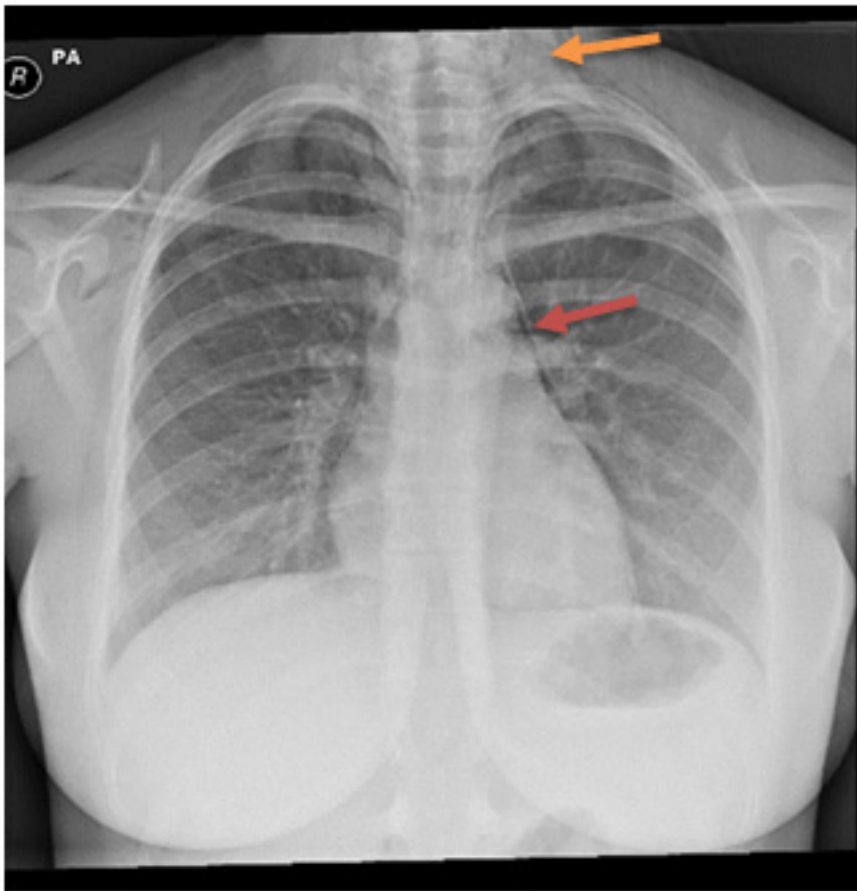


Figure 2: CT thorax coronal view showing pneumomediastinum as shown by the red arrow and subcutaneous emphysema indicated by the yellow arrow.

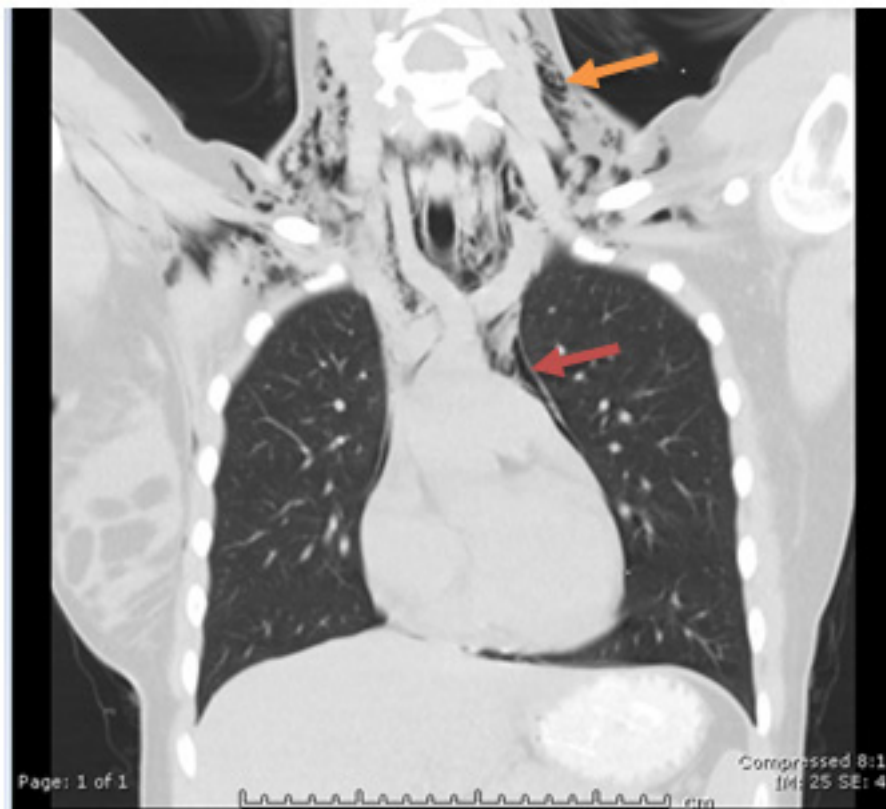
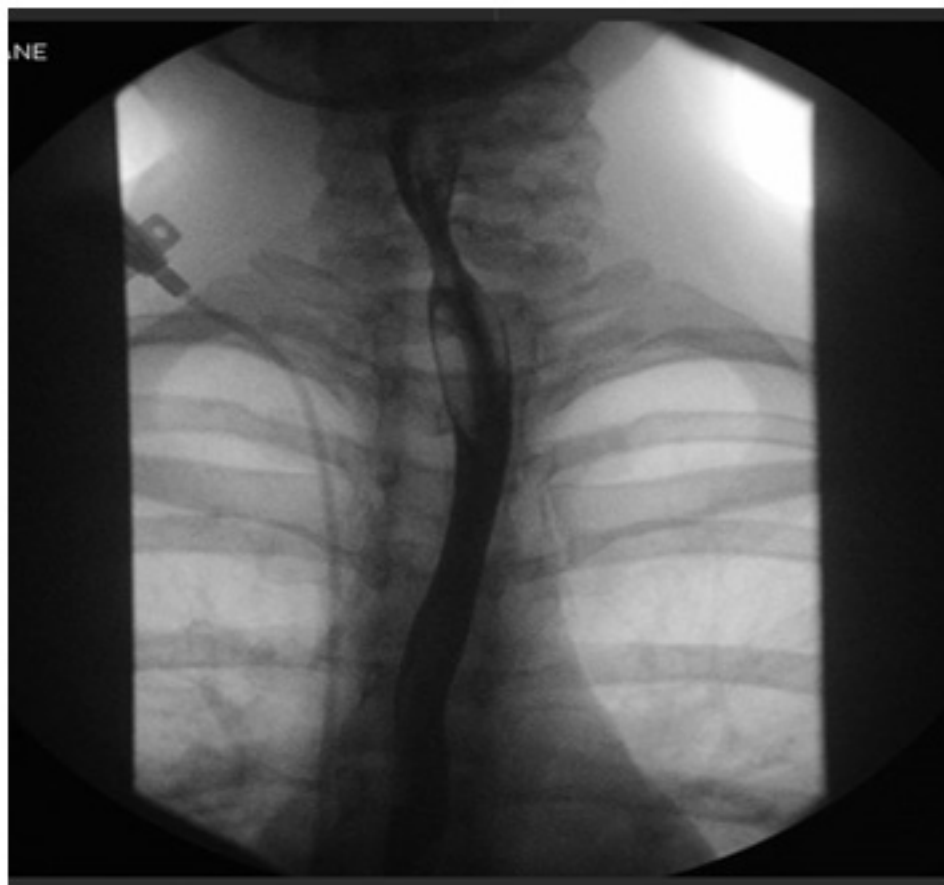


Figure 3: Water soluble contrast swallow where the contrast is seen flowing through the oesophagus with no evidence of perforation or leak on day 8 of admission. There is no bolus hold-up.



Discussion

Spontaneous transmural oesophageal rupture, also known as Boerhaave's syndrome, is a rare condition occurring due to sudden increased intra-oesophageal pressure. This is usually seen because of repeated vomiting or retching. The first case of Boerhaave syndrome was reported in the 18th century, where an elderly male developed vomiting after a large meal, leading to oesophageal rupture (1). The patient died 24 hours later with septic shock. Oesophageal ruptures most commonly occur in the distal third as it is inherently weaker than the rest of the oesophagus with lack of supportive surrounding tissues (2).

Vomiting is considered to be the most common cause for spontaneous oesophageal rupture, often linked to alcohol abuse, excessive food intake and bulimia (1,2,5). Other causes which can be implicated in increasing the oesophageal pressure are heavy lifting, during labour, straining at defecation and epileptic seizures (2, 5). Literature review shows men between the ages of 30-50 years are at greater risk for oesophageal perforation (5, 8). The rupture occurs typically in healthy oesophagus though there have been rare case reports of rupture seen in individuals with eosinophilic oesophagitis (5, 6, 7). In children under the age of 18 years, oesophageal rupture is extremely rare (6,10). Literature reviews in 2006 and 2009 found only 27 reported paediatric cases, with 12 being outside the neonatal period (8, 10).

This condition can have a wide spectrum of signs and symptoms; from relatively well looking individual to septic shock (4). The esophageal perforation can be a small air leak in the mediastinum to large air and fluid accumulation in the pleural cavity leading to life threatening complications. There can be non-specific chest pain that itself leads to a wide differential diagnosis (1). The non-specific vague symptoms or atypical presentation without the classic symptoms and signs causes a diagnostic delay, which can occur in up to one third of cases (3, 5, 9). The clinical symptoms may vary depending on the site of the rupture of oesophagus, degree of leakage and duration of perforation. The most common symptom is pain in the neck, chest or upper abdomen. The pain may radiate to the back and can be aggravated by swallowing (5, 9).

Commonly the rupture occurs in the left wall of distal third of oesophagus, though in children it is seen mostly on the right (10). This can lead to abdominal pain, mediastinitis, pleural effusion, emphysema, or necrosis due to spillage of gastric contents (5). As mid oesophagus lies next to right pleura, therefore mid-oesophageal or upper thoracic perforations tend to cause pleural effusion or hydropneumothorax on the right. Cervical oesophageal ruptures are usually benign as they often are more localized and their progression to the mediastinum through the retro-oesophageal space is slow, however it can cause neck or upper chest pain. The oesophageal perforation is the most lethal perforation of the gastrointestinal tract with mortality of 40-50% (1, 9). The life threatening complications like sepsis, shock, respiratory failure, cardiac and renal complications can occur (4,9).

Our reported case is unusual as a young 16-year-old girl with no known risk factors presented with spontaneous oesophageal rupture. The condition is rare in this age group. The patient presented with the classic features of the Mackler triad; vomiting, chest pain and subcutaneous emphysema. This triad is only present in 14 % cases of Boerhaave syndrome (1). These classical features can sometimes be accompanied with a crackling sound occurring with each heartbeat on auscultation, known as Hamman's sign, best heard in the left lateral decubitus position and is a manifestation of the pressure of the myocardium against the air infiltration in the mediastinal emphysema (3,5).

Initial management involves fluid resuscitation, broad spectrum antibiotics and gastric rest. The treatment options are conservative, endoscopic or surgical repair. Our patient underwent conservative management due to small contained rupture. With conservative management, the time to oral intake was 8 days similar to that reported in literature (4).

A retrospective study of 34 patients with oesophageal perforation, showed a diagnostic delay of more than 24 hours in 20 cases (4). Such a delay can lead to more tissue damage along with risk of sepsis and poorer outcome. The intensive care time was 1.5 days in early diagnosed and treated group compared to 6.5 days in those with delayed diagnosis (4). A delay of 24 hours or more in diagnosis doubles the mortality (9,10).

To our knowledge, this is first case of a teenage girl presenting to primary care where a prompt diagnosis of spontaneous oesophageal rupture was made clinically and emergency referral was made to the specialist services. This potentially fatal condition can present in children with no associated risk factors as seen in this case. A positive outcome solely depends on rapid recognition and prompt treatment.

Learning Points

- 1. Boerhaave syndrome is rare but can present in children.**
- 2. It should be considered if there is neck, chest or abdominal pain preceded by vomiting and retching, leading to focused clinical examination.**
- 3. Delay in the diagnosis can be fatal. Early diagnosis of oesophageal perforations, within 12 to 24 hours, have the best outcomes.**

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