Spontaneous esophageal rupture as the underlying cause of pneumothorax: early recognition is crucial

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Abstract: Boerhaave's syndrome (BS), also known as "spontaneous rupture of the esophagus", constitutes an emergency that requires early diagnosis if death or serious morbidity are to be prevented. First described in 1724, BS is thought to be more common than once thought. Its true incidence remains unknown. Mortality ranges between 20-40% with timely treatment but this rises to virtually 100% if treatment is delayed by more than 48 hours. This is unfortunately a common occurrence due to delayed diagnosis. The commonest precipitating factor is vomiting but BS can be truly "spontaneous". The classical clinical presentation described consists of vomiting, chest pain, and subcutaneous emphysema. However, and contrary to popular belief, this triad is actually uncommon accounting for the frequently delayed diagnosis. A less recognised presenting feature of BS is with pneumothorax due to associated rupture of the parietal pleura. Pneumothorax has been shown to be present in more than 20% of cases of BS-sometimes with a coexistent pleural effusion (hydropneumothorax). This article aims to raise awareness about pneumothorax as the sole initial presenting feature of BS and alert clinicians to consider BS in the differential diagnosis of any patient with respiratory symptoms and a recent history of vomiting.

Keywords: Boerhaave's syndrome (BS); esophagus; perforation; pneumothorax; hydropneumothorax

Boerhaave's syndrome (BS), also known as "spontaneous rupture of the esophagus" constitutes a life-threatening condition. The adjective "spontaneous" does not imply absence of a precipitating factor, but instead the fact that the rupture is not a consequence of direct trauma (usually caused by instrumentation or a foreign body). The commonest precipitating factor associated with BS is vomiting although other precipitants have been described such as straining, lifting or even laughing (1). BS contrasts with Mallory-Weiss syndrome where the tear is confined to the mucosa and commonly associated with hematemesis. In BS, the tear is transmural leading to esophageal perforation. Hematemesis is rarely present (2).

From a historical viewpoint, BS was first described by the Dutch physician, Hermann Boerhaave in 1724. It was a post-mortem diagnosis for the Grand Admiral of The Netherlands, Jan van Wassenaer, who died 18 hours after developing excruciating chest pain following vomiting. On autopsy, the tear was identified in the distal esophagus and undigested food seen in the left pleural cavity (1,3).

The true incidence of BS in the general population is unknown. However, it is thought to be more common than once thought as many cases of BS are diagnosed post-mortem resulting in under-reporting and thus an underestimate both with regards to its incidence and mortality (3).

The classical clinical presentation of BS traditionally taught at medical school and described in textbooks is Mackler's triad. This consists of vomiting, chest pain, and subcutaneous emphysema (4). However, and contrary to popular belief, this triad is actually uncommon. Characteristically, in a series of 14 patients eventually diagnosed with BS, only one presented with this triad (5). As such, reliance on those clinical features can be misleading. As a matter of fact, the symptoms associated with BS are more often than not, non-specific, resulting in delayed diagnosis.

The pathophysiology of BS involves a sudden rise in intraluminal esophageal pressure forcing the gastric contents against a tight cricopharyngeus muscle. This is most commonly the result of retching or vomiting, although as previously discussed, these may be completely absent (3). In addition to the situations described earlier, BS can be truly "spontaneous" with no apparent predisposing factor. Cases of BS have been described in patients during bending over, watching television or even during their sleep (6).

The perforation in BS has a predilection for the left lateral aspect of the distal esophagus (90% of cases) (7,8). There are several anatomical reasons to explain this. These include thinning of the muscle in the distal esophagus, weakening of its wall as a result of vessels and nerves entering it, lack of supporting neighboring structures and the fact that at the left diaphragmatic crus, the esophagus makes an anterior angulation (5).

Once the esophagus perforates, a number of events follow. All of these events pose an imminent threat to life. Their exact effect depends on where in the esophagus the perforation has occurred. Since the distal left lateral aspect is the most frequently affected site, signs and symptoms relate to this site in 75-90% of cases (2).

As the esophagus ruptures, the parietal pleura can either rupture with it or alternatively, become breached at a later stage secondary to the enzymatic effect of the gastric contents. In either case, a pleural breach will lead to a pneumothorax or hydropneumothorax depending on whether air only or air with fluid have leaked from the esophagus into the pleural cavity (2).

Other conditions that can lead to BS include pneumomediastinum, mediastinitis, abscess formation and septic shock. These are all a direct result of the esophageal and/or gastric contents spilling into the mediastinum. If the perforation is sealed, the patient may appear deceivingly well with no (or few) signs of systemic inflammatory response syndrome before they suddenly decompensate. Hence, none of the aforementioned features may be present

in the initial setting and there may even be absence of any preceding history of vomiting or retching to complicate things further (3). As such, pneumothorax may be the sole initial presenting feature of BS.

It is of paramount importance for clinicians of all specialties to appreciate this, i.e., that BS can masquerade as "spontaneous" pneumothorax with no other "classical" features. Rarely, this can be a tension pneumothorax (2,9). As already stated, Mackler's triad is uncommon (5), and vomiting or retching are not always present (3). This clouds the clinical picture and makes the timely diagnosis of BS unlikely (3).

Diagnostic delay carries a very high risk of death. Similar to other acute esophageal disorders, the mortality of BS is exceedingly high and rises steeply with time (10). It is reported to be in the order of 25% if treatment is started within 24 hours, but reaches almost 100% at 48 hours (7,8). These exceedingly high mortality figures illustrate the critical importance of timely diagnosis of BS. Any diagnostic delay is very likely to lead to patient death.

Clinically, BS should always be considered in cases of pneumothorax or chest pain if early diagnosis is not to be missed. As the Harvard physician Soma Weiss who described the eponymous Mallory-Weiss syndrome said, "A diagnosis is easy, as long as you think of it". When a patient is misdiagnosed, appropriate treatment is delayed, and in the case of BS, death becomes almost a certainty (2).

It is therefore crucial that the diagnosis of BS is rapidly made even before the patient leaves the emergency department. Like with all cases in clinical medicine, history taking followed by physical examination are mandated. Although history alone will sometimes give away the diagnosis, more often than not, this is not the case. Radiological investigations most commonly provide the diagnosis in BS but clinical suspicion is essential so as to request these in time and look for the relevant signs (2).

The first radiological investigation to be requested is a simple chest radiograph. A chest radiograph is likely to show the presence of pneumomediastinum or pneumothorax (or hydropneumothorax if a concomitant pleural effusion is present) most commonly on the left (3). The presence of pneumomediastinum with a preceding history of vomiting or retching followed by acute chest pain is virtually pathognomonic of BS. However, pneumomediastinum may take more than an hour to develop and is not present in 10-12% of cases (3). It is thus imperative for clinicians to be aware of this percentage of false-negative results on chest radiograph. This will prevent false reassurance and

diagnostic delay.

In the presence of clinical suspicion for BS, a contrast swallow study is mandated. This is irrespective of whether or not the chest radiograph has revealed any positive findings. A water-soluble contrast medium such as gastrografin is recommended. This is not only likely to confirm the diagnosis by showing the extravasation of contrast into the mediastinum and/or pleural cavity (although 15-25% false-negative results have been reported for this examination, too) (11,12), but will also delineate the anatomical site of the perforation and thus guide the surgeon in their attempt to close the defect (11). Surgery, as part of a multidisciplinary approach, constitutes the "gold standard" treatment of this otherwise fatal syndrome if it is diagnosed within 24 hours (13). Various techniques and technologies are available in modern esophageal surgery but these are beyond the scope of this article (14,15). Beyond 24 hours, the prognosis significantly worsens and conservative treatment is usually advocated with surgery reserved for patients with a septic profile (9). Other investigations for BS include CT with or without oral contrast and needle thoracentesis if a pleural effusion exists. In the latter, biochemical and cytological examination of the pleural fluid can give the diagnosis by revealing the presence of salivary amylase and undigested food contents respectively (11,16).

In this editorial, we are hoping to raise the awareness about BS and how important it is for every clinician to have this diagnosis at the back of their mind when consulting a patient with the primary complaint of acute dyspnea, chest pain, or where a pneumothorax has already been diagnosed; even in the absence of a "typical history" for BS. The pneumothorax may only represent the "tip of the iceberg" and has been shown to be present in more than 20% of cases of BS-sometimes with a coexistent pleural effusion (hydropneumothorax) (17).

Thus, when encountering a patient with a pneumothorax, it is paramount to directly ask on history taking for predisposing factors for BS (such as vomiting or retching) and take a detailed pain history, look for signs of sepsis on clinical examination and observations, feel for surgical emphysema and request the appropriate imaging modalities in a timely manner. The clinician needs to look for the relevant imaging signs described earlier but also appreciate that false-negative results can occur.

The esophagus is an unforgiving organ (18,19). As it originates in the neck, extends through the thorax and terminates in the abdomen, it bears no respect for the arbitrary boundaries that exist between specialties (20). All clinicians, no matter what specialty they belong to, need to be aware that BS is probably more common than generally thought and of the different ways it can present. This includes pneumothorax as the sole initial presenting feature. Early clinical suspicion will lead to timely diagnosis and maximize the survival chances for the patient.

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