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A Case Report of MEN Syndrome Type 1 with Boerhaave's Syndrome Presenting As Tension Hydropneumothorax

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Abstract Boerhaave Syndrome is a spontaneous rupture of the esophagus due to repeated vomiting and has a high mortality rate. Multiple endocrine neoplasia(MEN) syndromes are autosomal dominant disorders that can develop tumors in endocrine glands. A male patient in his 40s was admitted to the emergency room with complaints of chronic diarrhea and nausea. The patient was hospitalized in the gastroenterology department with a preliminary diagnosis of acute pancreatitis which was excluded later. On the first day of hospitalization, the patient developed dyspnea with desaturation after repeated retching. Thoracoabdominal CT revealed massive pleural effusion on the right side. Therefore, a chest tube was inserted. Six-hour later, he became hypotensive and a chest X-ray revealed a left-sided pneumothorax. A second chest tube was inserted. He was diagnosed with Boerhaave Syndrome by esophagography. Metallic stent was placed in the ruptured site.He eventually died of sepsis due to mediastinitis due to Boerhaave's syndrome.

Keywords: *Boerhaave's Syndrome, Dieulafoy's lesion, MEN Syndrome, pneumothorax, Zollinger-Ellison Syndrome*

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1. Introduction

Effort rupture of the esophagus, or Boerhaave syndrome, is a spontaneous perforation of the esophagus that results from a sudden increase in intraesophageal pressure combined with negative intrathoracic pressure. This condition was first documented by the 18th-century physician Herman Boerhaave, after whom it is named [1]. Although iatrogenic causes are common, other traumatic events such as penetrating, blunt force, or abrasive injuries can also cause esophageal rupture.

The syndrome is characterized by a combination of symptoms, collectively known as Mackler's Triad, including lower chest pain, severe vomiting, and subcutaneous emphysema [2]. However, these symptoms may not appear at the same time and can delay diagnosis. Mortality and morbidity rates are high; Mortality rates are 20-40% in treated patients and almost 100% in untreated patients [3]. Prognosis depends on the extent of mediastinal invasion and the degree of damage to the mediastinal pleura, which can lead to pulmonary symptoms like pleural effusion, pneumothorax, shock, or sepsis.

Early diagnosis is critical, and any patient with severe vomiting, pain, or fever following esophageal instrumentation or chest trauma should be thoroughly evaluated for esophageal perforation. Cine Esophagram is essential for accurate diagnosis. Treatment options for Boerhaave syndrome include conservative, endoscopic, and surgical approaches, depending on the severity of the condition and the clinical status of the patient [4].

Tension pneumothorax is a rare but life-threatening condition frequently seen in intensive care settings and poses a high risk for patients on mechanical ventilation [5]. Boerhaave syndrome presenting as tension hydropneumothorax is extremely rare. Symptoms in such cases typically include recent vomiting, dyspnea, chest pain, and epigastric pain, but Mackler's triad (chest pain, vomiting, and subcutaneous emphysema) is not consistently present. Treatment strategies vary; Conservative approaches are available for stable patients and surgical options are available for patients with a septic profile. An alternative method involves aggressive sepsis treatment and radiologically guided drainage to avoid major surgery; This may lead to better healing of the esophagus and reduced mortality. A recent study shows that Video Assisted Thoracoscopic Surgery (VATS) is comparable to thoracotomy [3].

Multiple Endocrine Neoplasia Type 1 (MEN1) is an autosomal dominant syndrome associated with benign and malignant tumors in various endocrine tissues, primarily parathyroids, entero-pancreatic tumors and pituitary tumors. Gastrinoma, Zollinger-Ellison syndrome, is the

most common type of pancreatic endocrine tumor and is a component of MEN1 syndrome [6].

Clinical symptoms of Zollinger-Ellison syndrome (ZES), such as esophagitis, peptic ulcers, and diarrhea, are caused by excessive acid production in the stomach. Important methods for diagnosing ZES include gastrin radioimmunoassay, secretin stimulation test and somatostatin receptor scintigraphy (SRS), and endoscopic ultrasound (EUS) [7].

Dieulafoy's lesion (DL) is a rare cause of gastrointestinal bleeding that can occur anywhere in the digestive tract, accounting for up to 4% of acute upper gastrointestinal bleeding cases. Risk factors for DL include Helicobacter pylori infection, gastrinoma, and excessive gum chewing [8]. In our case, we aimed to emphasize tension pneumothorax, which was the initial presentation of Boerhaave syndrome, and the relationship between MEN1 syndrome and Boerhaave syndrome.

2. Presentation of Case

A 45 years old male presented to the emergency department (ED) with a 2 day history of abdominal pain, nausea and emesis. The patient was admitted to the gastroenterology service with the preliminary diagnosis of acute pancreatitis due to high serum amylase and lipase levels. His abdominal pain was non-radiating, localized to the epigastric region, and mild which was not consistent with the acute pancreatitis. In addition, the abdominal CT requested in the ED showed that the pancreatic parenchyma was normal except for the 1 cm tumor in the body of the pancreas. The findings did not meet the criteria for acute pancreatitis, except for elevated amylase and lipase; Therefore, acute pancreatitis was excluded. On further questioning, the patient complained of chronic diarrhea and unexpected weight loss. We decided to investigate these symptoms. On initial examination, cachexia, sarcopenia and epigastric tenderness were present. Laboratory examination revealed normocytic anemia, mild hypoalbuminemia and hypokalemia.

The patient has no known diseases, surgical operation or medication and has been smoking for 25 years. There was nothing remarkable in his family history. The patient has been experiencing the following symptoms for two years: nausea, vomiting and watery diarrhea (occurring 7-8 times a day). Prior investigations regarding these symptoms in the last 6 months revealed parathyroid adenoma with hyperparathyroidism, non-secretory adenoma in left surrenal gland and 1 cm pancreatic tumor. Additionally, this pancreatic tumor showed neuroendocrine tumor characteristics on endoscopic ultrasonography. Fine needle biopsy with EUS was performed but was unsuccessful. At that time, the serum gastrin level was 524 ug/ml.

On the first day of hospitalization, the patient developed tachypnea and desaturation after repeated gagging. On physical examination, rebound tenderness with guarding in the abdomen, decreased lung sounds, and dullness on the right side of the thorax were detected. Thromboembolic events such as pulmonary embolism and mesenteric ischemia were suspected in the patient, whose arterial blood gas analysis revealed high lactate level, normal pH level, hypercarbia and hypoxia.; therefore, Thoracoabdominal CT angiography was requested and no thromboembolism was observed, but pleural fluid reaching up to 5 cm on the right side of the thorax was observed (Figure 1). A chest tube was applied to the right hemithorax. After a chest tube was inserted, his symptoms subsided. Pleural fluid samples were sent to the laboratory for culture, cell count and biochemical analysis.

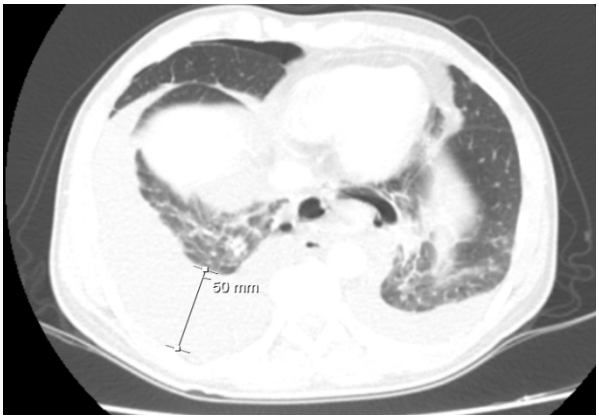

Figure 1. Bilateral pleural effusion on the thoracoabdominal CT angiography

Six hours after chest tube insertion, the patient developed hypotension that did not respond to intravenous fluids; Therefore, positive inotropic medication (noradrenaline) was commenced. On repeated physical examination, breathing sounds gradually decreased on the left side of the chest and heartbeats shifted to the right; Therefore, tension pneumothorax was suspected and confirmed by chest radiography (Figure 2). Another chest tube was placed in the left pleural space, immediately reducing the need for positive inotropic medication. Despite all interventions, signs of ischemic multiple organ damage such as increase in serum transaminase, creatinine level, and decrease in urine output were observed. He was transferred to the intensive care unit for close monitoring. He was transferred back to the gastroenterology service 3 days later.

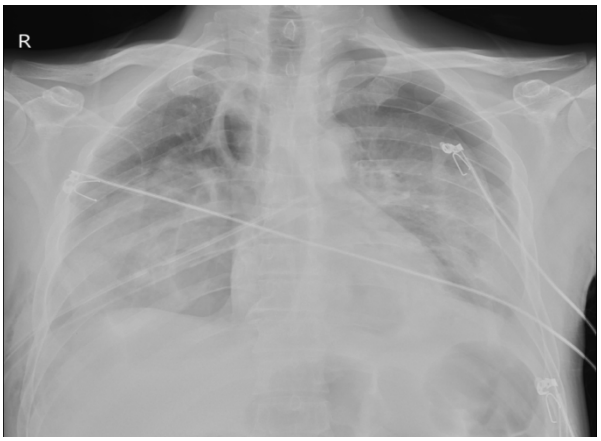

Figure 2. Chest X-ray: Trachea deviated to the right and pneumothorax line in the left lung

Boerhaave Syndrome was suspected because the patient's complaints such as shortness of breath and hypotension began after repeated retching. Exudative fluid, low pH 6.9, normal lipase/amylase and neutrophilic

leukocytosis were detected in the pleural fluid sample previously sent to the laboratory. The growth of polymicrobial oral flora bacteria in the pleural fluid culture further increased the suspicion of the diagnosis of Boerhaave syndrome. Extraluminal passage of contrast material to the right side of the pleural cavity was detected in the esophagram with peroral contrast material; Thus, he was diagnosed with Boerhaave syndrome (Figure 3).

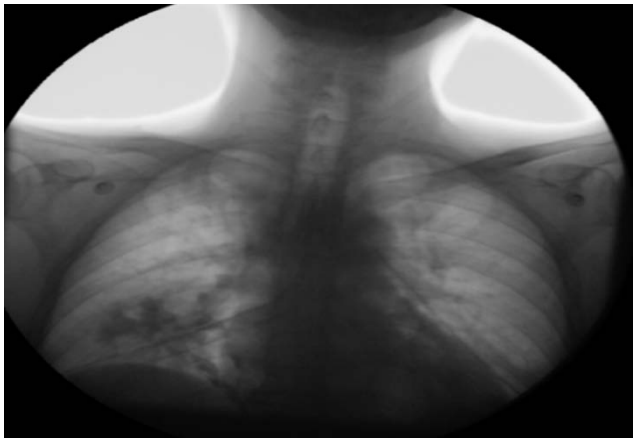

Figure 3. Cine Esophagram: Extraluminal passage of contrast material from the esophagus to the right side of the pleural cavity

The decision to place a metallic stent in the area of esophageal rupture was made by multidisciplinary consensus; A fully covered 10 cm metal stent was inserted to the ruptured area under general anesthesia at the endoscopy room (Figure 4). A second cine esophagram was requested and no extraluminal contrast material passage was observed.

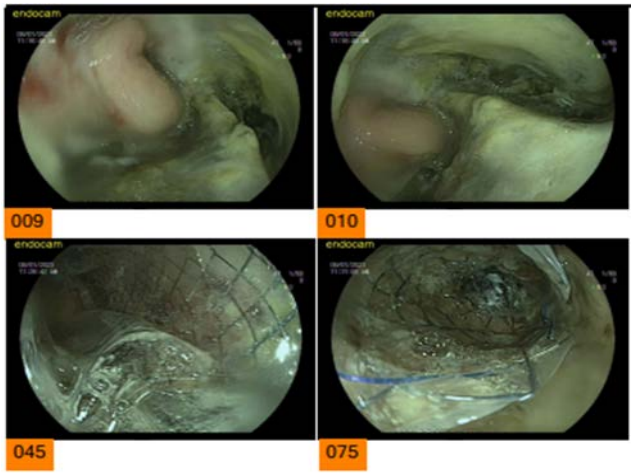

Figure 4. Placing a 10 cm diameter metal stent in the ruptured area via endoscopy

In the second week of admission, the patient developed hematochezia as well as hypotension; However, the source of bleeding could not be determined by gastroscopy and colonoscopy. The patient who did not respond to fluid replacement therapy was considered to have hemorrhagic shock; Therefore, he underwent emergency laparotomy and double balloon enteroscopy was performed in the same session. A Dieulafoy lesion with active bleeding was revealed in the 4th segment of the duodenum and a hemoclip was applied on it (Figure 5).

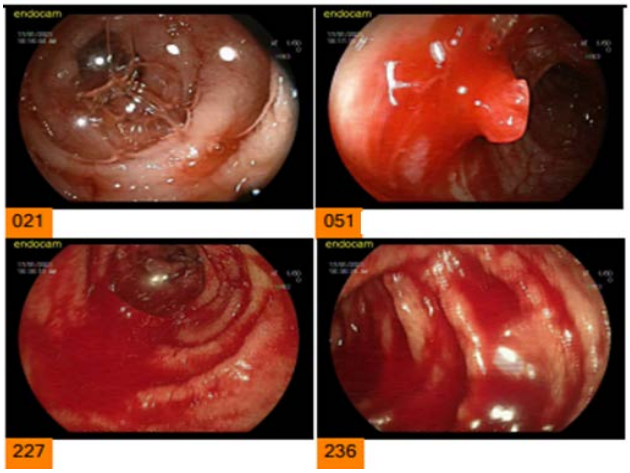

Figure 5. Double-Balloon Enteroscopy: Dieulafoy's lesion at the 4th part of the duodenum

The patient, whose pleural fluid culture after esophageal rupture was contaminated with many oral flora bacteria (Streptococcus mitis, Streptococcus salivarius, Neisseria subflava and Streptococcus vestibularis), was evaluated as mediastinitis and was followed up with a bilateral thorax tube. He was also treated with broad-spectrum antibiotics for weeks; he ultimately died of sepsis due to mediastinitis.

3. Discussion

MEN syndromes have diverse clinical presentations depending on the type and affected glands. The prevalence of MEN1 syndrome is approximately 1 in 20,000 to 40,000 people; Additionally, 10% of patients are the first person affected in the family [6].

Although genetic tests with high accuracy have been developed for diagnosis, they are not always affordable and accessible; Therefore, family history is important to make a diagnosis. Most patients are diagnosed with MEN1 Syndrome in middle age; nevertheless, studies show that tumors commence to develop at the age of 5 [6]. The patient's symptoms started 2 years ago and he was diagnosed with MEN syndrome at the age of 47. The patient was hospitalized multiple times due to recurrent acute pancreatitis. Elevated amylase/lipase levels and suspicious abdominal pain were always present in these admissions. While investigating the etiology of these pancreatitis attacks, a pancreatic NET (gastrinoma) and a parathyroid adenoma were detected. He had hyperparathyroidism, but his serum calcium and phosphorus levels were within normal limits. Further investigations of MEN1 syndrome, including Somatostatin Receptor PET/CT, pancreatic rebiopsy via endoscopic ultrasonography, hypophysis mri and genetic testing could not be completed as the patient's condition was never stable.

Boerhaave Syndrome usually occurs after repeated vomiting; On the other hand, our patient had retching instead of vomiting, and the first symptoms were shortness of breath, tachycardia and suspicious generalized abdominal pain. Three scans were performed within the first 24 hours after admission, and to our

surprise the evolving findings varied: no abnormality on thoracoabdominal CT, massive hydrothorax on thoracoabdominal CT angiography, and tension pneumothorax on chest X-ray. Esophagogram revealed passage of contrast medium into the right pleural space, which was rare as rupture of the left side of the distal esophagus occurred in almost 80% of cases [5]. Although primary surgical repair was recommended rather than endoscopic stent placement, it was decided to place a stent at the multidisciplinary meeting [9]. Endoscopic stent placement was performed without complications and the tear in the esophagus was successfully closed.

The most common site of Dieulafoy's lesion is the stomach, and it is seen in the gastroesophageal junction region in 71% of patients. These lesions are rarely seen outside the stomach, and the second most common place in patients is the duodenum with 15% [10]. In our case, the Dieulafoy lesion was located in the 4th part of the duodenum and manifested itself as hemorrhagic shock with hematochezia. Although the duodenum is the second most common site of Dieulafoy's lesion, a lesion in the 4th part of the duodenum as in our case is rare and it is challenging because routine gastroduodenoscopy cannot reach this area. We assume that this rare lesion and its uncommon site of location are related to the gastrinoma, which has been identified as a risk factor for Dieulafoy's lesion. Most of the patients with Dieulafoy's lesion are successfully treated with endoscopic intervention but some of them require operation to control the bleeding [10]. In our case, the patient underwent laparotomy and double balloon enteroscopy in the same session. Since the bleeding was successfully stopped with a hemoclip, no surgical intervention was required.

There are few case reports in the literature about Boerhaave syndrome occurring with tension hydropneumothorax; However, none of the cases are associated with MEN syndrome as in our case [11,12,13,14].

The key to successfully managing Boerhaave syndrome is early diagnosis and treatment; Therefore, high clinical suspicion may reduce the serious morbidity and mortality associated with Boerhaave Syndrome. If tension hydropneumothorax is detected on chest X-ray or CT, Boerhaave syndrome should be investigated. The most important risk factor for Boerhaave syndrome in our case was pancreatic gastrinoma due to MEN1 syndrome, which causes peptic ulcer and nausea.

We describe the first case of MEN Syndrome type 1 in which Boerhaave syndrome presented as tension hydropneumothorax.

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None declared.

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