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CASE REPORT

Boerhaave Syndrome Masquerading as Sudden Death: A Virtual Autopsy Perspective

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ABSTRACT

Boerhaave syndrome (BS) is a rare, life-threatening condition characterized by spontaneous esophageal rupture, commonly triggered by forceful vomiting. Due to its nonspecific presentation, BS is often misdiagnosed, leading to delayed intervention and high mortality. We report a case of BS presenting with hematemesis, respiratory distress, and collapse, emphasizing the forensic implications and the role of postmortem computed tomography (PMCT) in virtual autopsy. A 46-year-old male with a history of alcohol consumption experienced sudden-onset hematemesis and respiratory distress, followed by cardiopulmonary arrest. Despite resuscitation efforts, the patient was pronounced dead. Autopsy findings revealed a full-thickness esophageal rupture with mediastinal contamination and pleural effusions, confirming BS as the cause of death. PMCT successfully detected pneumomediastinum, free intrathoracic air, and mediastinal fluid, demonstrating its utility in forensic investigations. The case highlights key differential diagnoses of sudden death, including acute myocardial infarction, pulmonary embolism, and aortic dissection. This case aligns with existing literature where hematemesis, though less commonly reported, was a significant symptom preceding cardiovascular collapse. Delayed diagnosis of BS remains a critical factor contributing to poor outcomes and increased mortality rates. The integration of PMCT with conventional autopsy enhances the diagnostic accuracy of esophageal perforation-related deaths, particularly in forensic settings. Comparative analysis with previously reported cases underscores the need for heightened clinical suspicion and early imaging to prevent fatal outcomes. Boerhaave syndrome remains a diagnostic and therapeutic challenge, often leading to sudden and unexpected death. This case reinforces the medicolegal importance of virtual autopsy in identifying esophageal rupture, particularly in unwitnessed or unexplained deaths.

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Increased awareness of BS, combined with advanced imaging techniques, can aid in timely diagnosis, reducing misclassification in forensic pathology.

KEYWORDS

• Boerhaave's syndrome • Spontaneous esophageal rupture • Sudden death • Mediastinitis • Postmortem computed tomography (PMCT) • Hemorrhagic Shock • Hematemesis Introduction

Boerhaave syndrome (BS) is a rare but life-threatening condition characterized by a spontaneous, full-thickness rupture of the esophagus most commonly at the lower oesophagus, typically occurring due to a sudden increase in intraesophageally pressure following forceful vomiting or retching.[1] Current estimates suggest a global incidence of approximately 3.1 cases per million individuals annually worldwide.[2] The rare occurrence of this emergency limits physicians in gaining adequate clinical experience and makes it difficult to establish strong scientific evidence to guide patient management and clinical decision-making.[3] The classical clinical presentation, known as Mackler's triad vomiting, chest pain, and subcutaneous emphysema is observed in only 50% of cases, making early detection challenging.[4] Boerhaave syndrome (BS) is of significant forensic importance due to its potential to cause sudden and unexpected death, often leading to medicolegal investigations. The condition is frequently misdiagnosed because its symptoms mimic other fatal conditions, such as myocardial infarction, pulmonary embolism, or perforated peptic ulcer, delaying medical intervention and increasing mortality This syndrome represents the most severe form of esophageal perforation, often leading to mediastinitis, sepsis, and multi-organ failure if not promptly diagnosed and treated.[5] Postmortem computed tomography (PMCT) has emerged as a valuable tool in forensic investigations of BS. It can detect pneumomediastinum, subcutaneous emphysema, pleural effusion, and mediastinal contamination, providing critical diagnostic clues before a full autopsy is performed.[6]

We present a compelling case of Boerhaave syndrome (BS), where the patient suffered from hematemesis and acute respiratory distress, followed by a sudden and fatal collapse. This case underscores the diagnostic challenges associated with BS and highlights the critical role of virtual autopsy (postmortem computed tomography, PMCT) in forensic investigations. The integration of PMCT with traditional autopsy findings enhances diagnostic accuracy, providing invaluable insights into the cause of death, underlying pathology, and forensic implications.

CASE PRESENTATION

A middle age man 46-year-old with a known history of chronic alcoholism was brought to the emergency department of Pt. Madan Mohan Malaviya Hospital, Delhi, in an unconscious and unresponsive state. The patient had reportedly experienced hematemesis two hours prior and had complained of respiratory distress for the past three days. On examination, blood was observed oozing from the oral cavity, breath sounds were absent, and bilateral pupils were fixed, dilated, and non-reactive to light. Vital signs were unrecordable, and the patient was declared dead on arrival. The body was subsequently transferred to the Department of Forensic Medicine at AIIMS, Delhi, for a medicolegal autopsy to ascertain the cause of death.

Postmortem Examination

On External Examination the deceased exhibited generalized yellowish discoloration of the skin. Rigor mortis was well-developed, and postmortem lividity was fixed over the back and dependent regions. The conjunctivae were icteric, and sub-conjunctival hemorrhage was noted in the left eye. Approximately 100 mL of blood-tinged fluid was present in the oral cavity. The fingernail beds were pale, and blood-stained fecal matter was observed around the anal orifice. No external antemortem injuries were detected.

The postmortem computed tomography (PMCT) was conducted using an Aquilion Lightning TSX-035A, a 16-slice multi-detector computed tomography (MDCT) spiral scanner (Toshiba America Medical Systems, Tustin, CA), with scan parameters of 120 KVp, auto mA, and exposure of 52 mAs. Image acquisition

was performed with a slice thickness of 1 mm and a pitch factor of 0.8, utilizing a 16 × 1 mm collimation. The images were evaluated using standard window settings on Vitrea Software version 7.10.1.20. The scan revealed a periesophageal densification by heterogeneous content measuring 62.9mm × 37.6mm in coronal plane (*Figure* 1A). Same lesion in sagittal plane is located in the upper left superior mediastinum i.e. above the line of Ludwig *Figure* 1B. The lesion exhibited varying Hounsfield unit (HU) values ranging from +25 to -972, consistent with mediastinal contamination due to esophageal rupture (*Figure* 1C).

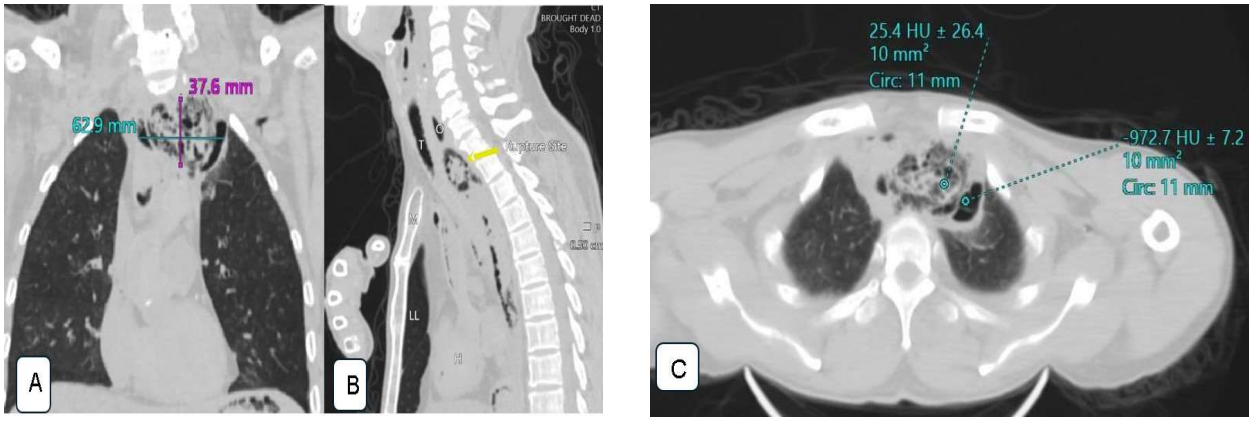
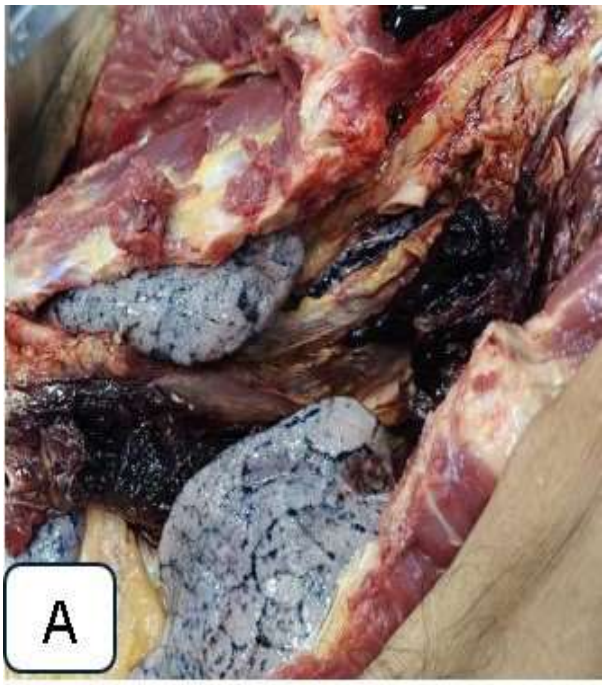


Figure 1: Postmortem computed tomography findings, **(A)** Coronal section of PMCT showing periesophageal densification by heterogeneous content measuring 62.9mm × 37.6mm (Figure 1A) in the upper left superior mediastinum, **(B)** Sagittal section showing the rupture site above the line of Ludwig with infiltration in surrounding (marked with yellow arrow), with anatomical landmark i.e. M- Manubrium, LL- Left Lung, T-Trachea, O-Esophagus and H-Heart, **(C)** Axial section showing pneumomediastinum with Hounsfield value (HU) values ranging from +25 to - 972, consistent with mediastinal contamination due to esophageal rupture

On internal examination, the dura mater and leptomeninges were intact, and the brain was edematous with diffuse congestion. Organ dissection using Letulle's method revealed a 5 cm long transmural rupture on the left posterior aspect of the lower one-third of the esophagus. Blood mixed with gastric contents was found leaking into the left mediastinum, with approximately 200 mL of accumulated material (*Figure* 2A). The stomach contained 1.5 L of blood-tinged partially digested food, with similar material present in the trachea extending to the primary bronchi (*Figure* 2B). The lungs were pale, adhered to the chest wall, and devoid of cyanosis. The pericardial sac was intact with 10 mL of straw-colored fluid. The coronary arteries exhibited calcifications but remained patent throughout. The heart chambers and valves appeared normal. The liver was markedly enlarged (1850g), with a shiny surface, nodular texture, and diffuse yellow discoloration suggestive of chronic alcohol-induced hepatic changes (Figure 2C). The kidneys were pale with indistinct cortico-medullary differentiation. Other organs were within normal weight limits.

The cause of death was determined as hemorrhagic shock due to esophageal rupture, with an estimated postmortem interval of 8–12 hours.



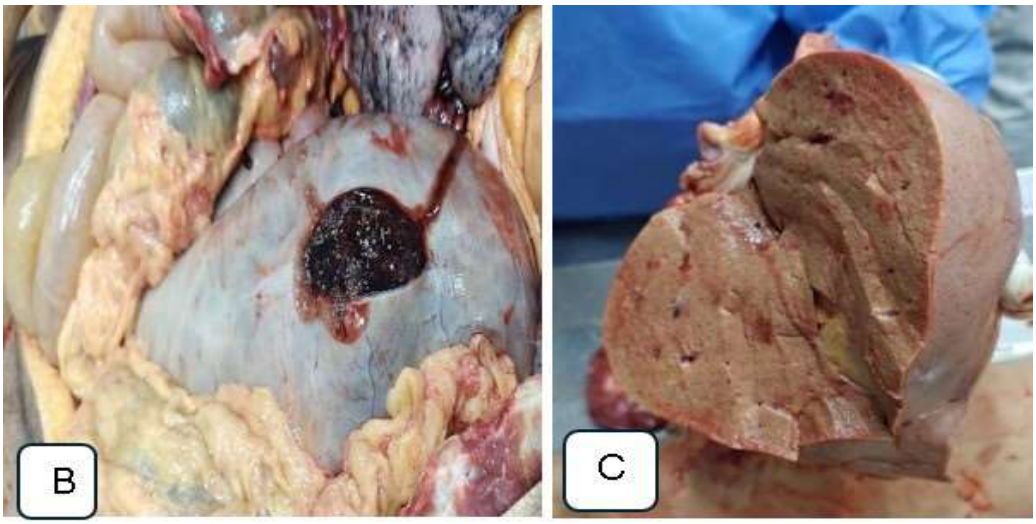


Figure 2: Gross internal examination, **(A)** Infiltration of site with looted blood in surrounding tissue at the esophageal rupture site, **(B)** The stomach is filled with approximately 1.5 L of blood-tinged, partially digested food due to bleeding from an esophageal rupture, **(C)** Enlarged liver (1850 g), with a shiny surface, nodular texture, and diffuse yellow discoloration

DISCUSSION

Boerhaave syndrome (BS) is a rare but fatal condition characterized by a spontaneous, full-thickness rupture of the esophagus, typically triggered by forceful vomiting or retching. The condition remains a diagnostic challenge due to its nonspecific presentation, leading to delayed recognition and increased mortality. The present case, where the patient experienced hematemesis and respiratory distress before a sudden collapse, aligns with existing literature emphasizing the critical nature of early diagnosis and intervention.[5,7]

Boerhaave's syndrome (BS) is frequently underdiagnosed due to its nonspecific clinical presentation and rapid deterioration, with the diagnosis often being established only at autopsy, as in our case. This diagnostic challenge has been highlighted in population-based studies as well. Vidarsdotti et al. reported an age-standardised incidence of oesophageal perforation of 3.1 per 1,000,000 per year in Iceland, and found that in 17% of cases (5 out of 29 patients), the diagnosis was missed during life and first made at autopsy.[2] Our findings are consistent with this observation, emphasizing that oesophageal rupture remains a condition prone to misdiagnosis, often discovered only on postmortem examination.

Boerhaave's syndrome (BS) is classically described by Mackler's triad of vomiting, chest pain, and subcutaneous emphysema; however, this triad is often incomplete and the condition may present atypically, making diagnosis difficult. Atypical presentations have been documented in the literature, such as the case reported by Saha et al., where a 54-year-old woman with Boerhaave's perforation presented with Enterococcal bacterial pericardial effusion rather than classical thoracic symptoms.[8] Similarly, our case was also atypical, as the patient presented with hematemesis as a prominent symptom, a feature less commonly reported in BS but seen in severe cases with intramural vascular injury. Such atypical manifestations pose a significant diagnostic challenge, often leading to delayed or missed recognition during life. In this context, our findings reinforce the importance of considering BS in the differential diagnosis even when classical features are absent, and highlight the value of imaging and autopsy in uncovering these unusual presentations.

Postmortem computed tomography (PMCT) is increasingly recognized as a valuable adjunct in forensic investigations, particularly for detecting thoracic injuries, pneumomediastinum, and mediastinal contamination in cases of Boerhaave's syndrome (BS).[7] In the present case, PMCT clearly demonstrated periesophageal densification with heterogeneous content and pneumomediastinum, findings that strongly suggested esophageal rupture even prior to conventional autopsy. These observations are

in line with those reported by Ribeiro TA et al., who described similar CT thorax findings of Boerhaave's syndrome in an elderly male patient.[6]

CONCLUSION

This case underscores the importance of considering BS in forensic investigations of sudden deaths, especially in individuals with a history of alcohol use and recent episodes of forceful vomiting. It highlights the forensic and clinical challenges associated with BS, emphasizing the need for heightened clinical suspicion, early imaging, and multimodal diagnostic approaches, including virtual autopsy. The integration of PMCT with conventional autopsy significantly aids in diagnosing esophageal rupture, preventing misclassification of sudden deaths. Increased awareness of BS in forensic pathology can facilitate earlier recognition, reduce diagnostic errors, and improve medicolegal investigations.

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