Early Identification and Treatment of Perforated Appendiceal Mucormycosis via cfDNA biomarker panel

Julian Casabar MD[1], Thomas Abbruzzese MD[1], Ji Fan MD[2]
[1]Department of General Surgery, HCA Healthcare / USF Morsani College of Medicine GME: HCA Florida Brandon Hospital, Brandon, FL
[2]Department of Gastrointestinal Oncology, Moffitt Cancer Center, Tampa FL.

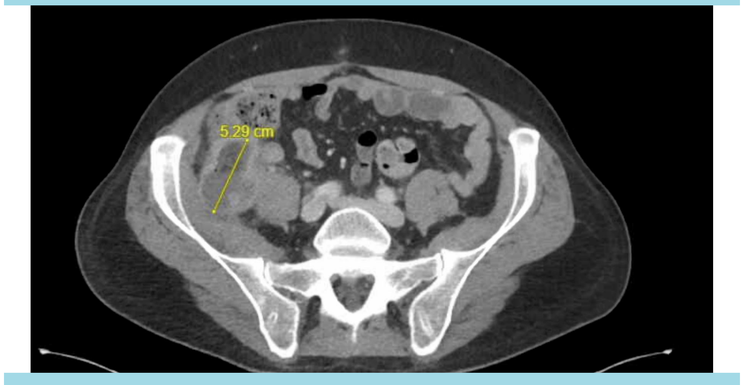
Abstract

Mucormycosis is a rare life-threatening opportunistic fungal infection typically seen in immunocompromised patients. Appendiceal mucormycosis is an uncommon presentation of an already rare infection. This can be mistaken clinically for bacterial appendicitis or typhlitis in this patient population. The mortality of mucormycosis reaches 95% once the infection disseminates from its primary site. The mainstay of treatment is early identification, early initiation of antifungals, and prompt surgical debridement. Histopathology is the gold standard for diagnosis. However, this is not always available given that patients in this population are typically poor surgical candidates.

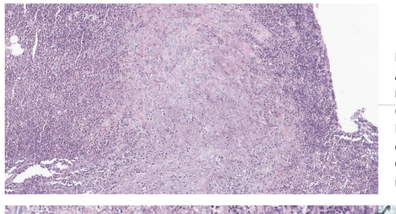
Case Presentation

We present the case of a 55-year-old male with recently diagnosed acute myelogenous leukemia (AML) who underwent two lines of chemotherapy and developed severe pancytopenia, fevers, and abdominal pain. He was found on CT imaging to have perforated appendicitis with an associated 5 cm abscess. Due to his pancytopenia, we initially elected to take a conservative approach with antimicrobials and percutaneous drainage believing this was of a bacterial etiology. However, a cell-free DNA (cfDNA) biomarker panel (Karius) identified an active mucormycosis infection. Early identification prompted early initiation of antifungal medications and necessitated an emergency trip to the the operating room for an interval appendectomy. He successfully underwent his surgery without evidence of disseminated disease, completed a perioperative course of antifungals, and fully recovered from his infection.

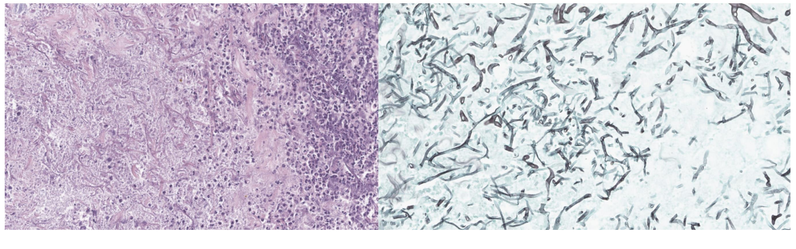
Imaging



Histology



Periappendiceal abscess.
A. Low power showing fibrinopurulent exudate with central necrosis. Fungal organisms are seen at this magnification. (Hematoxylin and eosin, 8X).
B. Abundant acute inflammation and necrosis with fungal organisms. (Hematoxylin and eosin, 20X).
C. Broad, ribbon-like hyphae with few septae. (Grocott methenamine silver, 20X)



Discussion

While bacterial appendicitis remains the most common cause of appendicitis, there are other rare opportunistic infections to consider that affect the immunocompromised patient population. Diagnosis of a mucormycosis is difficult because the gold standard for diagnosis is a surgical specimen. However, these patients are often poor surgical candidates due to pancytopenia, malnutrition, or active treatment with chemotherapy, making immediate appendectomy high risk.

In this case, we demonstrate how a cfDNA panel (Karius) was a useful adjunct to guide medical and surgical management of this patient. While perforated appendicitis with a large abscess is typically managed with percutaneous drainage and antibiotics, the positive Rhizopus results prompted early initiation of antifungals and emergent appendectomy with debridement which ultimately saved this man's life.

Citations

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