Introduction: Chronic granulomatous disease (CGD) is an inborn error of immunity (IEI). The CYBB gene is responsible for the most frequent genetic forms of the CGD. The affected cells have an impaired ability to generate reactive oxygen species (ROS), thus failing to kill intracellular bacteria and fungi, and excessive granulomatous inflammation. Fungal pathogens include, notably, the Aspergillus fumigatus complex, Aspergillus nidulans and Candida spp.; however, other genders have also been described. We describe two unrelated CGD patients with pulmonary infections due to Geotrichum spp., an emerging and opportunistic pathogen.

CASE 1. A 9-year-old male at 3-years-of-age, he suffered a cervical abscess and received intravenous antibiotics. His admission to the hospital was necessary due to a history of cough, fever, and dyspnea with progressive respiratory failure, which required mechanical ventilation and admission to the ICU.

Chest X-ray and CT scan images showed diffuse interstitial thickening in the lung parenchyma, multiple intrathoracic lymph node enlargements, pneumomediastinum, and subcutaneous emphysema, which suggested a fungal etiology; therefore, liposomal amphotericin B (L-AmB) was added empirically.

(Figure). Pseudohyphae and arthroconidia were detected in cultures of bronchoalveolar lavage (BAL) samples (Figure), and Geotrichum capitatum was identified with MALDI-TOF Mass Spectrometry. A mutation was found in the CYBB through next-generation sequencing (p.P383L/y), confirming CGD diagnosis. L-AmB was suspended after six weeks due to a good clinical response, and itraconazole was changed to a prophylactic dose.

Figure 1. A. Chest X-ray (case-1) with hilar and parenchyma interstitial infiltrates, multiple calcifications in axillary right margin (arrowhead). B. Thorax CT scan (case-1) with bronchiectasis, parenchyma infiltrates, pneumomediastinum (arrowhead), and subcutaneous emphysema (arrowhead). C. Cream-colored, dry and wrinkled yeast colonies of Geotrichum capitatum on Sabouraud agar. D. Microscopic examination of bronchoalveolar lavage culture showing the presence of arthroconidia, lactophenol cotton blue stain, 40 X magnification.

CASE 2. The second case is a 19-year-old male urban resident who was previously diagnosed with CGD secondary to a pathogenic variant in the CYBB gene (p.H115Q/y) at 14 years old.

He had a history of inflammatory bowel disease and multiple pneumonias, one of which was diagnosed as tuberculosis. The young patient had poor adherence to prophylactic antimicrobial treatment.

He arrived at the emergency room with myalgia, malaise, and dyspnea; the physical examination revealed fever, tachycardia, and tachypnea, and the oxygen saturation was low (SpO2 70%). Parenchymal nodules suggestive of pneumonia were visualized through thorax CT scan, and the administration of meropenem and vancomycin was initiated. In the first 24 hours, the patient required mechanical ventilation due to refractory hypoxemia; during the bronchoscopy sample-BAL culture was collected.

On the 6th day in the absence of pneumonia improvement (Figure 2), empirical amphotericin B (AmB) was initiated. On the 38th day, Geotrichum spp. grew from the admission BAL culture. AmB was replaced with voriconazole and administered for 12 weeks with a favourable response (Figure 2). Two years later, he continued to be monitored at the hospital as an outpatient with prophylactic treatment.

Figure 2. A. Thorax CT scan (case-2) showing bronchiectasis and parenchyma interstitial infiltrates. B. Thorax CT scan (case-2). Evident changes images accord to clinical improvement pneumonia.

Conclusion: Geotrichum capitatum, (also known as Magnusiomyces capitatus) is known to cause disseminated opportunistic infections, especially in neutropenic patients with haematologic malignancies. G. capitatum, however, is rarely reported as a pathogen in the literature. Erman et al. reported an adolescent with CARD 9 deficiency (an IEI) who presented with G. capitatum cholangitis. Geotrichum spp. have not been previously reported in CGD patients. The occurrence of geotrichosis should raise the suspicion of an underlying IEI; We described two patients with CGD presenting with geotrichosis, an emerging and opportunistic pathogen associated in this disease.