ANSWER TO PHOTO QUIZ (PAGE 259)

AN UNUSUAL CAUSE OF HYPERAMMONAEMIA

DIAGNOSIS

In adults, unrecognised end-stage liver disease is the most likely cause of high ammonia levels.[1] In this case liver function tests, abdominal ultrasound and CT scan ruled out the presence of liver cirrhosis and portal flow anomalies. Our patient was not using any drugs that might interfere with the urea cycle, such as valproate, which inhibits the synthesis of N-acetylglutamate, a cofactor that promotes the first step in the detoxification of ammonia.[1] A normal amino acid profiling and normal urine orotic acid levels render a defect in the urea cycle unlikely.

Infections with urea-producing bacteria, often present in complex urinary tract infections, may cause hyperammonaemia by hydrolysis of urea into carbon dioxide and ammonia.[1] Patients with an IL-12/23Rb1-deficiency are, however, particularly prone to develop atypical mycobacterial infections because of an inability to produce interferon gamma.[2] As a consequence, we suspected an atypical mycobacterium to be the cause of the hyperammonaemic encephalopathy in this patient. By means of FDG PET-CT the infection was localised in the bone marrow which, in retrospect, explains the pancytopenia. Targeted bone marrow biopsy, showing positive PCRs and Ziehl-Neelsen stains, confirmed the presence of atypical mycobacteria. Cultures ultimately showed *Mycobacterium genavense* (*figure 2*). Although this infection has been reported in patients who are immunocompromised (e.g. after solid organ transplantation), there has been only one published case of *M. genavense* presenting with hyperammonaemia.[3] Of interest, previous studies have shown that this mycobacterium has urease activity,[4] which renders this microorganism the most likely cause of the hyperammonaemia in the present case.

During the diagnostic workup, the patient was treated with interferon gamma. Furthermore, treatment of hyperammonaemia was initiated by administering lactulose, rifaximin, continuous infusion of dextrose 10% solution, restriction of dietary protein intake, and eventually continuous veno-venous haemofiltration. Once the diagnosis was confirmed triple therapy with rifabutin, isoniazid and clarithromycin was started.

Despite the early start with ammonia-lowering therapy and the initial improvement of laboratory parameters, the patient's consciousness fluctuated during admission. Unfortunately, he eventually died from a disseminated infection.

DISCLOSURES

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