Diagnostic ileoscopy assisting in the diagnosis of Whipple’s disease

A 70-year-old white man had been aware of mild memory loss, arthralgia, and altered bowel habits for 3 years prior to his referral. His gastrointestinal symptoms had worsened during the 2-month period preceding his referral, with diarrhea (five to six bowel movements of fatty stools per day) and a 4-kg weight loss. His laboratory workup, including thyroid function and human immunodeficiency virus (HIV) screening, was unremarkable. Colonoscopy showed normal colonic mucosa but ileoscopy revealed a diffuse brown appearance of the mucosa with scattered white plaques (Fig. 1).

Biopsy specimens taken from the terminal ileum revealed foamy macrophages in the lamina propria that stained positively with the periodic acid–Schiff (PAS) reaction. A negative acid-fast stain excluded Mycobacterium avium complex (MAC) infection. The duodenum appeared normal and biopsies showed no abnormalities. Treatment with trimethoprim–sulfamethoxazole for 1 year was instituted with prompt symptom resolution and 12-kg weight gain after 6 months.

Whipple’s disease is a rare chronic multisystem infection caused by Tropheryma whippelii. PAS-positive small-bowel biopsy specimens obtained during upper gastrointestinal endoscopy are usually diagnostic in the appropriate clinical setting, although polymerase chain reaction (PCR) tissue analysis is increasingly being used [1]. The reported small-bowel endoscopic features include edema and brown discoloration of the mucosa, erythematous spots, and subepithelial hemorrhages [2], which seem to disappear distally, usually in a continuous fashion [3]. Concomitant endoscopic disease of the terminal ileum has also been reported [4, 5] but to our knowledge this is the first description of endoscopic findings isolated to the terminal ileum and biopsy findings diagnostic of Whipple’s disease.

Full imaging of the small bowel (including capsule endoscopy) was not performed due to institutional constraints but we found no other evidence of disease at upper or lower gastrointestinal endoscopy even after biopsy sampling of the duodenum and colon. In the future, colonoscopy with ileoscopy should be considered in the diagnostic workup of patients suspected of having Whipple’s disease.

Competing interests: None

J. Dinis Silva1, V. Fernandes2, M. C. Alves3, J. Freitas2

1 Department of Gastroenterology, Hospital do Espírito Santo de Évora EPE, Largo Senhor da Pobreza, Portugal
2 Department of Gastroenterology, Hospital Garcia de Orta EPE, Almada, Portugal
3 Department of Histopathology, Laboratório Joaquim Chaves, Lisbon, Portugal
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Corresponding author
J. Dinis Silva, MD
Serviço de Gastroenterologia
Hospital do Espírito Santo de Évora EPE
Largo Senhor da Pobreza
7000-811 Évora
Portugal
Fax: +351-266-740117
jdinisilva@gmail.com