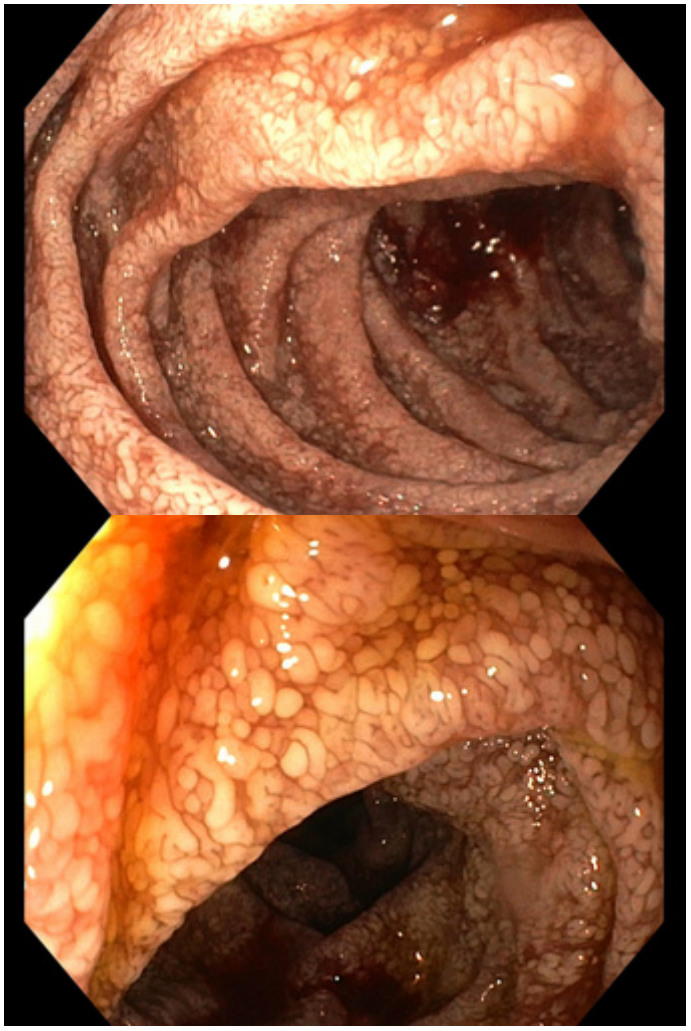
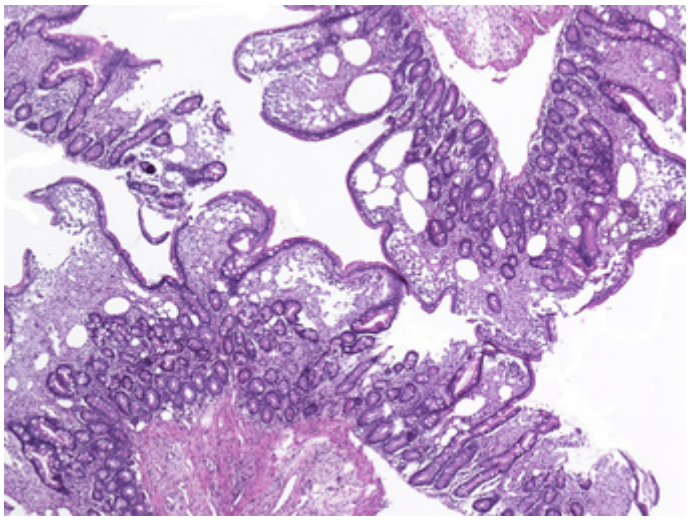
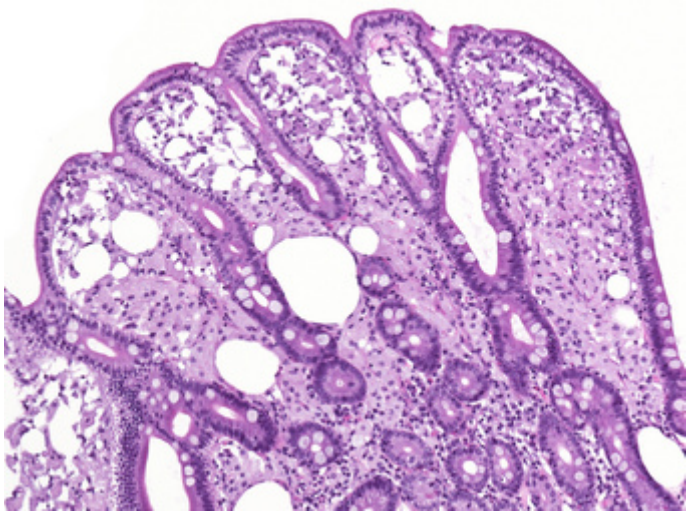
October 2017

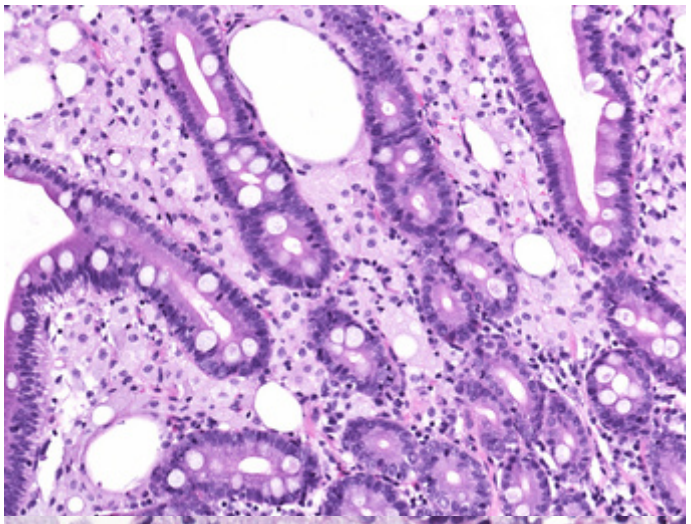
68-year-old male with chronic diarrhoea, malabsorption and weight loss.

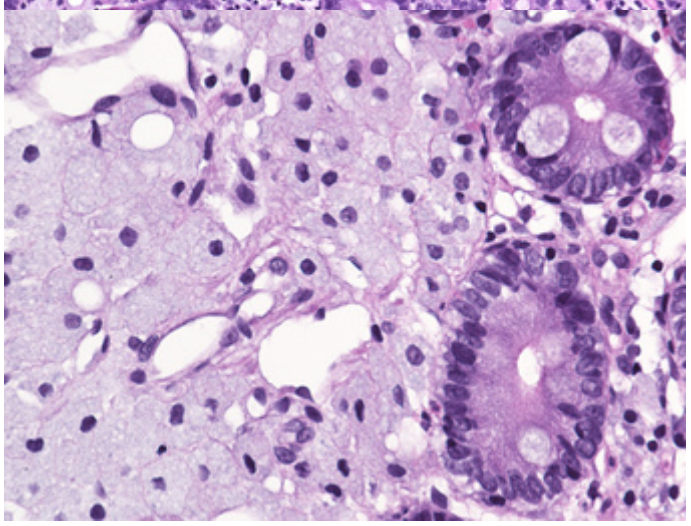
What is your diagnosis?

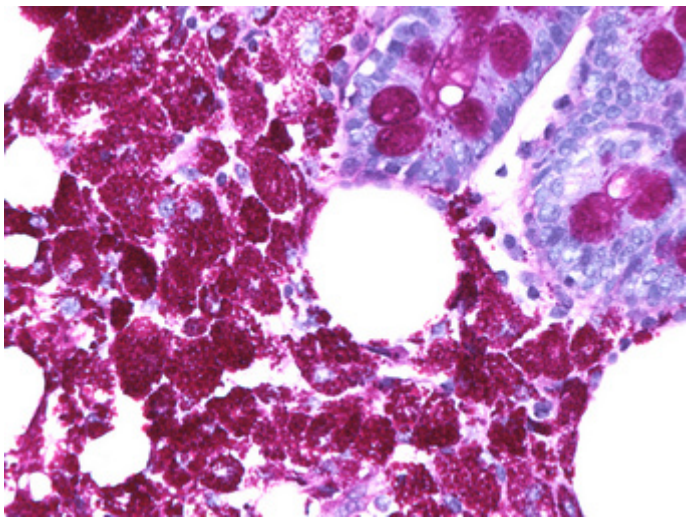












Diagnosis:

Whipple's disease.

Comment:

A 68-year-old male patient presented with a history of chronic diarrhoea (24 months) and significant weight loss (15 kg). Upper gastrointestinal endoscopy was performed and showed thickened villi with ectatic lymph vessels and marked mucosal oedema within the duodenum (Panels A-B). Stomach and oesophagus were unremarkable. Multiple biopsies were taken.

The histological picture of the duodenal mucosa was characterized by marked lymphangiectasia and prominent infiltration of the lamina propria with foamy macrophages (Panels C-F). The macrophages were strongly positive on PAS stain, which demonstrated a granular pattern and also decorated macrophages within the superficially sampled submucosa (Panel G). Polymerase chain reaction (PCR) confirmed the presence of Tropheryma whipplei and diagnosis of Whipple's disease was made.

Whipple's disease is a rare chronic systemic infectious disorder, affecting multiple organs and systems, such as small bowel, cardiovascular and central nervous system as well as joints. The disease shows strong male predominance (about 85% of patients) and occurs mostly in 40 to 50-year-old individuals.

The disease is caused by Tropheryma whipplei, which belongs to Gram-positive bacilli, supposed to be an obligatory intracellular pathogen. Bacteria are PAS-positive and negative on acid-fast-stain. Infection occurs in patients with impaired cellular immunity. Unfortunately, the major manifestations are rather nonspecific, which makes Whipple's disease difficult to diagnose. Symptoms include weight loss, diarrhoea with different features of malabsorption, abdominal pain, lymphadenopathy, fever and often arthritis. Although digestive tract involvement is observed in the majority of cases, atypical cases have been described lacking of gastrointestinal symptoms. Interestingly, these patients often present with neurologic symptoms and also endocarditis or uveitis may occur.

Without treatment the infection tends to be lethal. Most patients recover under antibiotic therapy. However, relapse may occur even years after treatment and CNS symptoms may be the first sign of recurrence.

For further reading:

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