EDITOR'S QUIZ: GI SNAPSHOT

When does severe diarrhoea disclose a hereditary disease?

Robin Spiller, Editor

A 30-year-old man of Mediterranean origin was admitted for intractable diarrhoea and severe malabsorption. He presented with a 2 month history of frequent watery stools and acute weight loss of 10 kg. He is of Mediterranean origin. His medical history included episodic diffuse abdominal pain of sudden onset, severe intensity and short duration without diarrhoea. He did not take any drugs. His clinical evaluation showed fatigue, muscle weakness and paraesthesias. The arterial blood pressure was 90/70 mm Hg and the heart rate was 120 beats/min. He had microcytic iron deficiency anaemia. His haemoglobin concentration was 8 g/dl, mean corpuscular volume 65 fl, serum iron 10 µmol/l and serum ferritin 15 µg/l. He had increased C reactive protein of 250 mg/l. Blood tests showed decreased levels of albumin, calcium and magnesium, but normal prothrombin time. The stool cultures were sterile. Electrocardiogram, plain abdominal film and chest x ray examination were normal. Parenteral alimentation was started. A gastroscopy was performed with duodenal biopsies that showed no mucosal lesions and no villous atrophy or infectious colonisation. Serum immunoglobulin A anti endomysium was negative. A colonoscopy with ileoscopy was performed (figs 1, 2). Biopsy specimens were taken from the lesions (figs 3, 4).

Figure 1 The ileocaecal valve.

Figure 2 Right colon.

Figure 3 Biopsy of the right colon.

Figure 4

Question

What do the colonoscopy and ileoscopy show and what is your differential diagnosis? Following biopsy, what is your final diagnosis and which blood test would you perform to confirm it?

See page 357 for answer

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