Double-balloon enteroscopy for the detection of diffuse small-bowel polypoid ganglioneuromatosis mimicking Crohn’s disease in a patient with von Recklinghausen disease

A 51-year-old woman with an 8-month history of recurrent episodes of abdominal pain, mild bloody diarrhea, and weight loss (6 kg in 3 months) was referred to our unit for evaluation of suspected Crohn’s disease. She had carried a diagnosis of type 1 neurofibromatosis for 30 years. Physical examination revealed multiple café-au-lait spots and multiple cutaneous neurofibromas. Her abdomen was mildly tender in the lower abdomen with no detectable palpable mass. Laboratory test results were as follows: hemoglobin level 9.6 g/dL, sedimentation rate 40 mm/h, C-reactive protein 23 mg/dL. Other biochemical tests were unremarkable. A colonoscopy revealed a normal-appearing colon and an edematous terminal ileum with a 1-cm pedunculated polyp covered by exudate (Fig. 1). A magnetic resonance enterography showed thickening of the jejunum and terminal ileum, and a pedunculated polyp, about 1 cm in diameter, located in the terminal ileum (Fig. 2). An oral double-balloon enteroscopy showed multiple, raspberry-like, 3–5-mm, sessile polyps, which were covered by faint exudates located in the proximal jejunum (Fig. 3). Biopsies of the polyps in the jejunum and ileum revealed intestinal ganglioneuromatosis.

Type 1 neurofibromatosis, also known as von Recklinghausen disease, may affect the gastrointestinal tract in 25% of patients in whom intestinal neurofibromas, gastrointestinal stromal tumors, or ganglioneuromatosis can be detected [1,2]. The disease may affect any part of the gastrointestinal tract. The most common symptoms are abdominal pain, change in bowel habit, diarrhea, and gastrointestinal bleeding, which resemble Crohn’s disease.

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