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]. Intestinal ganglioneuromatosis is a rare neoplastic condition characterized by proliferation of nerve ganglion cells, nerve fibers, and supporting cells of the enteric nervous system. It occurs in three forms: as an isolated polyp, as multiple polyps (ganglioneuromatous polyposis), and as diffuse involvement of the bowel wall (diffuse intestinal ganglioneuromatosis) [1]. 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.

Competing interests: None

Serta Kilincalp1, Mevlüt Hamamcı1, Hakan Akinci1, Şahin Çoban1, Aydın Şeref Köksal2, İlhami Yüksel2,3
1 Department of Gastroenterology, Diskapi Yıldırım Beyazıt Education and Research Hospital, Ankara, Turkey
2 Department of Gastroenterology, Yüksel İhtisas Training and Research Hospital, Ankara, Turkey
3 Department of Gastroenterology, Yıldırım Beyazıt University School of Medicine, Ankara, Turkey

Fig. 1 Colonoscopy showing an edematous terminal ileum with a pedunculated polyp (1 cm in size) covered by exudates.

Fig. 2 Magnetic resonance enterography showing neurofibromas on the skin (white arrow), thickening of the jejunum and terminal ileum, and a pedunculated polyp, about 1 cm in diameter, located in the terminal ileum (black arrow).

Fig. 3 Oral double-balloon enteroscopy showing multiple, raspberry-like, 3–5-mm, sessile polyps, which were covered by faint exudates located in the proximal jejunum.
References

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Corresponding author
Serta Kilincalp, MD
Department of Gastroenterology
Diskapi Yildirim Beyazit Education and Research Hospital
Altidag
06120, Ankara
Turkey
Fax: +90-312-3186690
serta80@gmail.com

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