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.

Competing interests: None

Serta Kilincalp¹, Mevlüt Hamamcı¹, Hakan Akinel¹, Şahin Çoban¹, Aydın Şeref Köksal², İlhami Yüksel²,³

1 Department of Gastroenterology, Diskapi Yildirim Beyazit Education and Research Hospital, Ankara, Turkey
2 Department of Gastroenterology, Yüksel Ihtisas Training and Research Hospital, Ankara, Turkey
3 Department of Gastroenterology, Yildirim Beyazit 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

Bibliography
DOI http://dx.doi.org/10.1055/s-0034-1377434
Endoscopy 2015; 47: E4–E5
© Georg Thieme Verlag KG Stuttgart · New York
ISSN 0013-726X

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