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.

Endoscopy_UCTN_Code_CCL_1AC_2AC

Competing interests: None

Serta Kilincalp1, Mevlüt Hamamcı1, Hakan Akinci1, Şahin Çoban1, Aydin Şeref Köksal2, İlhamı 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

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 Yıldırım Beyazıt Education and Research Hospital Altıdag 06120, Ankara Turkey Fax: +90-312-3186690 serta80@gmail.com