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

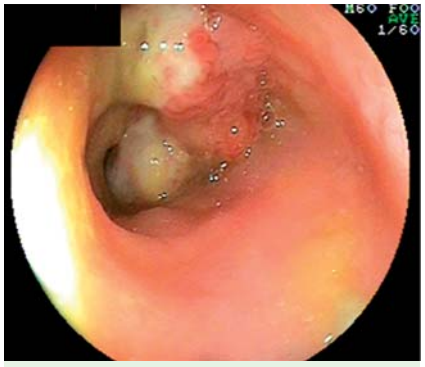

Fig. 1 Colonoscopy showing an edematous terminal ileum with a pedunculated polyp (1 cm in size) covered by exudates.

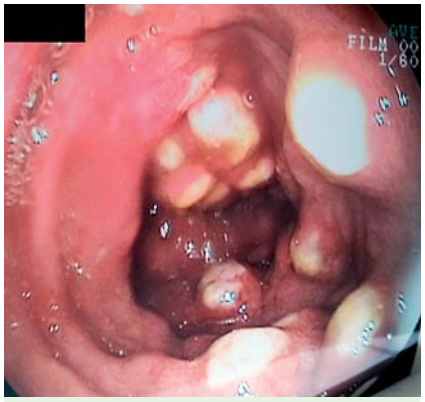

Fig. 3 Oral double-balloon enteroscopy showing multiple, raspberry-like, 3 – 5-mm, sessile polyps, which were covered by faint exudates located in the proximal jejunum.

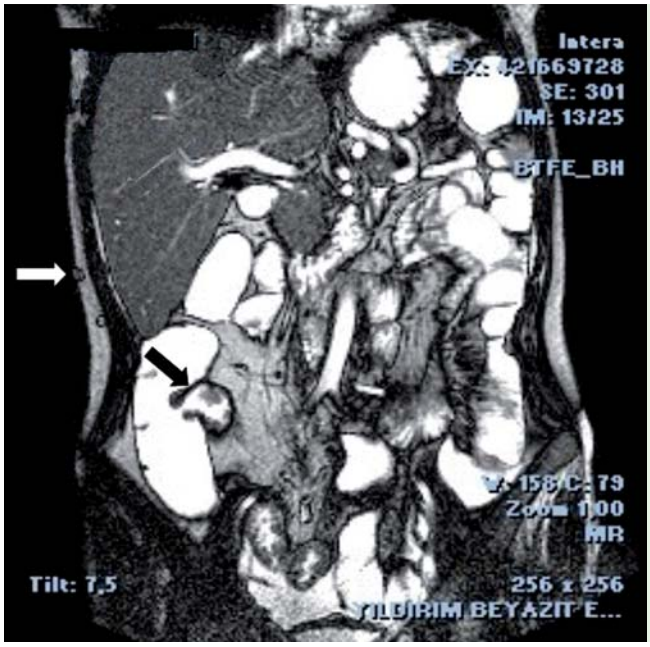

Fig. 2 Magnetic resonance enterography showing neurofibromas on the skin (white arrow), thickening of the jejunum and terminal ileum, and a pedunculated polyp, about 1 cm in diameter, located in the terminal ileum (black arrow).

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.

Endoscopy_UCTN_Code_CCL_1AC_2AC

Competing interests: None

Serta Kilincalp[1], Mevlüt Hamamcı[1], Hakan Akinci[1], Şahin Çoban[1], Aydın Şeref Köksal[2], İlhami Yüksel[2,3]

[1] Department of Gastroenterology, Diskapi Yıldırım Beyazıt Education and Research Hospital, Ankara, Turkey
[2] Department of Gastroenterology, Yüksek Ihtisas Training and Research Hospital, Ankara, Turkey
[3] Department of Gastroenterology, Yıldırım Beyazıt University School of Medicine, Ankara, Turkey

References

1 *Shekitka KM*, *Sobin LH*. Ganglioneuromas of the gastrointestinal tract: relation to Von Recklinghausen disease and other multiple tumor syndromes. Am J Surg Pathol 1994; 18: 250 – 257

2 *Karaahmet F*, *Coban S*, *Basar O* et al. Successful treatment of a large choledochal inflammatory polyp in a patient with cutaneous neurofibromatosis. Endoscopy 2014; 46 (Suppl. 01): E113. doi: 10.1055/s-0034-1364882

Bibliography

DOI http://dx.doi.org/10.1055/s-0034-1377434
Endoscopy 2015; 47: E4–E5

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