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Coexistence of small bowel gastrointestinal stromal tumor, small bowel adenocarcinoma, and ganglioneuroma in a patient with neurofibromatosis type I and caused intussusception

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Acquisition of data: Yazhuo Zhao, Liangrong Shi. Drafting of the manuscript: Yazhuo Zhao. Critical revision of the manuscript for important intellectual content: Yazhuo Zhao, Liangrong Shi. All authors approved the final version of the manuscript, including the authorship list.
Dear Editor,
A 67-year-old woman was admitted to our department with a 2-month history of abdominal pain that was paroxysmal and relieved after defecation. Multiple neurofibromas and café au lait macules was found all over the patient’s body. The fecal latent blood test was positive. The enhanced computed tomography revealed a terminal ileal tumor which was significantly enhanced with secondary intussusception and multiple lymph nodes were seen around the lesion (Figure 1A, 1B, 1C). The patient's clinical signs and imaging data led to a preliminary diagnosis of Neurofibromatosis type I. So the tumor of the ileum was considered to be NF-I-associated intestinal neurofibroma. Thereafter, the patient underwent laparoscopic small bowel resection. Postoperative pathological examination showed the tumor was adenocarcinoma and ganglioneuroma (Figure 1D, 1E). Intraoperative exploration also revealed a gastrointestinal stromal tumor. The patient has recovered well since 1 year postoperatively.

Discussion
Neurofibromatosis is an autosomal dominant disorder. Patients with neurofibromatosis type I have a higher risk to develop benign or malignant tumors. Simultaneous occurrence of abdominal tumors of three types in NF-I and causing intussusception is rare. Since NF-I itself can involve the gastrointestinal tract, and the patient in this case had a family history of NF-I, the tumor was first considered a NF-I-associated intestinal neurofibroma. It has been found that NF-I-associated intestinal neurofibromas present mainly as thickened intestinal walls with isointense or slightly hypointense density lesions on unenhanced CT and reduced enhancement in the venous phase after enhancement. In the present patient, the ileocecal wall
showed obvious thickening and significant enhancement with multiple enlarged lymph nodes surrounding. This is different from what is described in the literature. Postoperative pathological examination showed the tumor was adenocarcinoma and ganglioneuroma. The final diagnosis of how to identify whether the gastrointestinal tumor is caused by the primary tumor or NF-I still requires surgery to obtain pathological tissue, immunohistochemical testing, and then further genetic testing to confirm the diagnosis\[3\]. However, CT plays an important role in preoperative diagnosis. The analytical study of this case helps to expand the diagnostic ideas of imaging physicians. Either neurofibromatosis involving the intestine or neurofibromatosis with small bowel tumors should be managed aggressively to improve the patient’s survival and quality of life. At the same time, NF-I, as a genetic tumor syndrome, has no curative means yet, surgical treatment can be considered for those with combined abdominal tumors.

References
Figure 1. (A, B, C) Enhanced computed tomography revealed a terminal ileal tumor which was significantly enhanced with secondary intussusception and multiple lymph nodes were seen around the lesion. (D, E) Pathological examination showed the tumor was adenocarcinoma. Immunohistochemistry (IHC): Ki-67(approximately 85%+), p53(approximately 10%+), EGFR e(approximately 100%+), HER-2 (approximately 35%+), MSH2(+), MSH6(+), EBER(-), PMS2(-), MLH1(-), BRAF V600E(-).