A 55-year-old man presented with presyncope. He reported that his stools were dark red and that he had experienced mid-abdominal pain in the preceding week. His abdomen was tender to palpation in the periumbilical region but was otherwise soft and without masses. There was maroon-tinged stool in the rectum. His skin had circular areas of hyperpigmentation and multiple fleshy papules; the patient reported that these had been present since childhood but he had never sought medical care.

Colonoscopy, esophagogastroduodenoscopy, and push enteroscopy did not reveal a source of bleeding. However, an active area of bleeding in either the duodenum or jejunum was seen on capsule endoscopy, and a 2-cm ulcerated submucosal mass 50 cm distal to the pylorus was subsequently found on single-balloon enteroscopy (Figure 1, above). A CT scan of the abdomen accordingly revealed a single exophytic jejunal mass.
More than a dozen café au lait spots, neurofibromas, and axillary freckling were identified on a detailed dermatologic evaluation (Figure 2, left). On further questioning, the patient stated that his son had had similar skin features and that he died at a young age of an unknown soft tissue tumor. The findings in the patient were consistent with neurofibromatosis type I (NF-1). Pathologic examination of the jejunal mass following surgical resection confirmed gastrointestinal stromal tumor (GIST). Patients with NF-1 have at least a 150-fold increased risk of GIST compared with the general population.¹ Most NF-1–associated GISTs are clinically indolent, with favorable histologic parameters.² Fortunately, this patient’s tumor was low-grade and was completely resected (Figure 3, below); he has had no recurrence.

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