A rare case of adult intestinal intussusception: Epithelioid type mesenchymal tumor of the small intestine

Nadir görülen bir erişkin invajinasyon olgusu: İnce barsakta epiteloid tip mezenkimal tümör

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Small bowel intussusception is a rare cause of adult acute abdomen, in which an intraluminal neoplasm is the most frequent cause. Small bowel tumors are uncommon and can have a long delay prior to diagnosis. We present a case of intestinal intussusception originating from a rare variant of small bowel mesenchymal tumor, presenting with a hematologic disorder.

Key words: Schwannoma, intussusception, small bowel, epithelioid type mesenchymal tumor

INTRODUCTION

Although common in the pediatric population, intussusception accounts for only 1% of cases of small bowel obstruction in adults. In 90% of adult cases, intussusception is caused by an intraluminal neoplasm (1). Early diagnosis is essential to avoid treatment delay, which can increase morbidity and mortality. We report a case of small bowel intussusception, caused by a mesenchymal tumor, which had been under-diagnosed because of the preceding hematologic disorder.

CASE REPORT

A 29-year-old man admitted to the emergency room with abdominal pain, nausea and vomiting. The patient had experienced colicky abdominal pain for three weeks, which gradually increased during the last two days. He had no stool or gas discharge for one day. The patient had fatigue, dizziness and 20 kg weight loss in the last three months. He had been followed up by a hematologist for anemia and thrombocytosis for five months, with the suspicion of essential thrombocytosis and paroxysmal nocturnal hemoglobinuria. However, the patient had not yet been diagnosed despite detailed examination, including both gastrointestinal endoscopy (gastroscopy and colonoscopy) and bone marrow examination. On physical examination, he had abdominal distention, diffuse pain and localized rebound tenderness on lower quadrants. Abdominal ultrasound revealed target lesion in a small bowel segment, indicating intussusception, intraabdominal fluid, hepatomegaly and splenomegaly. On abdominal computerized tomography (CT), distended bowel segments were seen and an 8 cm small bowel segment was invaginated with a hyperdense lesion inside it, which was probably the leading point. CT also revealed hepatomegaly and splenomegaly. His blood count on admission was as follows: Htc: 25.9% (35-60), Hb: 7.1 g/dl (11-18 g/dl), WBC: 10,700/uL (5-10.5x10³/uL), and PLT: 1,076,000/uL (150-450x10³/uL). The blood biochemistry was not significant. The patient underwent emergency la-
parotomy. On exploration, an ileo-ileal intussusception was found at 220 cm from the ligament of Treitz (Figure 1), the proximal bowel segments were dilated and a 0.5 cm perforated area was seen 20 cm distal to the invaginated segment. We performed a segmental small bowel resection and an end to end anastomosis. The patient recovered uneventfully and was discharged on the sixth postoperative day.

On gross examination of the resected bowel, polypoid mass lesions (6x4x3 cm and 4x3x1.5 cm) were identified (Figure 2). On histopathology, both tumors showed identical characteristics. The superficial mucosal layer was ulcerated, and the tumor had invaded the muscular layer, with a focal subserosal invasion. On immunohistochemical examination, the tumors were diffusely positive for S-100 protein and vimentin-positive, but negative for CD34, c-KIT, smooth muscle actin (SMA), Melan-A, chromogranin, cytokeratin and placental alkaline phosphatase (PLAP). Cellular proliferative activity was assessed immunohistochemically using monoclonal antibodies to Ki-67 antigen and the tumor proliferation index was less than 1%. The mitotic count was less than 5 per 50 high power fields. With these findings, the masses were diagnosed as an epithelioid variant of mesenchymal tumor, primarily a peripheral nerve sheath tumor (Figure 3-A-B-C). The resection margins were intact.

On follow-up, the patient showed a rapid improvement. After one week, his blood count had returned to normal limits; by the end of the first month, on abdominal ultrasonography, splenomegaly had disappeared and the patient had gained 17 kilograms.

DISCUSSION

Gastrointestinal mesenchymal neoplasms are a heterogeneous group of tumors referred to as GISTs (gastrointestinal stromal tumors), which can arise anywhere within the gastrointestinal tract and have distinctive histologic and clinical features that vary according to their primary site of origin. The small bowel is the second most common site of GISTs, with approximately one-third arising in this location. Unlike GISTs of the stomach, those that occur in the small bowel are composed of spindle cells, and epithelioid variants are rare (2). As a group, a higher percentage of small bowel GISTs are malignant compared with those in the stomach and they should be evaluated with the use of site-dependent morphologic and prognostic factors.
Peripheral nerve sheath tumors are uncommon in the gastrointestinal tract. They represent 2-6% of stromal tumors of the gastrointestinal tract (3). In the tubal gut, they arise most commonly in the stomach and rarely in the esophagus and intestines (4). Schwannomas are mainly benign tumors derived from the cells of Schwann that form the neural sheath, and may become malignant if left untreated (5). They typically involve the submucosa and the muscularis propria. The overlying mucosa may be intact or ulcerated. Some have an intraluminal polypoid component as well (4). There are only a few reported cases of intestinal schwannomas causing intussusceptions.

In the case presented here, the patient actually had anemia caused by occult bleeding from the ulceration of the tumor, and thrombocytosis was probably secondary as an acute phase reactant. After a negative stool examination for occult blood, and negative gastroscopy and colonoscopy, the clinician was misled to a hematologic disorder. However, the small bowel was missed, and the patient suffered symptoms caused by anemia, until bowel obstruction developed. In 30-40% of KIT mutation-negative GISTs, PDGFR mutations were reported (6). The thrombocytosis could have also been the result of a PDGFR mutation, but unfortunately molecular studies are needed to confirm this hypothesis.

Small bowel tumors are uncommon and can have a long delay prior to diagnosis. Malignant small bowel tumors are more likely to produce symptoms and signs than benign tumors, particularly caused by small bowel obstruction. Abdominal CT is the best radiological investigation for small bowel tumors (7).

Prediction of biological behavior of schwannomas is also problematic and depends on the site, size and mitotic activity of the tumor. Mitotic count, size and cellularity are the best predictors of clinical outcome in small intestinal stromal tumors (8). Radical excision with negative margins is the treatment of choice, since their response to both chemotherapy and radiotherapy remains uncertain (9, 10). Regarding the size of the mass and the possible malignancy potential, our patient entered into a follow-up program, and was doing well at the 19th month.

In conclusion, epithelioid variant mesenchymal tumors of the small intestine are rare tumors, which may have a malignant potential; radical surgical resection is essential and will determine the overall outcome.

REFERENCES


