Gastrointestinal stromal tumor: a very rare cause of jejunoileal intussusception in a 6-year-old girl

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Abstract A 6-year-old girl was admitted to the emergency department with abdominal pain and bilious vomiting of 3 days in duration. Abdominal ultrasound examination showed an 8-cm-long intussuscepted intestinal segment with a target sign. There was a 26 × 28 × 23 mm nonperistaltic anechoic cystic mass suggestive of a duplication cyst. At laparotomy, the ileocecal region was normal with many enlarged lymph nodes from which biopsies were taken. There was a 20-cm-long intussuscepted segment at the proximal ileum close to the jejunum. After manual reduction, a 2-cm-long edematous segment resembling a duplication cyst served as the lead point. The segment was excised, and a primary bowel anastomosis was performed. She was discharged on the fifth postoperative day. The histopathologic examination revealed that the excised segment contained a gastrointestinal stromal tumor measuring 2.5 cm, with a mitotic rate of 2 to 3 mitoses per 50 high-power fields (low-risk group) showing an infiltrative growth pattern. On immunohistochemistry assay, some of the tumor cells were CD117 and CD34 positive, whereas all of them were smooth muscle actin and S-100 positive but CD10 negative. Staining index with Ki-67 was 5%. Surgical margins were free of tumor. The lymph nodes showed reactive hyperplasia. She was referred to the pediatric oncology department for further evaluation. Gastrointestinal stromal tumors are common in adults and may lead to intussusception. To the best of our knowledge, this is the first childhood case of gastrointestinal stromal tumor causing jejunoileal intussusception in the literature.

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Gastrointestinal stromal tumors (GISTs) account for only 0.1% to 3% of all gastrointestinal (GI) malignancies [1]. They are the most common mesenchymal tumors arising from the GI tract. They are relatively rare neoplasms, with an estimated prevalence of 15 to 20 per 1,000,000 [2]. Mesenchymal tumors may appear anywhere throughout the entire GI tract as well as the omentum and mesentery. They may present as huge intraabdominal masses at the time of diagnosis as seen in the elderly or as incidentally detected small nodules. Gastrointestinal stromal tumors occur predominantly in adults, and the literature on pediatric GISTs is inadequate to generalize the risk of the behavior, tumor recurrence, and patient survival. The tumors most commonly originate in the stomach (50%-70%), followed by the small intestine (25%-35%), colon and rectum (5%-10%), and...
esophagus (<5%). A range of 15% to 50% of cases are metastatic at diagnosis [3,4]. The fact that the disease can occur in many areas of the GI tract can, in part, explain the broad spectrum of its clinical presentations that span from GI bleeding, hemoperitoneum, anemia, and abdominal mass to the complete absence of symptoms [5]. Many cases of GIST, in fact, are discovered incidentally during imaging, endoscopy, or laparotomy for unrelated problems. A population-based study reported that 69% of GISTs were symptomatic, 21% were discovered incidentally during surgical procedures, and 10% were discovered at autopsy.

1. Case report

A 6-year-old girl presented to the emergency department with abdominal pain and bilious vomiting of 3 days in duration. Ultrasonographic examination performed at another facility demonstrated an intestinal invagination. Abdominal ultrasonography in our clinic revealed a 8-cm-long invaginated intestinal segment resembling a target sign. The intestinal segment had peristalsis and showed efficient circulation on Doppler study with minimal anechoic fluid around it. However, just inferior to this, there was a 26 × 28 × 23 mm nonperistaltic anechoic cystic mass suggestive of a duplication cyst.

She was transferred to the pediatric surgery clinic. On physical examination, she had neither palpable mass nor vomiting throughout her examination and during the investigative period. A repeat ultrasonography study confirmed the presence of an invaginated intestinal segment. At laparotomy, the ileocecal area was normal, but many enlarged lymph nodes were noted from which biopsies were taken. The ileum was followed proximally. There was a 20-cm-long invaginated segment at the proximal ileum close to the jejunum (Fig. 1). After manual reduction, there appeared a 2-cm-long edematous segment resembling a duplication cyst that was identified as the lead point for invagination. The tumor mass was excised with a 3-cm margin on each side, and a primary anastomosis was performed. The mass had encroached on the bowel lumen causing partial obstruction (Fig. 2). Nasogastric drainage tube was maintained preserved for 2 days postoperatively, and thereafter, enteral nourishment was started. She was discharged on the fifth postoperative day.

Macroscopically, cross-sectional surface of the specimen showed that the solid tumor mass had destroyed the smooth muscle layers in one area and ulcerated the mucosa at its largest width (Fig. 3). The pathology report stated that the excised ileal segment included a GIST measuring 2.5 cm, with a mitotic rate of 2 to 3 mitoses per 50 high-power fields (HPFs; low-risk group) showing an infiltrative growth pattern with necrosis and hemorrhage. Histologic examination

Fig. 1 At laparotomy, a jejunoileal intussusception with viable bowel was observed.

Fig. 2 The intraluminally protruding portion of the lesion resulted in partial bowel obstruction and macroscopically resembled a duplication cyst.

Fig. 3 Cross-sectional surface of the surgical specimen. The solid tumor mass had destroyed the smooth muscle layers (short blue arrows) in one particular area (long blue arrow) and ulcerated the bowel mucosa (red arrows) at its largest width (stars).
under low power showed a solid infiltrative tumor advancing from the muscularis propria to the mucosa. High-power fields showed spindle cells with atypical nuclei, organized in perpendicular bunches. On immunohistochemistry assay, some of the tumor cells were focally weakly positive for CD117, whereas all of them were strongly positive for smooth muscle actin and S100 but CD10 negative (Fig. 4A-D). Staining index with Ki-67 was 5%. Surgical margins were free of tumor. Lymph nodes showed reactive hyperplasia. She was referred to the pediatric oncology outpatient clinic where no other treatment was initiated. The finding of an abdominal ultrasound study 14 months postoperatively was normal. She will be followed up for at least 2 years.

2. Discussion

Gastrointestinal stromal tumors can occur throughout the GI tract from the esophagus to the rectum. They arise from Cajal cells in the intestinal wall or precursors of these cells, which are thought to undergo neoplastic transformation [6-8]. Gastrointestinal stromal tumors occur predominantly in adults, most being detected in the sixth or seventh decades, whereas only 10% are seen in patients younger than 40 years, and it is much rarer in children [5,6]. The literature on pediatric GISTs is inadequate to generalize the risk of the behavior, tumor recurrence, and patient survival. Gastrointestinal stromal tumors may be asymptomatic in 10% to 30% of patients. The most common presenting findings of GISTs are abdominal pain, a palpable mass, or GI bleeding, which is usually associated with the ulceration of the tumor into the lumen of the bowel [6,9]. They may rarely present with partial intestinal or biliary obstruction, hemoperitoneum, dysphagia, intussusception, or hypoglycemia. The symptoms of pediatric GISTs include hematemesis and anemia accompanied by fatigue and passage of tarry stools. A pediatric GIST is more common in girls than in boys. Gastrointestinal stromal tumors are identified by histologic, immunohistochemical, and molecular genetic assays [10,11]. However, clinical diagnosis, particularly of small or

Fig. 4 A-D, On histopathologic examination, HPFs revealed spindle cells with atypical nuclei, organized in perpendicular bunches (hematoxylin-eosin, ×200) (A). On immunohistochemistry assay, the tumor cells stained diffusely and strongly positive for smooth muscle actin (B) and S100 (C) but stained only focally and weakly positive for CD117 (D).
intramural GISTs, may be difficult. The most useful techniques for imaging and monitoring disease progression are endoscopic examinations and fused positron emission tomography/computed tomographic imaging.

Surgery is the first-line treatment and may lead to full remission in patients with a completely resectable nonmetastatic (localized) tumor. Endoscopic resection could be preferred in the treatment of small-sized GISTs. There are not much data on whether to perform resections in patients with positive margins or to resect sites of metastases. Although tyrosine kinase inhibitors are the mainstay of treatment for patients with metastatic GISTs, surgical resection of residual disease can sometimes be considered.

Benign GISTs are more common, but many tumors have an uncertain malignant potential. Tumor size, mitotic index, location of the primary tumor, CD117 and CD34 negativity in immunohistochemical studies, mucosal ulceration, and the presence of necrosis and KIT mutation status are the most reliable criteria for predicting the risk of the behavior, recurrence of the tumor, and patient survival [8]. Approximately 30% of newly diagnosed GISTs are malignant or have a high potential for malignancy. Gastrointestinal stromal tumors arising from the small intestine are generally associated with a less favorable outcome than primary gastric tumors.

Most GISTs are larger than 5 cm in diameter at the time of diagnosis, with a diameter of 10 cm being associated with a higher risk of local spread or distant metastasis. Petite (5 cm) small bowel GISTs with low mitotic indices (5 mitoses per 50 HPFs) rarely recur (<5% risk), whereas small bowel GISTs measuring more than 5 cm are at least at moderate risk (25%) for metastasis, independent of the mitotic index. The small intestinal GISTs with a mitotic rate greater than 5 mitoses per 50 HPFs have a 50% risk of recurrence. Although there are not much statistical data on the outcome of metastatic GISTs, some authors suggest that primary tumor size greater than 5 cm, mitotic index greater than 5 mitoses per 50 HPFs, spindle cell morphology, shorter disease-free interval, and unresected metastatic disease are associated with a worse prognosis.

Gastrointestinal stromal tumors are common in adults and may lead to intussusception [12,13]. Small tumor size (2 cm), low mitotic index, smooth muscle actin and S-100 positivity, poor C117 staining and CD10 negativity on immunohistochemical evaluation, low Ki-67 staining index (5%), and complete tumor resection are all indicators of good prognosis in the present case. To the best of our knowledge, this is the first childhood case of GIST causing jejunoileal intussusception in the literature.

References