Reception: **October 8, 2024** | Acceptance: **October 8, 2025** | Publication: **February 1, 2026**

Secondary intestinal intussusception to a GIST tumor in a girl

—

Rubén Martín Alvarez-Solis[1] • rubenalsolecito@hotmail.com
ORCID: 0000-0001-9703-4626

Armando Quero-Hernández[2] • ORCID: 0009-0003-5588-9265

Marcela del Pilar Vargas-Vallejo[3] • ORCID: 0009-0002-4150-8508

David Bulnes-Mendizába[4] • ORCID: 0009-0001-2437-3384

Nadia Selene Gómez-Villegas[5]

1 División de Cirugía Pediátrica. Hospital del Niño "Dr. Rodolfo Nieto Padrón. Villahermosa, Tabasco. México

2 Servicio de Oncología Pediátrica. Hospital General "Dr. Aurelio Valdivieso". Servicios de Salud de Oaxaca. Oaxaca de Juárez, México.

3 Servicio de Oncología Pediátrica del Hospital del Niño "Dr. Rodolfo Nieto Padrón" de Villahermosa, Tabasco. México

4 Servicio de Patología. Hospital del Niño "Dr. Rodolfo Nieto Padrón". Villahermosa, Tabasco. México

5 Cirugía pediátrica Hospital del Niño. "Dr. Rodolfo Nieto Padrón. Villahermosa, Tabasco, México

To quote this article:
Alvarez Solis, R. M., Quero Hernández, A., Vargas Vallejo, M. del P., Bulnes Mendizábal, D., & Gómez Villegas, N. S. Invaginación intestinal secundaria a un tumor de GIST en una niña. *Espacio I+D, Innovación más Desarrollo, 15*(43). https://doi.org/10.31644/IMASD.43.2026.a08

— Abstract—

GIST tumors are the most common mesenchymal tumors of the gastrointestinal tract in adults, but are very rare in children. We report the case of a 9-year-old female patient with abdominal pain and the ultrasound suggested intestinal intussusception. During the exploratory laparotomy, an intraluminal tumor was found, which subsequently confirmed the diagnosis of Gist tumor. She did not receive chemotherapy treatment. She has been under observation for more than 5 years without evidence of metastasis or recurrence. We carry out a brief discussion and analysis of the literature on GIST tumor in children.

Keywords:
GIST tumor in children; causes of secondary intussusception; Digestive tract bleeding.

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors of the digestive tract; they originate in the interstitial cells of Cajal, which are responsible for peristaltic activity in the digestive tract, and are characterized by the expression of the tyrosine kinase receptor CD117 (KIT). Most GISTs harbor gain-of-function mutations in the v-KIT (KIT) or platelet-derived growth factor receptor alpha (PDGRFA) genes, resulting in the activation of kinases that promote neoplastic proliferation in 80% of cases.[1]

Currently, the term GIST refers to CD117-positive, spindle-shaped or epithelioid mesenchymal tumors that are primary to the gastrointestinal tract, mesentery, and retroperitoneum. This term was first used in 1983. Mazur and Clark used it to describe non-epithelial tumors of the gastrointestinal tract that lacked the ultrastructural features of smooth muscle and the immunohistochemical characteristics of Schwann cells.[2]

It is the most common sarcoma of the gastrointestinal tract (GIT). It accounts for 2% of tumors in this region but 80% of gastrointestinal sarcomas. Its incidence is estimated at 10 to 20 cases per million people per year. Its prevalence is higher due to the long clinical course of the disease (10–15 years).[2,3]

The peak incidence occurs between the 4th and 6th decades of life, with a similar distribution across genders. However, recent studies suggest a slightly higher incidence in males. It is rare in the pediatric population, with an annual incidence of 0.02 to 0.44 cases per million in individuals under 20 years of age. [3,4]

GISTs share immunophenotypic similarities with Cajal interstitial cells, which are located around the myenteric plexus and scattered throughout the muscularis propria. Characteristics such as the expression of KIT, CD34, the heavy chain of smooth muscle myosin, and nestin are common to both. Cajal interstitial cells act as pacemaker cells mediating between the nervous system and the smooth muscle system. They express CD117 in 95% of cases, just like GISTs, and exhibit a mixture of neural and myogenic features in their ultrastructure.[5]

In 1998, Kinblom suggested that GISTs originate from a stem cell that differentiates into a Cajal interstitial cell. The precursor cell hypothesis would explain why mesenchymal tumors with histology similar to GISTs, which are CD117-positive, can occur in the omentum and mesentery.[5]

Our aim is to present a rare case of GIST in a pediatric patient that presented with symptoms of intussusception, along with the results of treatment and a review of the literature.

CLINICAL CASE

A 9-year-old female patient presenting with abdominal pain that began 20 days prior. She had previously received multiple treatments with antiparasitic and antidiarrheal medications without clinical improvement, leading her to seek care at our facility for persistent abdominal pain. Physical examination revealed generalized pallor; a holosystolic murmur in the precordial area secondary to “anemic heart”; tachycardia;

mild respiratory distress; a soft, depressible abdomen that was not tender on palpation; decreased peristalsis; and no signs of peritoneal irritation.

A complete blood count (CBC) was performed, revealing severe hypochromic microcytic anemia (4 gr/dl), a white blood cell count of 6,500/mm^3, and normal differential and platelet counts. An abdominal ultrasound was ordered and revealed intermittent colonic intussusception. She was evaluated by pediatric surgery, and an exploratory laparotomy was performed; a jejuno-ileal intussusception was observed secondary to a palpable mass in the intestinal lumen measuring approximately 2 cm in diameter. Resection of the segment and an entero-entero anastomosis were performed. Examination of the remainder of the small intestine and colon was normal.

The histopathological report indicated a gastrointestinal stromal tumor (GIST) with polypoid growth into the intestinal lumen (Figure 1), describing epithelioid and spindle cells with more than 5 mitoses per field. Immunohistochemistry confirmed positivity for: CD117, DOG1, and CD34.

The patient remains asymptomatic and has been tumor-free for 5 years following surgical resection.

DISCUSSION

The etiology of intussusception is usually inflammation of Peyer's patches in infants. In a patient over 2 years of age presenting with intestinal intussusception, one must always look for an organic or secondary cause. In this pediatric case, which appears to be the first reported in Mexican literature, the jejuno-ileal intussusception was caused by a gastrointestinal stromal tumor.

Kinblom and colleagues conducted a population-based study, collecting epidemiological and prognostic data on GISTs. In a Swedish population of 1.3–1.6 million people, with four referral hospitals, they investigated c-KIT-positive mesenchymal tumors between 1983 and 2000. They studied 650 cases, of which 398 met the diagnostic criteria for GIST. Seventy-two percent of the GIST cases had been diagnosed with another condition. 28% were diagnosed with GIST, 34% with leiomyomas, 18% with leiomyosarcomas, 13% with leiomyoblastomas, and 7% with other diagnoses. Regarding clinical presentation, 69% were symptomatic, with most symptoms being nonspecific: vague pain, anorexia, anemia, weight loss, and nausea. Less common were gastrointestinal bleeding (due to mucosal ulceration) or intraperitoneal bleeding (due to tumor rupture).[6]

In our patient, the tumor was found at the jejuno-ileal junction, which is very rare, as Kindblom reports that they can be located in the upper gastrointestinal tract (GIT), the mesentery, or the retroperitoneum. Between 50% and 60% of cases originate in the stomach, 20% to 30% in the small intestine, approximately 10% in the colon, and the remaining 15% in other sites: the rectum, esophagus, mesentery, and retroperitoneum. The esophagus accounts for 5% of cases.[3,4]

The macroscopic appearance depends on the size, which ranges from 1 to 32 cm, although most are larger than 2 cm. In our patient, the intraluminal tumor measured 1.5 cm. They appear as serous, intramural, or submucosal nodules. Large tumors typically exhibit exophytic growth, toward the lumen, with or without mucosal ulceration (20–30% of cases). They may also protrude toward the serosa with a large extraparietal component, which masks their gastrointestinal origin on imaging studies. Histologically, three architectural patterns are distinguished in GIST tumors: spindle cell (70%), epithelioid (20%), and mixed (10%). In our patient, a mixed type was found: spindle or spindle-shaped cells and epithelioid cells.[7]

The presentation of GIST varies in terms of symptoms and signs, depending on the tumor's size and location. In our patient, chronic abdominal pain was the predominant symptom. She has no history of hematemesis or melena; however, occult blood in the stool is possible due to chronic, unnoticed gastrointestinal bleeding.[7]

Ultrasound is often the initial diagnostic technique used in many patients with GIST tumors. These tumors are typically large and hypoechoic; in our patient, ultrasound did not detect the tumor prior to surgery, leading to a diagnosis suggestive of intussusception. Other recommended imaging studies include contrast-enhanced abdominal CT, considered the most sensitive technique, in which GIST tumors are described as hypervascular masses with a heterogeneous appearance due to hemorrhagic necrosis or cystic degeneration, and magnetic resonance imaging (MRI) if a rectal tumor is suspected.[8]

GIST tumors have immunohistochemical characteristics that are useful for confirming the suspected diagnosis. Approximately 95% are positive for KIT (CD117), 98% for DOG-1, 80% for PDGFRA, 70% to 80% for CD34, and 30% to 40% for smooth muscle actin and others [2,9,10] Thirty percent of GISTs are malignant, and currently there are at least four risk stratification systems (Fletcher, Miettinen, the Armed Forces Institute of Pathology [AFIP], and the risk criteria of the National Comprehensive Cancer Network [NCCN]), which generally consider the following variables: tumor size, mitotic rate per high-power field, and tumor site. In the case we present, the tumor size was less than 5 cm, the mitotic rate was greater than 5 per field, and the location was in the jejunoileal region; thus, based on AFIP criteria, it would be classified as moderate risk.[11] In children, the most common location is the stomach, and the disease typically presents with multiple tumor nodules (multifocal disease). In Mexico, Morales et al. reported two cases located at the esophagogastric junction and within the gastric cavity, presenting with chronic anemia; treatment consisted of surgical resection in one case and neoadjuvant therapy with imatinib in the second case.[12]

The prognosis shows some association with anatomical location, with a tendency for those located in the small intestine to be more malignant than those located in the stomach.[11] In a study of 1,700 gastric GISTs, Miettinen[10] observed that 83% were benign. In the duodenum, however, half are typically malignant. In the esophagus,

they are rare. Leiomyomas are the most common. However, more than half of GISTs are malignant. Other locations have been described, such as in the lymph nodes.[13,14]

The primary treatment for GISTs has been surgical resection, especially for localized tumors; whereas malignant GISTs are treated with various regimens of chemotherapy and/or radiation therapy, with a reported 5-year overall survival rate of approximately 12%, particularly in unresectable or metastatic tumors that relapsed or progressed and were resistant to cytotoxic chemotherapy and/or radiation therapy.[15] In recent decades, with a better understanding of the molecular mechanisms that give rise to these neoplasms, therapies have emerged that target these mutations at specific sites, thereby altering tumor proliferation and angiogenesis. Tyrosine kinase inhibitors block KIT and PDGFRA receptors (present in 90% and 80% of GISTs, respectively) and represent the current standard of care for reducing relapses and for the high-risk group; Imatinib is one of the first drugs with this mechanism of action, reducing the risk of disease progression or death by 54%.[15,16]

Given the rarity of these tumors in the pediatric stage, there are no specific treatment recommendations; the reported approaches have been extrapolated from the management of adult patients. Arimatias et al., in their systematic review that included 184 patients—68% of whom were women, with a mean age of 14 years—found that the tumor was located in the stomach in 90% of cases and in the intestine in 7%. Initial management consisted of partial to total gastrectomy; 46% of cases had metastatic disease at diagnosis and received standard chemotherapy in 2%, first-generation tyrosine kinase inhibitors in 34%, and second-generation tyrosine kinase inhibitors in 13%, achieving complete remission in 52% and an overall survival of 24% at 6 years.[17]

In our case, based on the histological findings, the tumor was classified as moderate-risk; the treatment plan involved complete resection without any adjuvant therapy, and at 5 years of follow-up, the patient remains free of tumor activity.

CONCLUSIONS

It should be noted that intussusception is idiopathic in 90% of cases and occurs in infants. When intussusception occurs in children older than 2 years, there is always an organic or secondary cause, as in the case described here.

This is the first reported case of a GIST in a 9-year-old child, treated with surgery alone, who remains free of tumor activity 5 years after diagnosis.

REFERENCES

1. **Naito**, Y., Nishida, T., & Doi, T. (2023). Current status of and future prospects for the treatment of unresectable or metastatic gastrointestinal stromal tumours. *Gastric Cancer, 26*(3), 339-351. https://doi.org/10.1007/s10120-023-01381-6
2. **Tyagi**, I. V., & Anand, S. (2025). Gastrointestinal Stromal Tumors Market Share, Report 2035. En *Market Research Future*. https://www.marketresearchfuture.com/reports/gastrointestinal-stromal-tumors-market-1583/?utm_term=&utm_campaign=&utm_source=adwords&utm_medium=ppc&hsa_acc=2893753364&hsa_cam=23142125492&hsa_grp=190076755354&hsa_ad=779362054048&hsa_src=g&hsa_tgt=dsa-2443880216606&hsa_kw=&hsa_mt=&hsa_net=adwords&hsa_ver=3&gad_source=1
3. **Popoiu**, T., Pîrvu, C., Popoiu, C., Iacob, E. R., Talpai, T., Voinea, A., Albu, R., Tăban, S., Bălănoiu, L., & Pantea, S. (2024). Gastrointestinal Stromal Tumors (GISTs) in Pediatric Patients: A Case Report and Literature Review. *Children, 11*(9), 1040. https://doi.org/10.3390/children11091040
4. **Hallie** J. Quiróz, Brent A. Willobee, Matthew S. Sussman, Bradley R. Fox, Chad M. Thorson, Juan E. Sola et al. Pediatric gastrointestinal stromal tumors- a review of diagnostics modalities. *Transl Gastroenterol Hepatol*. 2018;3:3-54 doi:10.21037/tgh.2018.07.08
5. **M**. Shimomura, S. Ikeda, Y. Takakura, Y. Kawaguchi, M. Tokunaga, Haruka Takeda, et al. Gastrointestinal stromal tumors of the small intestine in pediatric populations: a case report and literature review. *Pediatr Surg Int, 26* (2010), pp. 649-654
6. **Kinblom** L-G, Remotti HE, Aldenborg F, et al. Gastrointestinal Pacemeaker cell tumor (GIPACT). Gastrointestinal stromal tumors show phenotypic characteristics of interstitial cells of Cajal. *Am J Pathol* 1998; 152: 1259-69. 5.
7. **Saund** MS, Demetri GD, Ashley SW. Gastrointestinal stromal tumors (GISTs). *Curr Opin Gastroenterol* 2004; 20: 89-94.
8. **Corless** CL, Fletcher JA and Heinrich MC. *Biology of gastrointestinal stromal tumors. J Clin Oncol* 2004; 22: 3813-25
9. **Bucher** P, Villiger P, Egger J-F, et al. Management of gastrointestinal stromal tumors: from diagnosis to treatment. *Swiss Med Wkly* 2004; 134: 145-53
10. **Miettinem** M, Lasota J. Gastrointestinal stromal tumors (GISTs): definition, ocurrence, pathology, differential diagnosis, and molecular genetics. *Pol J Pathol* 2003; 54: 3-24.
11. **Inga**-Marie Schaefer, Adrián Mariño Enríquez, Jonathan A Fletcher. What is new in gastrointestinal stromal tumor?. *Adv Anat Pathol*. 2017;24(5): 259-267
12. **Morales** Peralta Adrián, Covarrubias Espinoza Gilberto, Rios García Candy Guadalupe, Larios Farak Tania Clarisa, Millán Valenzuela Luis Omar, Galván Ruiz Vanessa Guadalupe. Tumor de GIST pediátrico. Presentación de dos casos y revisión de la literatura. *Bol Clin Hosp Infan Edo Son*. 2017;34(2): 127-135

13. **B**. Samarji, T. Walter, F. Dijoud, S. Collardeau Frachon, F. Hameury, R. Dubois, et al. [Pediatric gastrointestinal stromal tumors: report of three cases] *Gastroenterol Clin Biol, 34* (2010), pp. 407-40
14. **Stiles**, Z.E.; Fleming, A.M.; Dickson, P.V.; Tsao, M.; Glazer, E.S.; Shibata, D.; Deneve, J.L. Lymph Node Metastases in Gastrointestinal Stromal Tumors: An Uncommon Event. *Ann. Surg. Oncol.* 2022, 29, 8641–8648.
15. **Ng** EH, Pollock RE, Munsell MF, Atkinson EN, Romsdahl MM. Prognostic factors influencing survival in gastrointestinal leiomisarcoma. Implications for surgical management and stag ing. *Ann Surg.* 1992; 215: 68-77
16. **Prof** Yoon-Koo Kang, Min-Hee Ryu, Changhoon Yoo, Prof Baek-Yeol Ryoo, Hyun Jin Kim, Jong Jin Lee, et al. Resumption of imatinib dosing to control metastatic gastrointestinal Stromal Tumors (GIST) after failure of Imatinib and Sunitinib: Results of a randomised, placebo-controlled, phase 3 trial (RIGHT). *Lancet Oncol.* 2013; 14(12): 1175-1182. doi:10.1016/S1470-2045(13)70453-4.
17. **Arimatias** Raitio, Adeline Salim, Dhanya Mullassery, Paul D Losty. Current treatment and outcomes of pediatric gastrointestinal stromal tumors (GIST): a systematic review of published studies. *Pediatr Surg Int. 2021; 37*(9):1161-1165

Annex

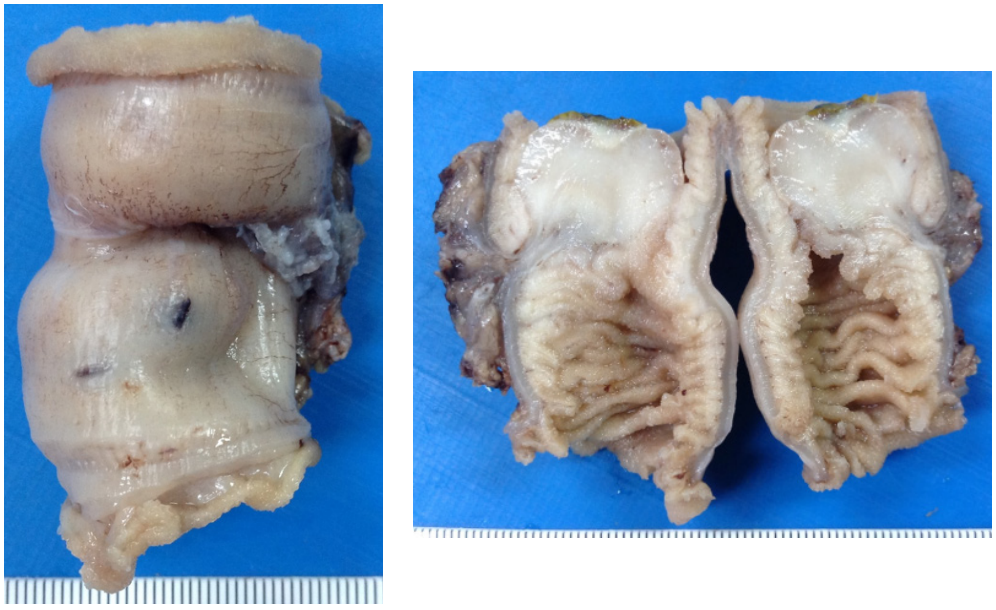

Figure 1. Macroscopic Description: A 6 × 3 cm Segment of the Jejunum with a polypoid mass showing traction on the intestinal wall

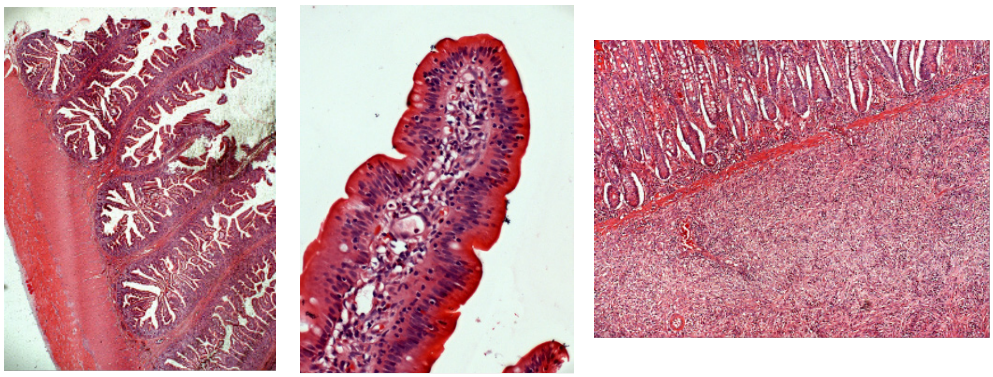

Figure 2. Microscopic Description. Proliferation of cells forming a polypoid mass originating from the lamina propria and muscle layer, without involvement of the superficial mucosa, is observed