Case Report

Intraluminal gastrointestinal stromal tumor leading to intussusception in an adult

Neha Parashar[1], Navpreet Kaur[2], Reena Tomar[3], Nita Khurana[4], C B Singh[5]

From [1]Senior Resident, [3]Professor, [4]Director Professor and Head, Department of Pathology, [5]Director Professor, Department of Surgery, Maulana Azad Medical College and Associated Lok Nayak Hospital, New Delhi, India, [2]Department of Pathology, Dr. B.R. Ambedkar State Institute of Medical Sciences, Mohali, Punjab, India

ABSTRACT

Gastrointestinal stromal tumor (GIST) is the most common mesenchymal tumor of the abdomen. Most of these tumors occur in the stomach. They arise from the interstitial cells of Cajal. GIST usually occurs in adults of more than 50 years of age, with a median age of 62–63 years. Fewer than 10% of cases occur at <40 years of age. It commonly presents as exophytic growth. GIST may lead to symptoms due to the effect of the mass. Mucosal ulceration may cause blood loss leading to anemia. Asymptomatic GIST may be discovered as an incidental finding during imaging or endoscopy. Intraluminal GIST presenting clinically as intussusception is extremely rare. This case report presents a case of intestinal GIST in a 35-year-old man. The tumor presented as an intraluminal nodule leading to intussusception. A complete resection of the tumor was done. Histopathological examination with immunohistochemistry helped to arrive at a definite diagnosis and determine the prognosis.

Key words: Gastrointestinal stromal tumor, Intussusception, Stomach tumor

Classical gastrointestinal stromal tumor (GIST) arises from the interstitial cells of Cajal (ICC) in the muscularis propria layer of the gastrointestinal (GI) tract [1]. It is one of the most common sarcomatous tumors of the GI tract [2]. GIST has a tendency to present as an exophytic growth arising from the serosal aspect of the GI tract [1]. Intraluminal growth is a rare presentation [2]. Anemia, weight loss, GI bleeding, and abdominal pain are the most common clinical findings in patients diagnosed with GIST [3]. GIST-causing intussusception is extremely rare [4]. The annual incidence of intussusception in adults is approximately 2–3 per 1,000,000 [2].

The present case report highlights a rare presentation of intussusception due to GIST in a 35-year-old male.

CASE REPORT

A 35-year-old male presented to the hospital with episodes of colicky pain in the central and lower abdomen for the past 3 months. The pain aggravated after heavy food intake. It was associated with decreased appetite and weight loss of about 5 kg in the past 3 months. He also had a history of passing thin, streaky stools.

On examination, the patient was conscious and alert. His conjunctiva was not pale, and his sclera was anicteric. The patient was afebrile with a blood pressure of 110/80 mmHg and a heart rate of 104 beats/min. On abdominal examination, a vague mass was palpable in the right lower quadrant. It had ill-defined margins and was not moving with respiration. No shifting or dullness was noted.

Laboratory investigations showed a normocytic normochromic blood picture hemoglobin 14.5 g/dL (normal value 13–17 g/dL), total leukocyte count of 6730/µL (4000–10,000/µL), differential leukocyte count of polymorphs 78%, lymphocytes 18%, monocytes 2%, eosinophils 2% (polymorphs 40–80%, lymphocytes 20–40%, monocytes 2–10%, eosinophils 1–6%), and platelet count of 2.32 lakhs/mm^3 (1.5–4 lakhs/mm^3). Ultrasound abdomen showed telescoping of bowel loops, giving a target appearance in the right lower abdomen suggestive of intussusception.

A laparoscopic-assisted open right hemicolectomy with ileotransverse anastomosis was performed. A segment of the intestine with cecum and appendix altogether measuring 21 cm was received as a specimen in the department of pathology. On cutting open, a polypoidal mass measuring 3.5 × 3 × 2 cm was present within the lumen of the intestine (Fig. 1). The cut section was gray-white with areas of hemorrhage. On microscopy, interlacing fascicles and whorls of spindle cells with elongated cigar-shaped nuclei were seen with inflammatory cells comprising neutrophils, lymphocytes, and eosinophils (Fig. 2).

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Correspondence to: Dr. Reena Tomar, Department of Pathology, Maulana Azad Medical College, New Delhi - 110 002, India. E-mail: drreenat@yahoo.com

On immunohistochemistry, cells were positive for CD 117, DOG 1 (Fig. 3), and vimentin but negative for Desmin, SMA, and ALK. A final diagnosis of GIST was made. Based on the morphology, size (2–5 cm), mitotic count (<5/50 HPF), and location (ileal), the polypoidal mass was classified as a low-grade GIST with a low risk of progression and metastasis [5,6].

The patient had an uneventful recovery and was symptom-free on follow-up. The patient was not prescribed imatinib mesylate as the tumor was small in size with a low mitotic index.

DISCUSSION

GIST has an incidence of approximately 16–22 cases/million people in Asia. There has been an increase in the number of cases reported as GIST after the introduction of the anti-CD117 antibody for immunohistochemical staining. The increase in incidence has also been associated with changes in diagnostic methods and the reclassification of many mesenchymal GI tumors previously diagnosed as smooth muscle tumors [2].

GIST arises from the ICC. It arises most commonly from the stomach (60–70%), followed by the small bowel (20–25%). Exophytic growth is seen in 79% of the GIST. Intraluminal or mixed growth is rare [7]. In the present case, the intraluminal nodule was present in the ileum. Intussusception and obstruction are very uncommon presentations of these lesions because of their tendency to grow in an extra-luminal manner. GIST presenting as an intraluminal nodule with intussusception is extremely rare. Only a few cases have been reported in the literature to date. GISTs can arise at any age. It is most common between 40 and 70 years of age [3]. Soreide *et al.* reviewed 29 studies consisting of 13,550 patients and found the median age of patients diagnosed with GIST to be 65 years. In the present case, the patient was 35 years of age.

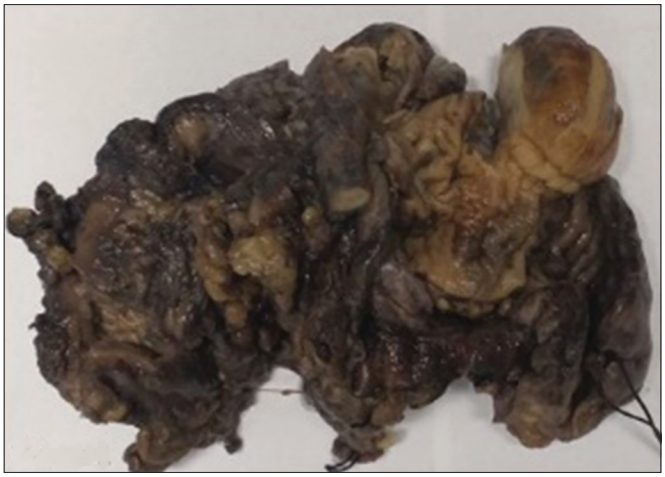

Figure 1: Polypoidal mass within the lumen of intestine

The classic triad of intussusception (abdominal tenderness, palpable abdominal mass, and hemoglobin-positive stools) is rare in adults. Intussusception causes only 1% of all bowel obstructions in adults. It comprises 0.02–0.03% of all cases reported to the hospital and 1–3% of all cases of surgical intestinal obstructions. Polypoid lesions such as inflammatory fibroid polyp, adenoma, leiomyoma, and Peutz–Jeghers syndrome; tuberculosis; and Meckel's diverticulum are the causes of intussusception involving the small intestine in 90% of the cases [5]. In a review article by Ssentongo *et al*., intussusception secondary to GIST was found to be reported in 18 cases worldwide between 1983 and 2018. Of these 18 cases, four had been reported in India of which only 1 case showed intussusception in the ileum [2]. The present case is probably the second case of ileal intussusception secondary to GIST to be reported in India to the best of our knowledge. In Portugal, Giestas *et al*. reported an unusual case of jejunojejunal intussusception caused by a GIST that presented with long-standing obscure GI bleeding as the first symptom [4]. In the present case, the patient presented with abdominal pain and thin streaky stools. On examination, an abdominal lump was palpable. Radiological findings were suggestive of intussusception. Diagnosis of small bowel GIST is difficult as these are inaccessible to endoscopy [8].

Surgery is the gold standard in the case of localized GIST [2]. GISTs are submucosal lesions that appear to arise from the muscularis propria of the bowel wall. However, in the present case, the lesion was intraluminal. An accurate diagnosis of such lesions can be made on the basis of medical history, physical examination, imaging modalities, and confirmation by a histopathological examination of the tumor [2].

The diagnosis of GIST can be confirmed with the help of histopathology and immunohistochemistry. The spindle cell type of GIST comprises cellular proliferation of bland spindle cells with pale to eosinophilic fibrillary cytoplasm. Cells are arranged in whorls or short intersecting fascicles. Benign forms usually show minimal pleomorphism with <5 mitotic figures/50 HPF [9]. Patho-histologically, GIST is defined by positive immunostaining for c-Kit protooncogene CD117 (overexpressed in 95%) and CD34 (positive in 60% to 70%) [2].

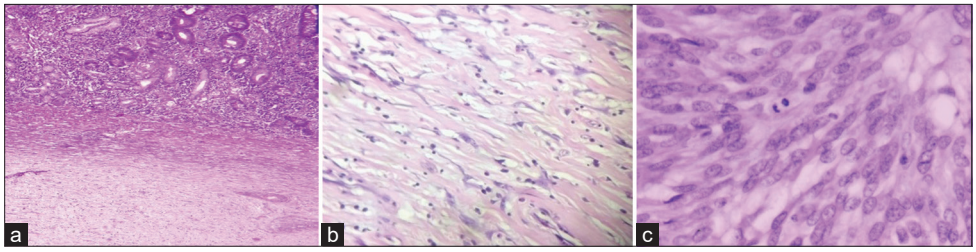

Figure 2: (a) Intestinal mucosal lining with fascicles of spindle cells underneath (HE × 100), (b) interlacing fascicles of spindle-shaped cells with mild inflammation (HE × 200), (c) mitosis 1-2/HPF (HE × 400)

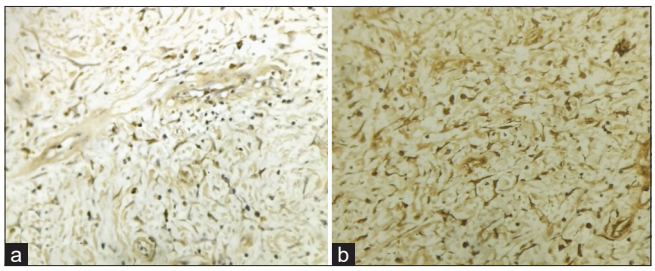

Figure 3: (a) CD117 positive (IHC × 200); (b) DOG 1 positive (IHC × 200)

DOG1, a recently described immunohistochemical marker, has high sensitivity and specificity for GIST. DOG1 is a calcium-dependent, receptor-activated chloride channel protein expressed in GIST. The expression of DOG-1 is independent of mutation type and can be used in the diagnosis of KIT-negative tumors [10]. In the present case, interlacing fascicles and whorls of spindle cells were seen. Cells were positive for CD 117, DOG 1, and vimentin. They were negative for CD 34, desmin, SMA, and ALK. The immunohistochemistry helps in differentiating it from inflammatory myofibroblastic tumors and solitary fibrous tumors.

CONCLUSION

Intussusception is a rare presentation of GIST. Although the presentation of GIST as an intraluminal nodule leading to intussusception is extremely uncommon, pathologists should always consider GIST as a differential diagnosis in an adult presenting with an intraluminal nodule.

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