Diffuse Intestinal Lipomatosis Presenting as Adult Intussusception

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Abstract

Background: A 25-year old female presented to the Emergency Department with intermittent abdominal pain. Computed tomography imaging demonstrated multiple fat-density tumors in the small intestine and colon associated with two long-segment intussusceptions.

Methods: Colonoscopy showed a partially obstructing multilobulated mass in the descending colon; biopsy was non-diagnostic. At exploratory laparotomy, she was found to have hundreds of variable-sized fatty submucosal tumors distributed diffusely throughout the small intestine and colon. Proximal ileal resection and near-total abdominal colectomy with ileorectal anastomosis was performed to remove the heavily diseased and intussuscepted bowel segments.

Results: Pathology confirmed the diagnosis of diffuse intestinal lipomatosis, identifying extensive submucosal lipomas in the small and large bowel. The largest lipomas, which had served as intussusception lead points, measured 4 cm in the colon and 6 cm in the ileum. Her post-operative course was uncomplicated and she remains symptom-free at over one year after surgery.

Conclusions: Symptomatic diffuse intestinal lipomatosis is a very rare condition with variable presenting symptoms. It is readily diagnosed by cross-sectional imaging and treated by surgical resection of the intestinal segments causing symptoms. The etiology may involve fetal somatic genetic mutations. Long-term risk of malignancy is believed to be low, but optimal follow-up remains undefined.

KEYWORDS: diffuse intestinal lipomatosis, lipoma, gastrointestinal tract, intussusception
Introduction
A 25-year old African-American female presented to the Emergency Department (ED) complaining of abdominal pain. The pain was sharp, intermittent, and localized to the left upper quadrant. It was associated with small-volume non-bilious emesis that occurred after the pain started. She reported having episodes of similar symptoms approximately once per month for the past two years. She had presented to ED on several previous occasions, but since her symptoms always resolved spontaneously they were attributed to gastroesophageal reflux. She had no other systemic, gastrointestinal, urologic, or gynecologic symptoms. Her past medical history was significant only for two normal pregnancies with uncomplicated vaginal deliveries. Family history was negative for any gastrointestinal problems or benign or malignant tumors. Her vital signs and physical examination were normal. Laboratory studies, including complete blood count, electrolytes, liver function tests, urinalysis, and urine pregnancy test were normal. A computed tomography (CT) scan of the abdomen and pelvis with oral and intravenous contrast was obtained in the ED. The CT scan was grossly abnormal, demonstrating fat-density masses throughout the walls of colon and small bowel (Figure 1), with a long-segment colonic intussusception in the left upper quadrant (Figure 2) and a second intussusception of the distal ileum. Oral contrast was able to pass through the areas of intussusception. There was mesenteric congestion and adenopathy but no evidence of bowel ischemia or metastatic disease.

Methods
The patient was admitted to the Gastrointestinal Surgery service. At the time of admission, her symptoms had abated spontaneously and she was passing flatus. An exploratory laparotomy with bowel resection was recommended, but the patient initially refused. She did consent to colonoscopy, which was performed by the attending surgeon; this demonstrated a large, partially obstructing, multilobulated polyp in the descending colon. The colon proximal to the polyp could not be evaluated due to the presence of solid stool. Multiple biopsies of the polyp were taken, but revealed only colonic mucosa with non-specific reactive changes. After further discussion, the patient gave her informed consent and was taken to the operating room. Upon entering the abdomen through a midline incision, the small bowel and colon were found to be studded with hundreds of variable-sized fatty tumors from the proximal jejunum to the distal sigmoid. The epiploic appendages were grossly hypertrophied throughout the colon. There was a massive long-segment intussusception of the proximal ileum with grossly distended bowel, with multiple very large fatty tumors serving as a lead point. There was a second intussusception starting at the terminal ileum with intussusception into the right colon, and this segment was subsequently
intussuscepted into the distal transverse colon. There was no evidence of necrosis or perforation. The distal ileum between the two intussuscepted segments contained a few small fatty tumors, but appeared healthy. The stomach, duodenum, and distal sigmoid colon appeared normal and healthy.

Results
A near-total abdominal colectomy was performed with a stapled side-to-side anastomosis of the distal ileum to the sigmoid colon. The heavily diseased segment of the proximal ileum was then resected separately, and small bowel continuity was reestablished with a second stapled side-to-side anastomosis. Multiple fatty tumors were left in place throughout the remaining small bowel, but all appeared to be small with no other segments of intussusception. The patient’s post-operative course was uncomplicated. She tolerated advancement of her diet, resumed bowel function, and was discharged home on the fifth post-operative day. She has had no recurrence of her gastrointestinal symptoms at over a year after surgery. Pathologic examination of the resected specimens identified extensive submucosal lipomas in the small and large bowel, confirming the diagnosis of diffuse intestinal lipomatosis (Figure 3). The largest lipomas, which had served as lead points for the intussusceptions, measured 4 cm in the colon and 6 cm in the ileum (Figure 4).

Discussion
As of this writing, thirteen other cases of diffuse intestinal lipomatosis have been reported. Seven cases predominantly involved the small bowel and its mesentery (1-4), four predominantly involved the colon (5-7), and two were associated with extraintestinal lipomatosis (8, 9). Age of presentation was highly variable, ranging from the neonatal period to the seventh decade of life. Although most patients presented subacutely with intermittent obstructive symptoms, at least one patient presented with colonic perforation and peritonitis. Treatment always involved resection of the severely diseased bowel. One case has been reported in association with Proteus syndrome, a rare disorder characterized by patchy hyperplasia of multiple tissues and organs but low risk of malignancy (10). The cause of this syndrome has recently been identified as a somatic (i.e. non-germline) genetic mutation in AKT1 that occurs during fetal development (11). It is possible that isolated intestinal lipomatosis arises from a similar mutation in mesenchymal cell lines of the midgut during fetal development.
In conclusion, symptomatic diffuse intestinal lipomatosis is a very rare condition with variable presenting symptoms, but can be readily diagnosed by cross-sectional imaging. Treatment for lesions causing symptomatic obstruction or intussusception is surgical resection. The etiology remains unknown, but may involve fetal somatic genetic mutations. Optimal long-term follow-up, aside from
instructing the patient to seek immediate medical care for recurrence of symptoms, remains undefined.

References

Figure captions
Figure 1: Multiple fat-density tumors in small bowel (left arrow) and colon (right arrow).
Figure 2: Long-segment intussusception of terminal ileum (open arrow) into distal transverse colon (solid arrows).
Figure 3: Near-total abdominal colectomy specimen, demonstrating patches of normal mucosa (open arrow), multiple submucosal fatty tumors (solid arrows), and hypertrophied epiploic appendages (solid arrowheads).

Figure 4: Proximal ileum specimen, with examiner’s hand holding large multilobulated submucosal fatty tumor.