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CASE REPORT

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Congenital short-gut syndrome

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Abstract A case of congenital short gut is reported in a 4-month-old boy presenting with failure to thrive. Upper gastrointestinal examination (UGI) with small bowel follow-through (SBFT) demonstrated dilation of the duodenum and jejunum, with rapid transit to rectum. On barium enema (BE), rapid transit of barium was noted to the dilated proximal small bowel seen on UGI/SBFT. Neither study delineated the ileocecal region, and the overall length of bowel appeared short. The diagnosis of congenital short gut was confirmed at surgery. Congenital short-gut syndrome is a rare entity. The diagnosis can be suggested by imaging, but is usually confirmed operatively.

Keywords Barium enema
Congenital short-gut syndrome ·
Infant · Malrotation · Upper
gastrointestinal examination

Introduction

Congenital short-gut syndrome is a rare anomaly. The diagnosis can be suggested on a contrast examination. We present a patient with this entity in whom the diagnosis was suggested by imaging studies of the gastrointestinal tract and confirmed by surgery.

Case report

The patient, a male, was born at 37 weeks' gestation by uneventful spontaneous vaginal delivery. Two previous children of the same parents were normal. There was no family history of congenital gastrointestinal tract anomalies. The mother was treated for hyperthyroidism with levothyroxine sodium (Synthroid) during pregnancy. The infant was also treated with antibiotics after birth

for group B streptococcus positivity in his mother. He had jaundice that lasted 3–4 weeks. There was no history of any infections or diarrhea, but the child did have significant gastroesophageal reflux. The child was breastfed and the mother noted an increase in the frequency of nursing in the 2 weeks prior to presentation.

The patient presented to our institution at 4 months of age for work-up of failure to thrive. He weighed 3.8 kg (< third percentile for age) while his birth weight was 3.0 kg. A 0.17- kg weight loss had occurred since his 2-month well-child examination. On physical examination, the infant appeared somewhat cachectic. His abdomen was soft, non-tender, non-distended and without evidence of mass or organomegaly. The abdominal wall and umbilicus were normal. Normal bowel sounds were heard. Loosely formed stool was noted. Laboratory values showed acidosis (bicarbonate = 12 mEq/l), hyponatremia (sodium = 130 mEq/l) and hypokalemia (potassium = 2.5 mEq/l). Serum albumin was normal.

Supine and upright views of the abdomen (Fig. 1) showed diffuse gaseous distention of bowel loops. An upper gastrointestinal examination (UGI) and small-bowel follow-through (SBFT) showed marked dilation of duodenum and proximal jejunum with normal-caliber bowel beyond the proximal jejunum (Fig. 2). In the anteroposterior projection, the duodenal course appeared grossly normal, crossing the midline and extending cephalad to the left of the spine. Slow transit of the barium was noted through the dilated proximal bowel; however, once the contrast media had passed the dilated proximal jejunum, it quickly passed through the remaining gastrointestinal tract to the rectum. Anatomy of the distal small bowel, cecum, and colon was not well delineated because of overlying opacified loops of bowel.

An initial attempted barium enema was aborted as the infant suffered a brief respiratory arrest. He was readily resuscitated without sequelae. The exact cause of the respiratory arrest was unclear and was likely multifactorial. A subsequent barium enema was performed without complication. The enema failed to show a normal anatomic arrangement of small bowel and colon. The rectum, sigmoid and descending colon appeared relatively non-dilated. Additional loops of bowel on the left side of the abdomen were thought to represent colon. The cecum and ileocecal valve could not be identified. After transit through a relative short length of bowel, barium was noted to opacify partially the dilated proximal small bowel seen on preceding UGI/SBFT. An overhead film showed a small amount of barium refluxed into the stomach (Fig. 3). The possibility of a congenital short gut was considered with substantial uncertainty, given the rarity of the diagnosis.

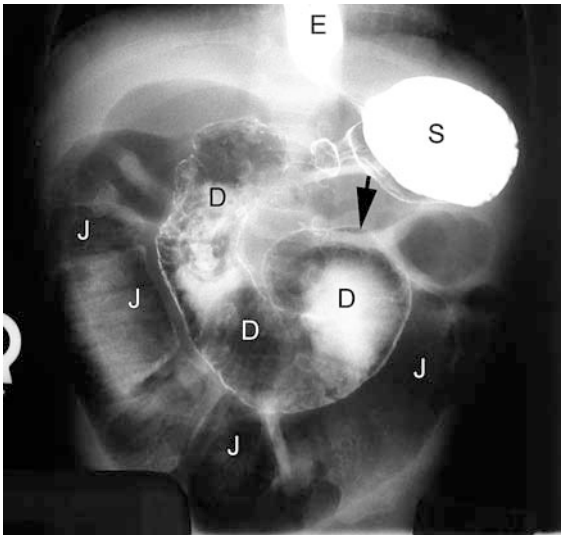

Fig. 2 Radiograph from small bowel follow-through shows moderate gaseous dilation of the distal duodenum (*D*). The duodenal-jejunal junction (*arrow*) appears to be in a relatively normal location, to the left of the spine and slightly inferior to pylorus; however, proximal jejunum (*J*) appears markedly dilated and extends back to the right upper quadrant. Gastroesophageal reflux is seen (*S* stomach, *E* esophagus)

An exploratory laparotomy confirmed the presence of a congenitally short small bowel. The ileocecal valve, cecum, appendix, and ascending colon were absent. The small bowel appeared diffusely dilated. The small bowel measured 56 cm from the duodenojejunal junction to where it connected with the transverse colon in an apparent autoanastomosis (Fig. 4). The duodenum was not fixed and passed anterior to the superior mesenteric artery and vein, indicative of a malrotation. Some adhesions were also noted. Additionally, a very tight stenosis was noted in the duodenum just distal to the pylorus due to Ladd's bands (no correlative finding was evident on the UGI). The bands were lysed, a gastroduodenostomy was performed, and a gastrostomy-tube was placed.

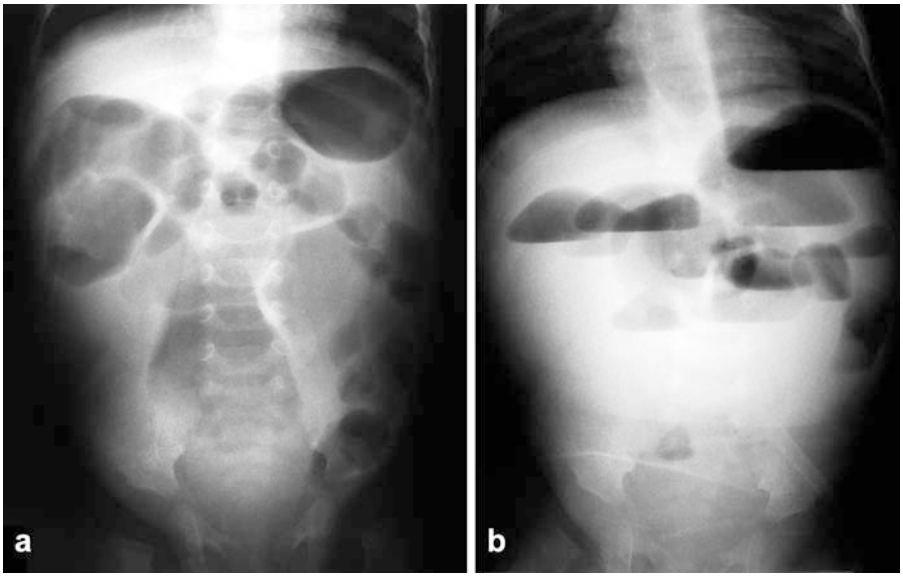

Fig. 1a, b Plain films of the abdomen. Supine (**a**) and upright (**b**) abdominal radiographs shows moderate gaseous distension of bowel loops in the mid-abdomen and right upper quadrant

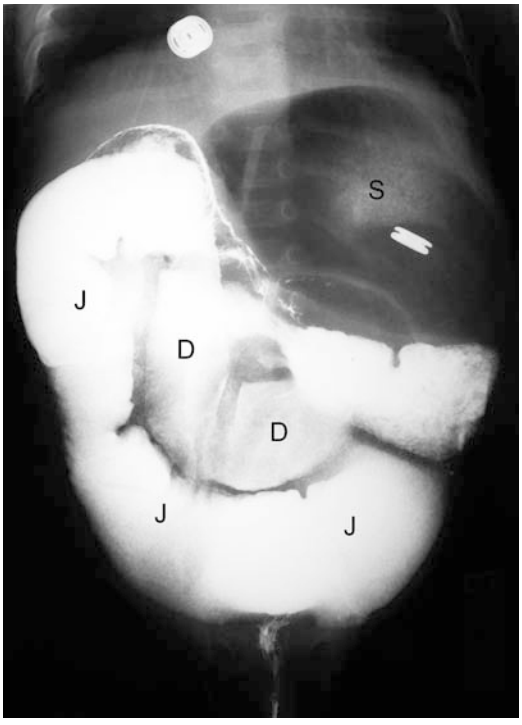

Fig. 3 Post-evacuation radiograph from barium enema shows barium had refluxed into dilated proximal jejunum (*J*) and duodenum (*D*). Faint barium is seen in the stomach (*S*). Most of the colonic barium has been evacuated or refluxed proximally

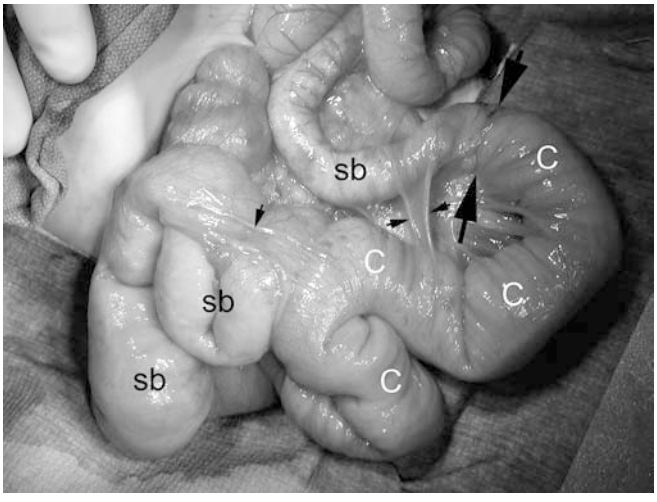

Fig. 4 Intraoperative photograph shows an autoanastomosis (*large arrows*) from small bowel to distal transverse colon. Adhesive bands (*small arrows*) arose from intrauterine events. Note the dilated appearance of small bowel (*sb*). (*C* colon)

Postoperatively, the infant was started on total parenteral nutrition (TPN) and followed by enteral feeds prior to discharge. He currently is on a mixture of parenteral and enteral nutrition, with the hope of tapering him off TPN. Since diagnosis, the infant has gained 4 kg in 8 months. During that interval, he has had nine hospital admissions, including four for catheter-related infection, three for dehydration and/or gastroenteritis, and one for presumed bacterial overgrowth.

Discussion

Short-gut syndrome in infants is most often acquired as a result of surgical resection of a diseased small intestine affected by multiple bowel atresias, gastroschisis, necrotizing enterocolitis, or malrotation with midgut volvulus and infarction. Acquired short-gut syndrome is relatively common. Congenital short-gut syndrome without atresias, on the other hand, is a very rare condition with relatively few reported cases. Some familial cases of congenital short-gut syndrome have been reported with autosomal recessive inheritance [1, 2, 3].

In a child with congenital short-gut syndrome, onset of symptoms within the first few weeks of life usually represents a more severe form of the entity and usually leads to a fatal outcome. A later clinical onset, after 2–3 months of age, bears a more favorable prognosis, although, in general, the diagnosis usually has a dismal outcome [2]. As with acquired short-gut syndrome, early use of TPN may improve prognosis and advancements in TPN care have improved outcome [4]. Small-bowel transplants for short-gut syndrome have been attempted with limited success, but offer promise [5].

Several theories have been postulated to explain the intrauterine development of short gut; however, the embryologic basis of the defect still remains obscure. Many reported cases have associated malrotation [1, 2]. Intrauterine volvulus may lead to ischemia and cause disturbances of growth and function of the developing small bowel [1]. Additionally, in utero infarction of bowel could lead to resorption and a decreased length of the remaining gut. This may be the etiology in our patient based on the surgical findings of malrotation and an apparent autoanastomosis. Other etiologic theories have focused on a defective neuroenteric system. For instance, Tanner and co-workers noted an abnormality of the myenteric plexus neurons in their postmortem study of children with congenital short-gut syndrome [6]. Others, however, have noted a normal myenteric plexus in children with congenital short-gut syndrome [7]. A mechanical etiology has been proposed for this rare disorder. Hamilton et al. suggested that the primitive digestive tube could not be accommodated in the intraumbilical space and remained in the abdominal cavity throughout gestation, resulting in a short small bowel [8]. Conversely, others have reported congenital short-gut syndrome associated with delayed return of fetal gut into the abdomen due to gastroschisis [9]. In these cases, it was speculated that extra-abdominal volvulus and infarction resulted in autolysis and reabsorption of the

bowel, rendering it short [9]. Malrotation would be an expected accompanying feature [9].

Clinically, infants with congenital short gut syndrome present with vomiting, diarrhea and, as in our case, failure to thrive. This syndrome complex is due to underlying short length of bowel, dilation, and hypo- or dysmotility. Acidosis, hyponatremia, and hypokalemia, as seen in our patient, occur as a consequence of losses owing to decreased absorptive capacity of the gut. Dilation of the small bowel has been attributed to a compensatory mechanism of the gut attempting to increase its absorptive capacity, as is also noted in acquired shortening of the small bowel [9]. Interestingly, hypertrophic pyloric stenosis has been reported in several cases of congenital short-gut syndrome [2].

Radiographic findings of congenital short gut syndrome have not been described in detail because of the sporadic reports of this rare diagnosis. In general, findings are non-specific; however, the diagnosis may be suggested if enteric contrast studies demonstrate short length of the gut [2, 3, 4, 7, 10]. Plain radiographs show non-specific bowel dilatation [1, 7, 10, 11]. In cases with underlying malrotation, UGI may diagnose malrotation, but other findings are often non-specific [2, 3, 9, 10]. Often, there is rapid transit of barium through small bowel [2]. Dilation may suggest obstruction. Findings on barium enema may include a rapid retrograde transit through shortened bowel to the stomach [4]. In cases with underlying malrotation, the enema may show a malpositioned cecum [7, 11].

The mainstay of management of short-gut syndrome is intravenous alimentation along with very gradual advancement of oral feedings [1, 4, 10]. This allows the small bowel to improve its functions of absorption and motility. TPN is necessary, but its complications often contribute to significant morbidity and mortality.

In conclusion, we present a case of congenital short-gut syndrome, a rare entity. The diagnosis was suggested on preoperative contrast studies of the gastrointestinal tract. Operative findings suggest the short-gut syndrome was a consequence of malrotation with in utero volvulus, bowel infarction and autoanastomosis.

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