

Case Report

CONGENITAL SHORT BOWEL SYNDROME

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Abstract

Congenital short bowel syndrome (SBS) is a rare congenital disease. Only about 37 cases were reported. Infants usually present with failure to thrive, recurrent vomiting and or diarrhoea. It is associated with significant mortality and morbidity. High degree of suspicion is necessary to diagnose this condition promptly. Early initiation of parenteral nutrition or surgery may result in a favorable outcome. We treated a case of infant aged eight-weeks presented with recurrent bilious vomiting and failure to thrive. On Laparotomy it was found that the baby had very short small bowel with malrotation. This case was reported and reviewed.

Key Words: Congenital malrotation, short bowel syndrome, Intestinal malabsorption.

Introduction

Short bowel syndrome (SBS) is a clinical disorder of functional or anatomical loss of enough small bowel length to markedly compromise intestinal absorptive capacity that is characterized by diarrhea and or malabsorption. Most often it is recognized as an acquired disorder following surgical bowel resection for conditions such as necrotizing enterocolitis or intestinal atresia in neonates. In rare instances, the SBS may be congenital in origin, with the neonatal bowel being short in length at birth.

Case report

An eight-week old girl presented with recurrent bilious vomiting and failure to thrive. She was the second issue of a term pregnancy from nonconsanguineous parents and her delivery was normal at home. Her birth weight was 3 kg. She was exclusively breastfed. Ten days later, she developed irregular vomiting,

frequently it was bilious. Gradually the baby lost her weight and during surgical consultation her weight was 1.5 kg only. Mother described her bowel habit as normal. Otherwise, she appeared clinically well, with no dysmorphic features, jaundice, ascites, edema, rash or mouth lesions. Her abdomen was soft, with only mild distension. Visible peristalsis was noted in upper abdomen. There was no organomegaly. Laboratory studies demonstrated hemoglobin levels at 8.0 g/dl, white blood cell count at $10.0 \times 10^9/L$, with a normal differential count and platelet count. Serum electrolytes were within normal limit. A plain film of the abdomen was unremarkable. An upper gastrointestinal (GI) water soluble contrast studies revealed malrotation with the duodenojejunal flexure positioned in the right midabdomen and proximal dilated small gut only. Preoperative blood transfusion was given. On 6 september 2010, during laparotomy, malrotation was noted and the small bowel length was measured to be 40 cm from the duodenum to the ileocecal valve. Ladd's operation was done. Postoperatively, she received parenteral fluid and blood transfusion. Gradual oral feeding with breast milk was started from third post operative day. Post-operatively she gained 0.8 kg weight within 15 days. Due to poor wound healing, her wound dehiscence needed mass closure of wound by a second operation.

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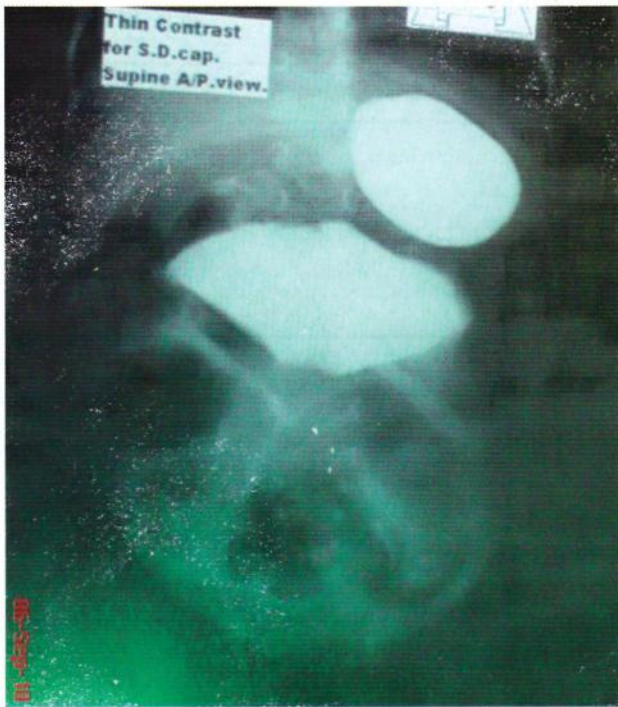


Fig.-1: Contrast upper GIT X ray- evidence of upper GIT obstruction



Fig II: Congenitally short small bowel

Discussion:

Congenital Short bowel syndrome (SBS) is a rare condition, with only 37 cases reported in the English literature¹. The length of the small intestine, as measured from the Treitz ligament to the ileocecal valve, has been shown to correlate with crown-to-heel length. During somatic growth, as the crown-to-heel length increases from 40 cm to 100 cm, the small bowel increases from 164 ± 54 cm (mean \pm SD) to 425 ± 90 cm². Thus, the mean length of the small intestine in full-term newborns is approximately 240 cm, increasing to 430 cm by 15 years of age³. In preterm infants, the expected jejunoileal length correlates with gestational age, with the bowel length expected to double from 28 to 40 weeks gestation⁴.

In full-term neonates, SBS may manifest when the small bowel length is less than 75 cm⁵. Newborns with congenital SBS may have a significantly shortened small bowel, with intestinal lengths reported as short as 20 cm. In the present paper, the patient had a small bowel length of 40 cm, measured at laparotomy.

During normal embryogenesis, midgut lengthening begins in the fifth week of gestation, initially forming a loop that extends into the umbilical celom. Following a series of rotations and to accommodate further intestinal elongation and growth, the bowel returns from its ventral herniation to the abdominal cavity at approximately 10 weeks gestation. Because congenital SBS occurs most often in association with malrotation, Hamilton et al¹ suggested that the normal elongation, rotation and herniation of the small intestine is interrupted or delayed because of a lack of space between the developing digestive tube and the umbilical celom. The etiopathogenesis of congenital SBS is poorly understood. Some have postulated an ischemic injury to the developing intestine; however, the patency of the bowel lumen and the presence of an intact mesentery in these cases would argue against an in utero bowel infarction. Still others have focused on the possibility of defective neuroenteric development, especially because intestinal dysmotility is recognized as an important component of the congenital SBS⁶. Several cases, including those in the original report by Hamilton et al¹ have occurred in a familial pattern⁷. Cases of congenital SBS have been reported in association with other congenital anomalies. No specific gene mutations have yet been identified for congenital SBS⁷⁻⁹.

Most infants with congenital SBS present with vomiting, diarrhoea and failure to thrive, or signs and symptoms consistent with intestinal obstruction. While malrotation is an almost universal feature (present in 96% of cases), associated volvulus with acute bowel infarction is an infrequent finding, perhaps because the short bowel length prevents significant intestinal twisting and subsequent ischemia. The typical presentation of failure to thrive and recurrent upper GIT obstruction as in our patient is likely a direct consequence of the short bowel length, malabsorption and malrotation.

The cornerstone to the management of these cases is perenteral nutrition (PN) with early introduction and advancement of enteral feedings¹⁰. While PN is necessary to provide adequate calories for growth, the delivery of nutrition parenterally has potential

complications, with sepsis being a leading cause for morbidity and mortality. While the mechanisms for small bowel adaptations are multiple and complex, early and aggressive introduction of enteral feeding is a critical component¹⁰⁻¹².

Of the cases with congenital SBS reported in the literature, one-third of the patients have survived to a mean age of 5.8 years (range, one to 18 years). Two-thirds had died, with most of the deaths having occurred early in life, well before one year of age (mean age of death was 84 days [range, one day to seven months]). Since the year 2000, reports have shown significantly improved survival rates for these patients. These enhanced outcomes are likely due to improvements in the overall care and management of PN therapy as well as the development of novel infant formulas that both facilitate the early introduction of enteral feeding and promote intestinal adaptation^{6,13,14}.

Conclusion

Congenital SBS is a rare condition of the newborn that presents with chronic diarrhea, failure to thrive or symptoms of small bowel obstruction in the first weeks of life. The diagnosis of congenital SBS is typically established at surgical laparotomy. Our patient, with 40 cm of small bowel, was able to successfully initial wean off. Long-term survival of children with congenital SBS is now possible if adequate calories are provided to stimulate growth, aggressive enteral feeds are introduced early and close monitoring is provided.

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