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Congenital Small Bowel Syndrome

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Authors' contributions

This work was carried out in collaboration among all authors. Authors SAH and HAA were responsible in managing the patient, supervising the study and reviewing it. Authors NAH and MM collected the data, managed the literature searches, and wrote the manuscript. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Congenital short bowel syndrome (CSBS) is a rare entity of short bowel syndrome, with only 37 cases reported in literature till this date, with numerous reports of its high morbidity and mortality rate.

We present a case of a 7-week-old boy who presented with abdominal distension, recurrent vomiting, and chronic diarrhea. The diagnosis of CSBS was confirmed by exploratory laparotomy, and was suggested by imaging studies of the abdomen and upper gastrointestinal tract. Post-operatively, the patient was started on total parenteral nutrition for a month, but subsequently passed away as a result of disseminated sepsis.

Keywords: Congenital; short bowel syndrome; malabsorption; nutrition.

1. INTRODUCTION

Short bowel syndrome (SBS) is a malabsorptive disorder that results from functional or anatomical loss of small bowel length. This loss is usually acquired due to extensive resection of the bowel for necrotizing enterocolitis, congenital intestinal atresia, volvulus, or extensive Hirschprung's disease. Congenital short bowel syndrome (CSBS), on the other hand, is a rare

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condition affecting neonates. It is associated with a high morbidity and mortality rate, as a result of a decrease in absorptive surface area and changes in bacterial flora, which lead to malnutrition, failure to thrive, and chronic diarrhea which can be severe [1]. The normal length of the small intestine in an infant is around 250 cm, measured from the Treitz ligament to the ileocecal valve. Infants with CSBS have a significantly shorter small bowel, with clinical manifestations appearing when the length is less than 75 cm [2]. The exact etiology and pathogenesis of CSBS is unknown, with several proposed theories. The theories range from interrupted or delayed elongation and rotation of the bowel, to defective neuroenteric development with subsequent intestinal dysmotility, to a postulated in-utero ischemic injury with CSBS representing the sequel of necrotizing enterocolitis [2].

Familial occurrence of CSBS, mainly affecting siblings, has been noted in approximately 60% of reported cases. Recently, the genetic basis of CSBS has been attributed to mutations in the CLMP gene and FLNA gene, causing the recessive form and X-linked form of the disease respectively [3].

Cases of CSBS have been associated with several abnormalities, including pyloric stenosis, appendiceal agenesis, dextrocardia and the most common being intestinal malrotation.

Hasosah et al (2008), reviewed all published cases of CSBS and noted that all but one was associated with intestinal malrotation. [2] In another study, by Huysman et al. (1991) intestinal malrotation in the context of CSBS was associated with a poor outcome, with only 3 out of the 16 patients with the mentioned abnormalities surviving [4]. The associated malrotation seen can either be due to an independent developmental abnormality, or it may be a consequence of the shortened small intestine. It is worth noting that in our patient, dextrocardia was present, however intestinal malrotation was not.

2. CASE PRESENTATION

A 7-week-old Bahraini baby boy, presented with abdominal distension, recurrent vomiting, intolerance to feed, and chronic watery diarrhea. He was the product of a normal vaginal delivery, from non-consanguineous parents at 31 gestational weeks, with an Apgar score of 9 and 10, at 1 and 5 minutes after birth, respectively. Birth weight was 2.850 kg. A history of dilated bowel loops on antenatal scan was reported. No history of delayed passage of meconium. On the fourth post-natal day, he developed the first symptom of watery, non-bloody, diarrhea which continued till presentation. In the ensuing weeks, the other mentioned features manifested, despite breastfeeding and trying different formula feeds.

On examination, the child appeared dysmorphic, and mildly dehydrated. The abdomen was soft and distended, with no organomegaly. Bowel sounds were present and normal. Groin and genitalia were normal. System examination was unremarkable, apart from noted dextrocardia. Vitals were within normal limits.

Laboratory investigations revealed a: white blood cell count of 5.24 x10^9/L, hemoglobin of 8.9 g/dL, platelet count 156 x10^9/L, triglycerides 3.4 mmol/L, normal liver function tests, sodium 137 mmol/L, potassium 3.6 mmol/L, chloride 103 mmol/L, and a bicarbonate of 15 mmol/L. Blood cultures were sterile.

Plain radiograph of the chest and abdomen was remarkable for diffuse bowel dilatation and dextrocardia. [Fig. 1] An upper gastrointestinal study showed dilated small bowel, normal large bowel, and rapid transient time from small bowel to rectum.

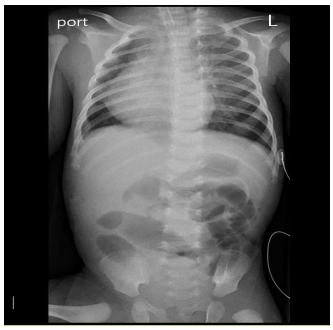

Fig. 1. Plain radiograph of the abdomen

Due to persistent vomiting and abdominal distension, the patient underwent exploratory laparotomy, which revealed the abnormal finding of dilated small bowel with a length of 32 cm. The large bowel was normal, there were no signs of obstruction or malrotation. [Fig. 2] [Fig. 3]. An appendectomy was performed and reported to be normal following histological examination.

Post-operatively, the patient was kept on total parenteral nutrition for a month. However, in his hospital stay, he succumbed to disseminated

Fig. 2. Intraoperative finding 1

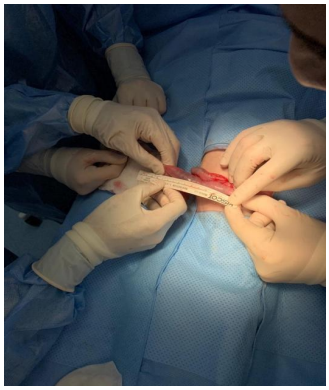

Fig. 3. Intraoperative finding 2

sepsis with secondary hemolytic uremic syndrome.

3. DISCUSSION

The diagnosis of CSBS relies on clinical, radiological, and surgical findings. Clinically, infants present with symptoms of malabsorption such as failure to thrive, vomiting, and chronic diarrhea. The severity of the symptoms correlates with the degree of decrease in intestinal absorptive surface area [1]. Although the diagnosis can be reached by an upper GI small bowel follow-through study, which can measure small bowel length and assess for the presence of malrotation, exploratory laparotomy is typically performed to confirm the diagnosis. The necessity of this lies in the fact that radiographic assessment of SBS has been reported to underestimate intra-operative intestinal length [5]. Early diagnosis and management of short bowel syndrome is essential to achieve favorable outcomes.

The foundation of management of SBS is nutritional support with early initiation of parenteral nutrition and advancement of enteral feedings. CSBS is considered to be a type of intestinal failure with resultant failure to sustain growth, hydration, and electrolyte homeostasis. Thus, compensating for the nutrients and caloric intake in the form of total parenteral nutrition (TPN) is a life-saving therapy in neonates. However, TPN is frequently associated with complications mainly intestinal failure-associated liver disease (IFALD) and catheter-associated bloodstream infections (CABSI). In IFALD, the deleterious effects to the liver are multifactorial and include TPN toxicity, recurrent sepsis and prematurity. Whereas in CABSI, the portal of entry of infection is the central venous catheter through which TPN is administered. CABSI, as in the case of our patient, is a major complication that can be life-threatening. In an attempt to decrease the risk of CABSI, other authors utilized ethanol locks, which penetrate the biofilm that forms on central lines, with promising results [6].

Weaning the patient from TPN therapy is a clinical decision that can be supported with laboratory investigations such as measuring levels of serum citrulline concentration. Citrulline is a product of the intestinal mucosa. Thus, its levels reflect mucosal mass, and the ability to wean TPN. A level persistently <12 mmol/L does not encourage weaning TPN [7]. However, it is important to note that growth and nutritional requirements are a dynamic process and so regular assessment of the ability to wean TPN is needed as the infant grows. It has been noted by other authors that the transition to enteric feed, aggressively and as early as possible, is a critical component in encouraging intestinal adaptation which leads to improved outcomes, and prevents the complications of TPN mentioned above [2][8]. In the case of our patient, a trial of enteric feed was not possible to undertake, given the clinical condition of the patient.

In a selected population of neonates with CSBS, refractory to medical treatment, a new bowel lengthening procedure called serial transverse enteroplasty operation (STEP) was introduced in 2003 [7]. The procedure aims to increase the length of the small bowel by extending pre-existing tissue to increase intestinal surface area to enhance the absorption process. It relies on the anatomy of the small bowel and its blood supply which comes from the mesenteric border and traverses the bowel perpendicular to its long axis. An endoscopic stapler is passed through the mesentery, with all staple lines kept parallel to the plane of the blood supply, and a zig zag

lengthening and tapering of the intestine proceeds. This results in a small bowel that is longer in length and smaller in diameter [6][9]. Clinically, in successful cases, neonates are able to tolerate and thrive on enteric feed. Other surgical options include combined liver-intestinal transplantation. However, this is usually reserved for neonates with concomitant irreversible life-threatening hepatic failure.

4. CONCLUSION

In conclusion, congenital short bowel syndrome is a rare entity of short bowel syndrome. It manifests in the neonatal period with features of malabsorption, mainly failure to thrive and chronic diarrhea. It is essential to diagnose and manage this condition early, since studies have shown that early initiation of enteral feed promotes intestinal adaptation and hence improves the long-term survival and the quality of life of these patients.

CONSENT

All authors declare that 'written informed consent was obtained from the patient and the parents for publication of this case report and that maximal patient anonymity was ensured and maintained during the writing of the case report.

ETHICAL APPROVAL

As per international standard guideline written ethical approval has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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