

Carmi syndrome associated with gastric perforation

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Congenital pyloric atresia with epidermolysis bullosa known as Carmi syndrome is a rare condition and is associated with a high mortality rate. Carmi syndrome complicated by gastric perforation is extremely morbid and unusual, with a fatal outcome. Here, we report a case of Carmi syndrome in a neonate who presented with an uncommon complication of gastric perforation. *Ann Pediatr Surg* 9:122–123 © 2013 Annals of Pediatric Surgery.

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Introduction

Isolated congenital pyloric atresia (CPA) is a rare malformation with a fairly good outcome [1]. CPA with epidermolysis bullosa (EB), also known as Carmi syndrome, is still a rare condition with an autosomal recessive inheritance, although its prevalence is unknown [2,3]. Neonatal gastric perforation (NGP) as an isolated case is also an uncommon condition associated with CPA [4]. Here, we report these three uncommon conditions, that is, CPA and EB with gastric perforation in a neonate, with a brief review of the literature. Such associations have rarely been reported in our literature search [5].

Case report

A 7-day-old boy (full term) presented with a history of abdominal distension and vomiting of 2 days' duration. On physical examination, the baby weighed 2.62 kg, appeared sick looking, and was dehydrated, with poor activity. The abdomen was grossly distended and tense. Radiograph of the abdomen showed gross pneumoperitoneum. Routine blood investigations were normal. The child was started with intravenous fluid, nasogastric aspiration, and antibiotics – piperacillin with tazobactam, amikacin, and metronidazole. Emergency laparotomy indicated a large perforation of about 5 cm on the anterior wall of the stomach near the greater curvature, with evidence of peritonitis. Pyloric atresia type-1 was also found.

Heineke-Mikulicz pyloroplasty, transgastric feeding jejunostomy, and gastrorrhaphy were carried out. On the third postoperative day, skin blisters were observed on the hands and patches of raw areas on the back (Fig. 1). Dermatologist opinion, followed by skin biopsy report confirmed the diagnosis of EB. Postoperatively, the child was managed with intravenous fluid and blood products, along with the antibiotics. Feeding through a jejunostomy tube was started on the fifth postoperative day. Oral feeding was started 1 week later. The baby recovered well and was discharged. On follow-up, baby was doing well 3 months later, tolerating feeds, gaining weight, and had minimal skin lesions.

Discussion

CPA is a rare malformation that constitutes less than 1% of all upper gastrointestinal atresia. Its incidence is reported to be 1 : 100 000 births [1]. It was first described by Calder in 1749 [6]. Although CPA occurs commonly as an isolated lesion, which has an excellent prognosis, not uncommonly, it is seen in association with either hereditary multiple intestinal atresia syndrome or EB and/or aplasia cutis congenital, which can have a negative impact on the final outcome [1,6].

EB is a rare autosomal recessive form of genetic disorder caused by mutation of the keratin gene. This disorder is characterized by the presence of extremely fragile skin and recurrent blister formation of the skin and mucous membrane resulting from minor mechanical friction or trauma. Its incidence is about 1/300 000 births [2,5]. EB is divided into three major types: EB simplex, dystrophic EB, and junctional EB [3]. EB with CPA is extremely rare, and its prevalence and incidence are unknown. The association of CPA and EB was first described by

Fig. 1



Back of the patient showing patches of raw areas in epidermolysis bullosa.

Swinburne and Kolher [7]. EB-CPA is usually lethal but nonlethal forms have also been reported. The long-term prognosis of EB-CPA depends on the severity of cutaneous manifestations.

Although most affected children succumb as neonates, those who survive may have blistering with the formation of granulation tissue on the skin, around the mouth, nose, fingers, and toes and internally around the trachea. However, some affected individuals have little or no blistering later in life [3].

NGP is a rare but frequently fatal condition of uncertain etiology. NGP can be categorized as spontaneous, ischemic, and traumatic. Traumatic NGP is usually the result of gastric tube insertion or attempted intubation [8]. The term spontaneous suggests a cause separate from necrotizing enterocolitis, ischemia, trauma from gastric intubation, distal intestinal obstruction, or accidental insufflation of the stomach during assisted ventilation [9]. NGP represents an immediate surgical emergency. The mortality rate of NGP is high, overall being 47% [4]. The most important factors affecting survival appear to be the onset of symptoms and the start of the definitive therapy, the extent of peritoneal contamination, the degree of prematurity, and the severity of other associated consequences of asphyxia. Very few cases of gastric perforation with CPA are reported in the English literature [10]. CPA associated with two fatal conditions, namely EB and gastric perforation, obviously has a very high mortality rate. All NGPs should be explored thoroughly during surgery for any distal intestinal obstructions such as CPA, duodenal atresia, annular pancreas, malrotation, and intestinal atresia, although intestinal obstruction is seldom the cause of NGP [11].

Conclusion

Carmi syndrome, although a fatal condition with a fatal outcome, should be operated. NGP, irrespective of its etiology, is a surgical emergency. All NGP during surgery should be explored to identify any distal obstruction. In case of NGP with distal obstruction, gastrorrhaphy with definitive surgery for distal intestinal obstruction should be decided on the basis of the degree of peritonitis for a favorable outcome.

Acknowledgements

Conflicts of interest

There are no conflicts of interest.

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