Pediatr Surg Int (1999) 15: 264–265

CASE REPORT

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Pyloric atresia associated with epidermolysis bullosa, malrotation, and high anorectal malformation with recto-urethral fistula: a report of successful management

Accepted: 26 May 1998

Abstract Pyloric atresia (PA) is an uncommon anomaly that may be associated with many other congenital anomalies, the commonest of which is junctional epidermolysis bullosa (JEB). Most of the cases of PA associated with JEB (Herlitz syndrome) reported have been fatal. A case of PA associated with JEB, malrotation, and a high anorectal malformation with a rectourethral fistula, which was hitherto undescribed, was successfully managed at our institution.

Key words Pyloric atresia · Epidermolysis bullosa · Malrotation · High anorectal malformation

Introduction

Pyloric atresia (PA) is an uncommon variety of intestinal obstruction [9]. It is usually an isolated finding, but may be associated with many other congenital anomalies, the commonest of which is junctional epidermolysis bullosa (JEB) [8, 9]. Most reported cases of PA associated with JEB (PA-JEB) have been fatal [3, 4, 6, 16], with a few reported survivors [7, 12, 18]. A case of epidermolysis bullosa (EB) with pyloric, esophageal, and anal atresia has been described [3]. We observed a case of PA-JEB in a child who survived and also had a malrotation and a high anorectal malformation (HARM) with a rectourethral fistula (RUF), an association that has not been previously described with PA-JEB.

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Case report

A 1.5-kg male was born at term of a consanguineous marriage with an absent anal opening. An antenatal ultrosound (US) scan done at 32 weeks of gestation because of the presence of polyhydramnios had detected a "duodenal atresia." Plain abdominal X-ray films done at 10 h of age showed a distended stomach shadow with no distal gas. A laparotomy revealed PA and a malrotation. The pylorus was obstructed by a 4-mm-thick membrane (type I). Excision of the pyloric membrane and a Heineke-Mikulicz-type pyloroplasty, gastrostomy, Ladd's procedure, and loop sigmoid colostomy was performed. A nasogastric (NG) tube was positioned across the anastomosis during surgery.

On the 2nd day of life the baby developed multiple bulla-like skin lesions, which started over the pressure points and spread all over. Electron microscopy of a skin biopsy from the edge of one such lesion confirmed the diagnosis of JEB. NG feeding was started on the 2nd postoperative day and oral feeding was instituted on the 5th day, which was tolerated well. The skin lesions healed without scarring over the next 2 weeks. Subsequent US scans revealed a normal genitourinary tract, and a cologram showed the blind rectal pouch ending proximal to the pubococcygeal (PC) line with a RUF. The patient had two further episodes of bullous eruptions in the follow-up period, which were managed conservatively. He subsequently underwent a posterior sagittal anorectoplasty and colostomy closure, and has been well thereafter.

Discussion

Since the first report of PA in 1937 [2] and the first successful repair in 1940 [1, 7], a lot has been written about this rare congenital anomaly, which forms 1% of all gastrointestinal atresias [9] and still carries a high overall moratlity. Many cases of familial occurrence [7, 11, 13, 14] with probable autosomal recessive inheritance [1, 5–7] have been reported. Associated anomalies have been reported in 30% of cases of PA [8, 9], of which JEB is the commonest [4, 8, 9, 15]. Since both PA and EB are rare autosomal disorders, the association between EB and PA is probably genetic rather than coincidental [7]. The report of an association of pyloric, esophageal, and anal atresia with EB [3] has led some authors to believe that in patients with EB the esophageal, gastric, intestinal and colonic mucosa are affected.

The favored treatment of type I PA is excision of the membrane and a Heineke-Mikulicz-type pyloroplasty [6–8]. Recently, a transgastric approach without a pyloroplasty has also been described [10]. Most deaths are now due to delayed diagnosis [1, 6], leading to severe dehydration with hypochloremic alkalosis, or because of associated

anomalies like JEB and multiple intestinal atresias [5, 15]. PA-JEB has been reported to be so uniformly fatal that nonoperative management of such cases has been advocated by some [13], but recently a few survivors have been reported [6, 11, 17]. Neither malrotation nor a HARM with RUF has previously been reported in association with PA-JEB.

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