Turkish Journal of Trauma & Emergency Surgery
Case Report
Ulus Travma Acil Cerrahi Derg 2012;18 (3):271-273
Olgu Sunumu
doi: 10.5505/tjtes.2012.13284

# Pyloric atresia associated with epidermolysis bullosa: report of two cases and review of the literature

## Pilor atrezisi ve epidermolizis bülloza birlikteliği: İki olgu sunumu ve literatür derlemesi

**Ünal BIÇAKCI,[1] Burak TANDER,[1] Fatma ÇAKMAK ÇELİK,[2] Ender ARITÜRK,[1] Rıza RIZALAR[1]**

The coexistence of pyloric atresia (PA) and epidermolysis bullosa (EB) is a rare but well-known surgical emergency in neonates. PA/EB is described by the association of atresia of the pylorus and bullous lesions on the skin. Ninety-one cases have been reported in the literature to date. We present two new cases and evaluate the association of PA/EB, its etiopathogenesis and the clinical properties. Case 1: A three-day-old female presented with nonbilious vomiting and bullous lesions 2-3 cm in diameter on the extremities. Abdominal X-ray showed a single air-fluid level in the left upper quadrant. At laparotomy, we found PA and performed a pyloro-pylorostomy. The patient died due to sepsis complication of EB two months after surgery. Case 2: A two-day-old male presented with severe dermal bullous lesions on the trunk, neck and extremities. His stomach was dilated and there was no gas distally. We found PA and performed gastroduodenostomy. Initially, he tolerated the feeding well, but he died due to severe sepsis on the postoperative 23rd day. Almost all neonates born with the PA/EB result in a fatal outcome in the first few years. The complications related to EB are usually the cause of death. Even after successful repair of PA, skin lesions lead to death due to infection.

***Key Words:*** Epidermolysis bullosa; Carmi syndrome; pyloric atresia.

Pilorik atrezi (PA) ve epidermolizis bülloza (EB) birlikteliği genetik etyolojisi iyi bilinen fakat ender rastlanılan yenidoğanın acil cerrahi hastalığıdır. PA/EB, pilor atrezisi ile cilt ve mukozalardaki bülloz lezyonların beraberliği olarak tariflenir. Literatürde bugüne dek toplam 91 olgu bildirilmiştir. Bu yazıda iki yeni olgu sunuldu, literatür taraması yapıldı. Olgu 1: Üç günlük hasta safrasız kusma ve ekstremitelerde 2-3 cm çapında bülloz lezyonlarla başvurdu. Karın grafisinide sol üst kadranda hava-sıvı seviyesi vardı. Laparotomide pilor atrezisi saptanarak piloropilorostomi uygulandı. Sorunsuz taburcu edilen hasta iki ay EB'ye bağlı ağır sepsisten kaybedildi. Olgu 2: İki günlük hasta, boyunda, diz altında, ön yüzde, el sırtında ve sol dirsek üstünde yaygın cilt lezyonları mevcuttu. Karın grafisinde, dilate mide ve distalinde gaz yokluğu dikkati çekti. Pilor atrezisi ön tanısı ile ameliyat edilerek gastroduodenostomi uygulandı. Başlangıçta oral beslenmeyi iyi tolere eden hasta ameliyat sonrası 23'üncü cilt lezyonlarına bağlı ağır sepsis nedeniyle kaybedildi. PA/EB tanısı alan hastaların hemen hemen hepsi ilk birkaç yıl içinde kaybedilmektedir. EB'den kaynaklanan komplikasyonlar ölümlerin en yaygın nedenidir. Başarılı PA ameliyatlarından sonra bile, EB'ye bağlı cilt lezyonları, enfeksiyon ve metabolik problemlere yol açarak, ölümleri neredeyse kaçınılmaz hale getirmektedir.

***Anahtar Sözcükler:*** Epidermolizis bülloza; Carmi sendromu; pilor atrezisi.

The association of pyloric atresia (PA) and epidermolysis bullosa (EB) is very rare, but is a life-threatening emergency in neonates. Its genetic background is well defined.[1,2] Surgical intervention for PA is effective only for short-term survival. Long-term outcome of PA/EB is almost always poor and most patients are lost due to complications related to EB. Ninety-one cases of this rare syndrome have been reported in the literature thus far.[3,4]

We report here two additional cases of PA/EB and review the related literature.

Departments of [1]Pediatric Surgery, [2]Neonatology, Ondokuz Mayis University Faculty of Medicine, Samsun, Turkey.
Ondokuz Mayıs Üniversitesi, Tıp Fakültesi, [1]Çocuk Cerrahisi Anabilim Dalı, [2]Neonatoloji Bilim Dalı, Samsun.

Correspondence *(İletişim):* Burak Tander, M.D. Ondokuz Mayıs Üniversitesi, Çocuk Cerrahisi Anabilim Dalı, Kurupelit 55139 Samsun, Turkey.
Tel: +90 - 362 - 312 19 19 / 3510 e-mail *(e-posta):* btander@omu.edu.tr

## CASE REPORTS

***Case 1-*** A three-day-old female was admitted to the emergency department with nonbilious vomiting on her third day of life (33 weeks gestation, 2000 g, spontaneous vaginal delivery). She had bullous lesions 2-3 cm in diameter on her extremities and peeling dermal lesions on her right arm and left wrist. Abdominal X-ray depicted prominent gastric air and lack of air distally. No intraabdominal calcifications were identified (Fig. 1). PA was found at laparotomy. A pyloro-pylorostomy described by Dessanti et al.[5] was performed, which provides the preservation of the pyloric sphincter. She was fed by trans-anastomotic tube on the second day of surgery. The postoperative course was uneventful, and she was discharged on the ninth day after operation. In the follow-up studies, feeding was well tolerated and bullous lesions had improved. After two months, however, she was admitted to our emergency department with increased bullous lesions of the skin in number and size, electrolyte imbalance (Na: 119 mmol/dl, K: 7.5 mmol/dl), findings of sepsis (tachypnea, tachycardia, hypotension, WBC: 23,000, body temperature: 38.7ºC), and dehydration. Some of the lesions of the skin contained purulent material. There were numerous bullous lesions 1-2 cm in diameter on the trunk and extremities and peeling dermal lesions covering the body. She had no feeding problem. Abdominal radiographic examination revealed no specific finding. Her clinical status deteriorated within a few hours and despite all medical efforts, the patient died of severe sepsis induced by EB.

***Case 2-*** A two-day-old male was admitted to our newborn intensive care unit due to extensive dermal lesions (35 weeks gestation, 2200 g, cesarean section). Dermal lesions were severe bullous type on the trunk, neck, and extremities (Fig. 2). He had nonbilious vomiting and was dehydrated. On X-ray examination, his stomach was dilated and there was no gas distally. The oral contrast X-ray study revealed dilated stomach and no distal passage. At laparotomy, we found PA and performed gastroduodenostomy. Oral feeding was started on the seventh day postoperatively and was well tolerated. Within the first two weeks after surgery, the dermal lesions were prone to healing, but they became aggravated thereafter. The general condition of the patient deteriorated progressively, and he died of severe sepsis on the postoperative 23rd day.

## DISCUSSION

Pyloric atresia (PA) is a rare surgical emergency in newborns with obscure etiology, and its incidence has been reported as 1 per 100,000 live births.[6] Anatomic subgroups consist of (a) pyloric membrane alone, (b) obstructed pyloric canal by a solid cord and (c) atrophic pylorus with a gap between the stomach and duodenum.[7] PA is also associated with EB and/or aplasia cutis congenitalis, as seen in our patient.[8]

The coexistence of PA and EB was first described by Swinburne and Kohler in 1968.[1,9] Carmi suggested the pathophysiology of the disorder; therefore, it is also referred to as 'Carmi syndrome'.[2] Both PA and EB are very rare and autosomal recessive entities. The combination of these pathologies appears more than coincidental and is not the result of chance association.[10,11] Chang et al.[12] proposed that PA occurs secondary to an intrauterine complication of EB where sloughing pyloric mucosa leads to fibrosis and obstruction of the pyloric canal. Indeed, any epithelial-lined structure may be affected by EB, such as the uroepithelium.[13] EB is divided into three main categories: (a) EB simplex occurs within the epidermis, (b) junctional EB occurs within the lamina lucida of the basement membrane, and (c) dystrophic EB occurs within the lamina densa of the basement membrane.[14]

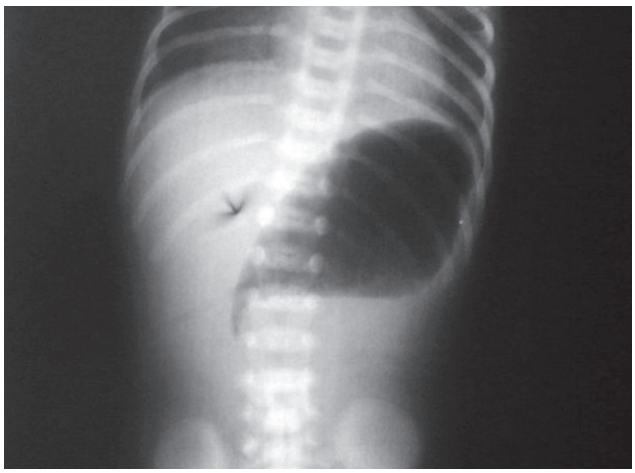

**Fig. 1.** Abdominal X-ray of Case 1 shows large gastric air-fluid with lack of air distally.

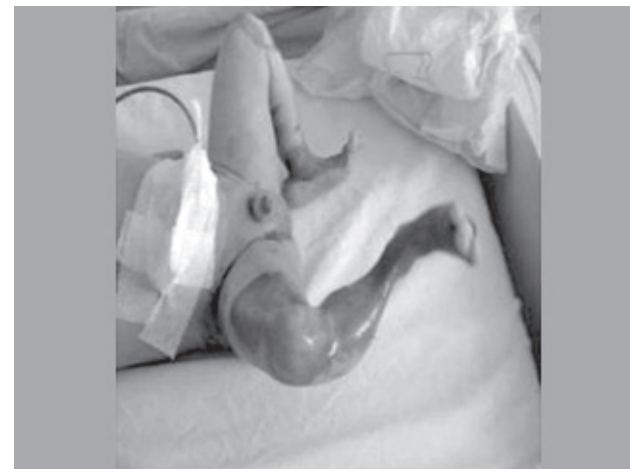

**Fig. 2.** Dermal lesion of Case 2 on the extremities.

Neonates with PA usually present with nonbilious vomiting soon after birth. Plain radiographs of the abdomen confirm the diagnosis of PA, with the stomach distended by air and an otherwise gasless abdomen.[7] Skin lesions may not appear until as late as 48 hours after birth. Disruption of the intestinal mucosa leads to malabsorption, increased antigenic sensitivity, bloody diarrhea, and protein losing enteropathy. Patients with PA/EB may have many associated disorders including gastrointestinal, urinary, pulmonary, and eye problems.[15]

The surgical treatment of PA depends on the anatomic variety. The recommended treatment for a pyloric web is excision of the web in combination with a pyloroplasty.[16] For solid PA, the treatment of choice is Heineke-Mikulicz pyloroplasty if the atresia is short. Excision of the atretic segment with gastroduodenostomy is the choice of treatment if the solid cord is long. Gastroduodenostomy is required in cases of PA with gap.[17] Dessanti et al.[5] described a new mode of surgical treatment. A longitudinal incision on the gap and dissection of two stumps of atresia are the essentials of their technique. We performed this technique in one patient with a good postoperative outcome. In the other patient, we performed a gastroduodenostomy. He also tolerated the oral feeding well.

Pyloric atresia (PA)/EB is a highly lethal combination, and death is almost a universal result. Hayashi et al.[10] reported four patients who had a survival between 17 months to 16 years, but they might have had alleles with lesser likelihood of lethality. Many infants die from extensive denudation of skin leading to septicemia, electrolyte imbalance, protein loss, and dehydration. Chronic blood and protein loss from denuded skin and mucosa causes anemia and hypoalbuminemia. Very few patients with limited and manageable skin blisters had long survival. As in our patient, most of the PA/EB infants die within the first few weeks or months of life despite successful surgical correction of PA.[3] Our cases developed aggravated skin lesions following minor trauma, and had severe sepsis induced by infected skin lesions of EB.

In conclusion, PA/EB combination is rare and a lethal pathology. Surgical intervention for PA is effective only for short-term survival, and the long-term outcome is almost always poor.

## REFERENCES


1. Birnbaum RY, Landau D, Elbedour K, Ofir R, Birk OS, Carmi R. Deletion of the first pair of fibronectin type III repeats of the integrin beta-4 gene is associated with epidermolysis bullosa, pyloric atresia and aplasia cutis congenita in the original Carmi syndrome patients. Am J Med Genet A 2008;146:1063-6.
2. Carmi R, Sofer S, Karplus M, Ben-Yakar Y, Mahler D, Zirkin H, et al. Aplasia cutis congenita in two sibs discordant for pyloric atresia. Am J Med Genet 1982;11:319-28.
3. Dank JP, Kim S, Parisi MA, Brown T, Smith LT, Waldhausen J, et al. Outcome after surgical repair of junctional epidermolysis bullosa-pyloric atresia syndrome: a report of 3 cases and review of the literature. Arch Dermatol 1999;135:1243-7.
4. Samad L, Siddiqui EF, Arain MA, Atif M, Parkash J, Ahmed S, et al. Pyloric atresia associated with epidermolysis bullosa--three cases presenting in three months. J Pediatr Surg 2004;39:1267-9.
5. Dessanti A, Iannuccelli M, Dore A, Meloni GB, Niolu P. Pyloric atresia: an attempt at anatomic pyloric sphincter reconstruction. J Pediatr Surg 2000;35:1372-4.
6. Muller M, Morger R, Engbert J. Pyloric atresia: A report of four cases and review of the literature. Pediatr Surg Int 1990;5:276-9.
7. Okoye BO, Parikh DH, Buick RG, Lander AD. Pyloric atresia: five new cases, a new association, and a review of the literature with guidelines. J Pediatr Surg 2000;35:1242-5.
8. Sencan A, Mir E, Karaca I, Günşar C, Sencan A, Topçu K. Pyloric atresia associated with multiple intestinal atresias and pylorocholedochal fistula. J Pediatr Surg 2002;37:1223-4.
9. Sarin YK, Nagdeve NG. Carmi syndrome complicated by pharyngo-esophageal perforation. Indian Pediatr 2006;43:61-4.
10. Hayashi AH, Galliani CA, Gillis DA. Congenital pyloric atresia and junctional epidermolysis bullosa: a report of long-term survival and a review of the literature. J Pediatr Surg 1991;26:1341-5.
11. Maman E, Maor E, Kachko L, Carmi R. Epidermolysis bullosa, pyloric atresia, aplasia cutis congenita: histopathological delineation of an autosomal recessive disease. Am J Med Genet 1998;78:127-33.
12. Chang CH, Perrin EV, Bove KE. Pyloric atresia associated with epidermolysis bullosa: special reference to pathogenesis. Pediatr Pathol 1983;1:449-57.
13. Reitelman C, Burbige KA, Mitchell ME, Hensle TW. The urological manifestations of epidermolysis bullosa. J Urol 1986;136:1320-2.
14. Shaw DW, Fine JD, Piacquadio DJ, Greenberg MJ, Wang-Rodriguez J, Eichenfield LF. Gastric outlet obstruction and epidermolysis bullosa. J Am Acad Dermatol 1997;36:304-10.
15. Lestringant GG, Akel SR, Qayed KI. The pyloric atresia-junctional epidermolysis bullosa syndrome. Report of a case and review of the literature. Arch Dermatol 1992;128:1083-6.
16. Moore CM. Congenital gastric outlet obstruction. J Pediatr Surg 1998;24:1241-6.
17. Al-Salem A, Nawaz A, Matta H, Jacobsz A. Congenital pyloric atresia: the spectrum. Int Surg 2002;87:147-51.