Case Report

Pyloric Atresia Associated with Epidermolysis Bullosa and Aplasia Cutis Congenita; Carmi Syndrome; Mimicking Esophageal Atresia

Canan KOCAOĞLU[1,a], Merve ATILGAN[1], Nuriye EMİROĞLU[2], Hüseyin ALTUNHAN[2]

[1]*Necmettin Erbakan Üniversitesi Tıp Fakültesi, Çocuk Cerrahi Anabilim Dalı, Konya, Türkiye*
[2]*Necmettin Erbakan Üniversitesi Tıp Fakültesi, Çocuk Sağlığı ve Hastalıkları Anabilim Dalı, Yenidoğan Bilim dalı, Konya, Türkiye*

ABSTRACT

Carmi syndrome, which involves epidermolysis bullosa and pyloric atresia, is a rare neonatal emergency surgical disease. We report Carmi syndrome in a neonate treated surgically. A one-day-old female neonate was consulted due to antenatal polyhydramnios, and no gas was observed on abdominal radiography. There were severe bullous skin lesions on the trunk and areas of aplasia cutis congenita on the neck and extremities. The orogastric tube could not be advanced into the stomach, and it was determined that the opaque contrast material did not pass into the stomach. The patient was operated on with a preliminary diagnosis of esophageal and pyloric atresia. During intraoperative exploration, pyloric atresia was detected. Gastroduodenostomy and gastrostomy were performed. On the 3rd postoperative day, while the proximal esophagus was aspirated, the orogastric tube reached the stomach. At first, it was thought that the reason why the orogastric tube did not reach the stomach was the narrowing of the his angle following excessive gastric dilatation. The patient was started on oral feeding on the 5th postoperative day. Postoperatively, the neonate began to deteriorate and eventually developed septicemia secondary to aplasia cutis and epidermolysis bullosa and passed away on the 24th postnatal day.

Keywords: *Aplasia Cutis Congenita, Carmi Syndrome, Congenital Pyloric Atresia, Epidermolysis Bullosa, Esophageal Atresia.*

ÖZ

Ösefagus Atrezisini Taklit Eden Pilor Atrezisi, Epidermolizis Bülloza ve Aplazia Kutis Konjenita Birlikteliği; Carmi Sendromu

Epidermolizis bülloza ve pilor atrezisi içeren Carmi sendromu nadir görülen bir yenidoğan acil cerrahi hastalığıdır. Şimdi cerrahi olarak tedavi edilen bir yenidoğanda Carmi sendromunu tanımlıyoruz. Bir günlük bir kız yenidoğan, antenatal polihidramnios nedeniyle konsülte edildi ve abdominal radyografide gaz yoktu. Gövdede şiddetli dermal büllöz lezyonlar ve boyun ve ekstremitelerde aplasia cutis congenita vardı. Orogastrik tüp mideye ilerletilemedi ve opak materyalin mideye geçmediği belirlendi. Hasta, özofageal atrezi ve pilor atrezisi ön tanısıyla ameliyata alındı. Perioperatif eksplorasyonda pilor atrezisi tespit edildi. Gastroduodenostomi ve gastrostomi yapıldı. Ameliyat sonrası 3. günde proksimal özofagus aspire edilirken orogastrik tüp mideye ulaştı. İlk başta orogastrik tüpün mideye ulaşamamasının nedeninin aşırı gastrik dilatasyon sonrası his açısının daralması olduğu düşünüldü. Hastaya ameliyat sonrası 5. günde beslenme başlandı. Ameliyat sonrası yenidoğan kötüleşmeye başladı ve sonunda aplasia cutis ve epidermolizis bülloza nedeniyle septisemi gelişti ve doğum sonrası 24. günde hayatını kaybetti.

Anahtar Sözcükler: *Aplasia Kutis Konjenita, Carmi Sendromu, Epidermolizis Bülloza, Konjenital Pilor Atrezisi, Ösefagus Atrezisi.*

Bu makale atıfta nasıl kullanılır: Kocaoğlu C, Atılgan M, Emiroğlu N, Altunhan H. Ösefagus Atrezisini Taklit Eden Pilor Atrezisi, Epidermolizis Bülloza ve Aplazia Kutis Konjenita Birlikteliği; Carmi Sendromu. Fırat Tıp Dergisi 2025; 30(4): 282-286.

How to cite this article: Kocaoglu C, Atilgan M, Emiroglu N, Altunhan H. Pyloric Atresia Associated with Epidermolysis Bullosa and Aplasia Cutis Congenita; Carmi Syndrome; Mimicking Esophageal Atresia. Firat Med J 2025; 30(4): 282-286.

ORCID IDs: C.K. 0000-0001-7111-3318, M.A. 0009-0000-3782-4079, N.E. 0000-0003-2444-4725, H.A. 0000-0003-0264-8671.

Carmi syndrome, which is characterized by the association of epidermolysis bullosa (EB) and pyloric atresia (PA), is a rare neonatal emergency surgical disease with an autosomal recessive inheritance and a well-known genetic etiology (1, 2). PA is a rare surgical emergency in neonates of unknown etiology, with an incidence of 1 in 100,000 live births. They constitute 1% of all intestinal obstructions. PA typically occurs in isolation, but associated anomalies are also commonly seen in 40% to 50% of the cases, and EB is the most common, while the incidence of EB is one in 300,000 (3, 4). Carmi syndrome carries a high mortality rate owing to its systemic manifestations, such as electrolyte imbalances and septicemia (5-7). We report a neonate born with Carmi Syndrome who had no gas in the abdomen and was treated surgically.

CASE REPORT

A one-day-old female (36+5 weeks gestation, birth weight of 2070 g) was born via cesarean section with spinal anesthesia; her mother was 31 years old, gravida 7, parity 2, abortion 4. She was admitted to the newborn intensive care unit due to extensive skin lesions and antenatal polyhydramnios, and there was no intraabdominal gas was seen on X-ray (Figure 1).

[a]Yazışma Adresi: Canan KOCAOĞLU, Necmettin Erbakan Üniversitesi Tıp Fakültesi, Çocuk Cerrahi Anabilim Dalı, Konya, Türkiye
Tel: 0533 239 4564 e-mail: drckocaoglu@hotmail.com
Geliş Tarihi/Received: 30.01.2025 Kabul Tarihi/Accepted: 29.05.2025
* This study was presented as a poster presentation at 41. Annual Congress of Pediatric Surgery Infectious Disease (10 November 2024, Nevşehir).

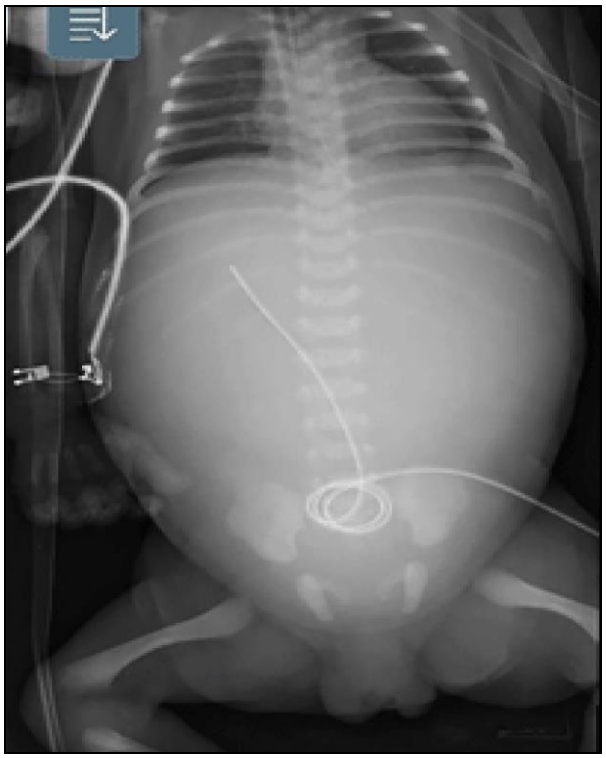
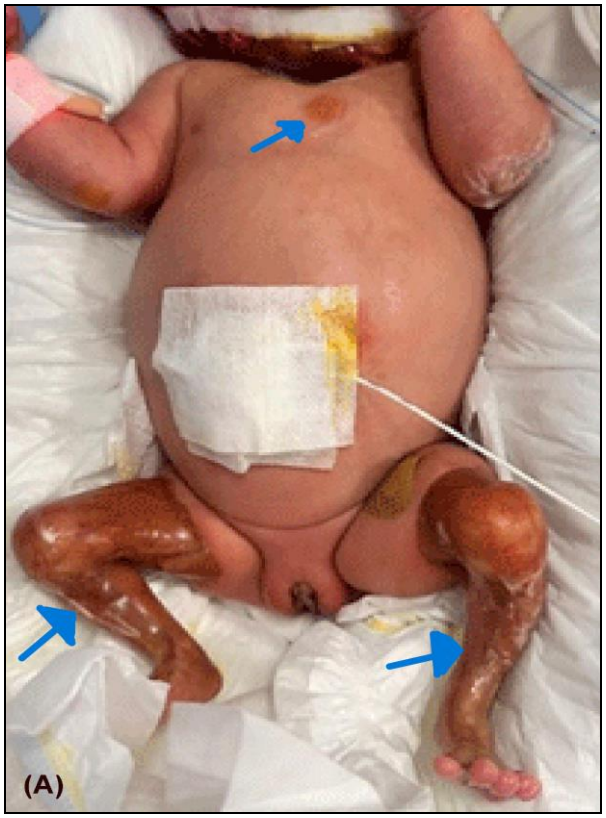


Figure 1. *Postnatal first day no gas on abdominal X-Ray examination.*

She had severe bullous skin lesions on the trunk because of epidermolysis bullosa and areas of aplasia cutis congenita on the neck and extremities (Figure 2A, B) and trisomy 18.



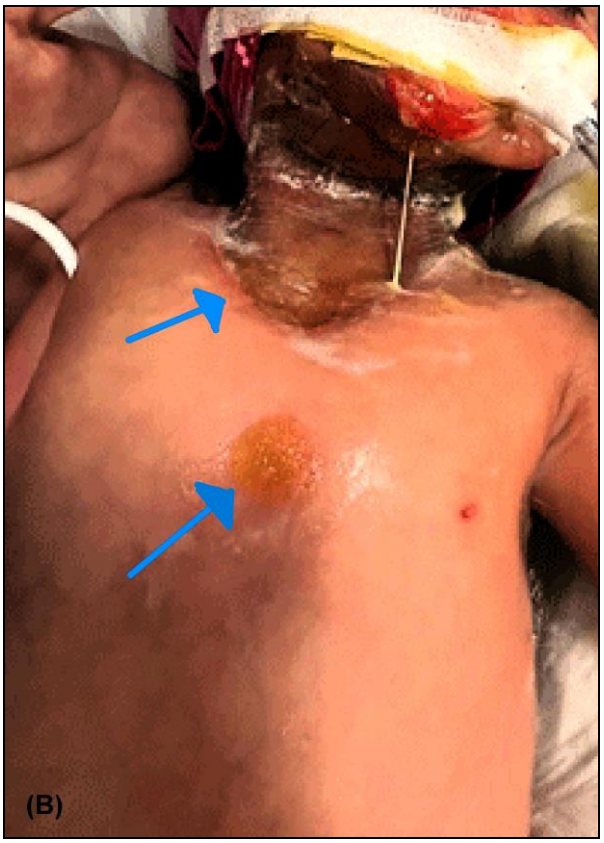


Figure 2A, B. *Aplasia cutis congenita ve epidermolizis bülloza.*

On laboratory tests, WBC 18.4 (4-10x103 uL), hemoglobin 17.4 (12.1-17.2 g/dL), sodium 137 (136-145 mmol/L), potassium 4.54 (3.5-5.1 mmol/L), chlorine 103 (98-107 mmol/L), and C- reactive protein <0.3 (0-5 mg/L) were normal. Abdominal ultrasonography revealed a 6x7 cm cystic lesion located intraperitoneally, distinctly identifying the left diaphragma superiorly in the midline of the abdomen, pushing the liver posteriorly. The identified cyst is primarily compatible with a mesenteric cyst. Its connection with the intestinal tract could not be demonstrated sonographically." The neonate was evaluated due to antenatal polyhydramnios, and no gas was present on abdominal X-ray. The orogastric tube could not be advanced into the stomach, and it was determined that the contrast material did not pass into the stomach (Figure 3).

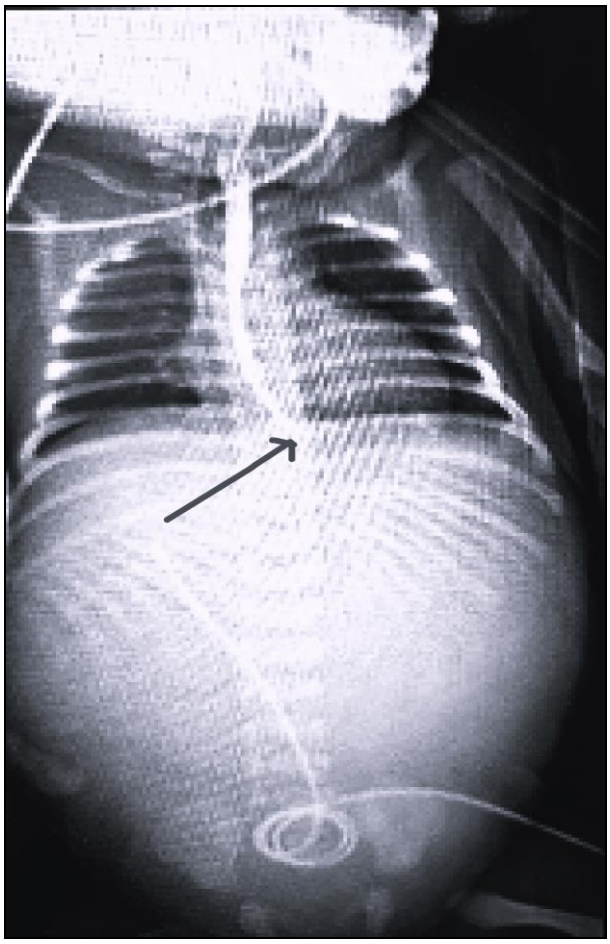
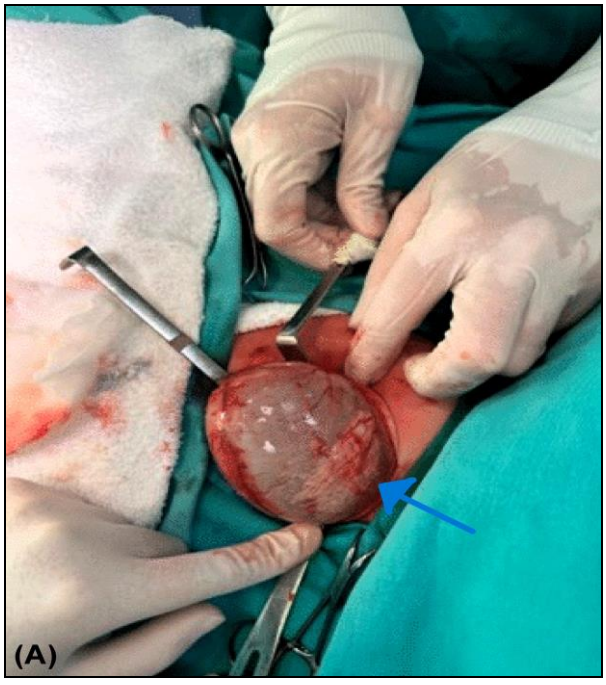


Figure 3. *Opaque material did not pass into the stomach.*

The patient was operated on with a preliminary diagnosis of esophageal and pyloric atresia. In the perioperative exploration, it was seen that the stomach was cystic and its wall was edematous (Figure 4A, B).



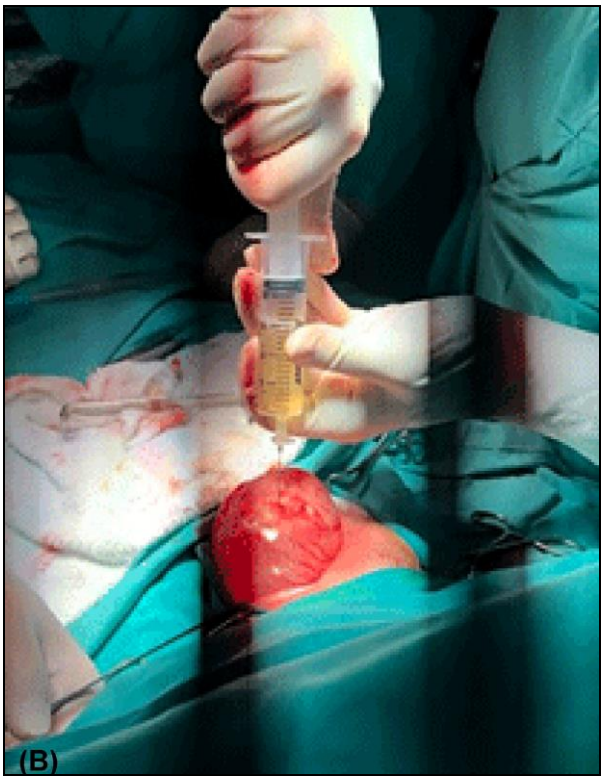


Figure 4. *Peroperative images (A) Cystic stomach, (B) Non-bilious serous fluid taken from the stomach.*

Type 2 pyloric atresia was identified. The stomach was opened near the pylorus, and it was ensured that there was no passage to the pylorus. Gastroduodenostomy and gastrostomy were performed (Figure 4C).

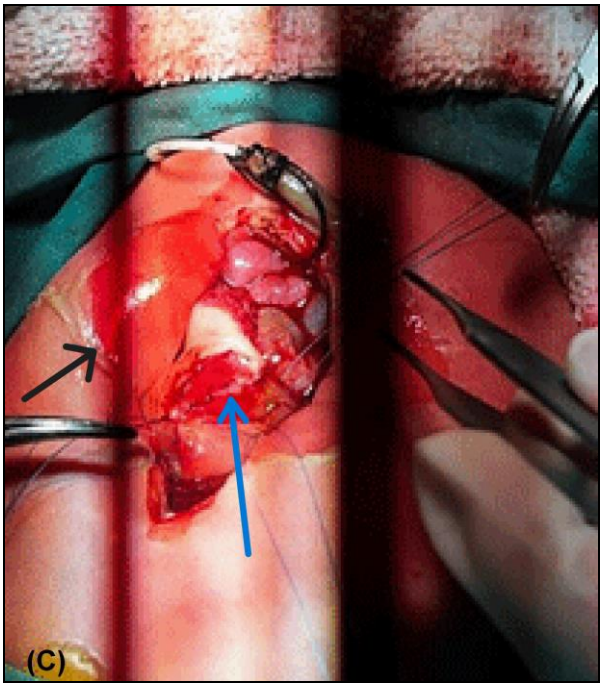


Figure 4C. *Gastroduodenostomy, gastrostomy, Epidermolysis bullosa.*

On the third postoperative day, while the proximal esophagus was aspirated, it was observed that the oro-

gastric tube reached the stomach. At first, it was thought that the reason for the orogastric not reaching the stomach was the disruption of the His angle after excessive gastric dilatation. The patient was started on oral feeding on the 5th postoperative day and passed stool thereafter (Figure 5).

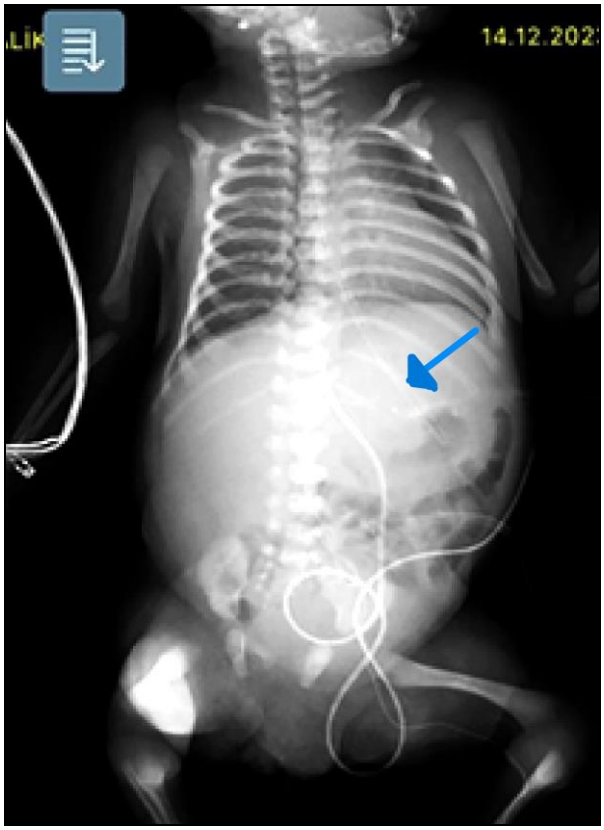


Figure 5. *Postoperative seventh days, Orogastric tube.*

On the 11th postoperative day, blood count was as follows: CRP 197.6 (0-5 mg/L), procalcitonin 1.26 (0-0.046 ug/L). Candida parapsilosis was isolated from the blood culture. On the postnatal 23rd day, WBC was 17.1 (4-10x103 uL), hemoglobin was 11.1 (12.1-17.2 g/dL), CRP was 86.4 (0-5 mg/L), and procalcitonin was 14.1 (0-0.046 ug/L). The neonate began to deteriorate and eventually developed septicemia secondary to aplasia cutis and epidermolysis bullosa and passed away on the 24th postnatal day.

DISCUSSION

PA can present with polyhydramnios or fetal gastric dilatation on antenatal ultrasonography (8). PA is usually suspected when neonates develop recurrent non-bilious vomiting and abdominal distension immediately after birth. On direct abdominal radiography, the stomach is dilated, and there is no other gas distal to the stomach. In our case, the angle of his was narrowed due to excessive gastric dilatation, and the abdomen was found to be free of gas because gas could not pass into the stomach (9). PA is anatomically divided into three subgroups: (a) only the pyloric membrane or web (57%), (b) the pyloric canal occluded by a solid cord (34%), and (c) an atrophic pylorus with a gap between the stomach and duodenum (9%). The pyloric membrane is treated with Heineke-Mikulicz pyloroplasty, while type b and c variations are usually treated by excision of the atretic segment and gastroduodenostomy (3, 10). In our case, there was a type B pyloric atresia, and we performed gastroduodenostomy. All patients with PA should be screened in terms of tracheoesophageal fistula, renal and ureteral anomalies, and EB (11).

EB is an inherited, autosomal recessive, bullous disease characterized by mucosal erosions and skin blisters. Numerous subtypes of EB are described and are divided into three major groups: EB simplex, dystrophic EB, and junctional EB. Junctional EB is further divided into three subgroups: Herlitz, non-Herlitz, and junctional EB with PA, also known as Carmi syndrome (12).

Carmi syndrome is an extremely rare autosomal recessive genetic disorder characterized by the coexistence of PA and junctional EB, and with aplasia cutis congenita is characterized by localized or widespread absence of skin in approximately 28% of patients (13). Our case was also diagnosed with aplasia cutis congenita. The prognosis of patients with Carmi syndrome is also poor, with mortality rates of up to 75%, mostly due to sepsis and renal failure, with a median time to death of 30 days (13).

Carmi syndrome should be excluded in every neonate with PA, regardless of the degree of skin blistering. Similarly, when a patient presents with clinical signs of EB, as in our case, even if no gas is seen in the abdomen, the possibility of Carmi syndrome should be considered in newborns. It should be noted that a gas-free abdominal x-ray and pyloric atresia may mimic esophageal atresia. Carmi Syndrome follows an autosomal recessive inheritance pattern. Families should be provided with genetic counseling before having subsequent children. Families can be informed about Carmi syndrome if findings such as polyhydramnios, gastric enlargement, and lower-limb anomalies are detected during prenatal diagnosis with ultrasonography.

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