CASE REPORT

A Case Report of Pyloric Atresia and Epidermolysis Bullosa: A Unique Co-Occurrence

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ABSTRACT

Pyloric atresia, a rare congenital anomaly, comprises 1% of intestinal atresia cases. It can occur alone or with conditions like epidermolysis bullosa and renal anomalies. Epidermolysis bullosa causes fragile skin and mucous membranes leading to blistering. The case describes a premature baby with low birth weight and epidermolysis bullosa, who developed stomach issues. Radiographs revealed gastric obstruction, prompting pyloric atresia type 1 diagnosis and Heineke-Mikulicz pyloroplasty. Sadly, sepsis caused the baby's death four days post-surgery. This highlights the rarity of combined conditions, the value of specialized care, and the challenges posed by complex congenital anomalies requiring multidisciplinary treatment.
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INTRODUCTION

Pyloric atresia is an exceedingly uncommon congenital anomaly, occurring in approximately 1 out of every 100,000 live births and constituting about 1% of all cases of intestinal atresia. The earliest documented case of pyloric atresia was reported by Calder in 1749, and the first successful surgical intervention was carried out by Touroff in 1940. The condition manifests as symptoms of obstruction at the outlet of the stomach, and it can either appear in isolation or be linked to other anomalies such as multiple intestinal atresia and epidermolysis bullosa (1). Pyloric atresia combined with epidermolysis bullosa involves both pyloric obstruction and blistering skin lesions (2). In this report, we present an unusual case of a nineteen-day-old female who was diagnosed with both pyloric atresia and epidermolysis bullosa. The patient underwent Heineke-Mikulicz pyloroplasty as part of her treatment.

CASE REPORT

A female infant was delivered via caesarean section after 33 weeks of gestation, weighing 1860 g. The mother's prenatal check-up revealed excess amniotic fluid (polyhydramnios). There was no family history of blistering diseases or congenital abnormalities. The baby's Apgar scores were 6 at 1 minute and 7 at 5 minutes following birth. Physical examination revealed the presence of blisters and peeled skin lesions on various parts of her body, including her cheeks, chin, thorax, abdomen, hands, left knee area, lower legs, and oral mucosa (Figure 1). These blisters contained clear fluid.

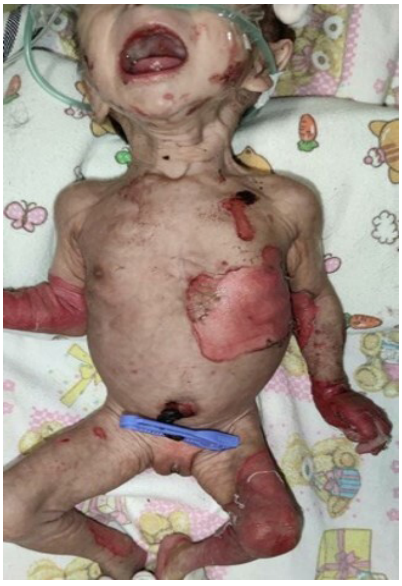

Figure 1: Clinical photograph showed skin peeling characteristic of epidermolysis bullosa

By the second day after birth, the infant exhibited feeding intolerance. The patient experienced repeated episodes of vomiting without the presence of bile and developed swelling in the upper abdomen (epigastric). The abdominal distention somewhat improved after the infant vomited. To address this, an orogastric tube was inserted to alleviate the pressure, and oral feeding was withheld. At five days of age, a babygram examination was conducted, revealing a swollen stomach with a solitary gas bubble in the gastric region, while no air was observed further down the digestive tract (Figure 2). Following this finding, the patient was admitted to our hospital.

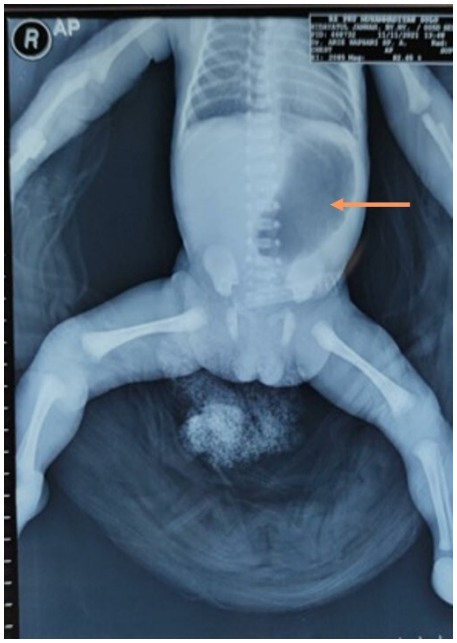

Figure 2: Babygram examination showed a single gastric air bubble, with no air in distal

Total Parenteral Nutrition (TPN) immediately for for the infants to meet their nutritional needs and prevent malnutrition while addressing the gastrointestinal obstruction. TPN provides a balanced mix of glucose, amino acids, lipids, electrolytes, vitamins, and trace elements tailored to the infant's specific requirements, and after stabilizing her general condition, laparotomy exploration was performed on day 19. A distended gaster with pyloric atresia was found and Heineke-Mikulicz pyloroplasty was performed (Figure 3).

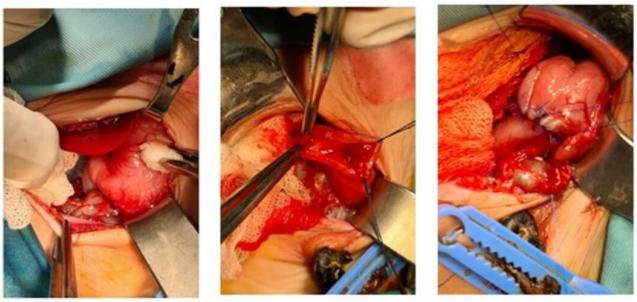

Figure 3: Intraoperative imaging showed distended gaster with pyloric atresia type 1 was found and Heineke-Mikulicz pyloroplasty was performed.

Skin care was managed through consistent cleansing, application of topical antibiotics, and dressing using antiseptic materials. Following the surgery, the infant experienced a critical condition of sepsis accompanied by dysfunction in multiple organs. Based on the results of blood culture, Vancomycin and Metronidazole were administered. Despite diligent efforts in infection management, assisted breathing, wound treatment, and nutritional assistance, the patient's life unfortunately came to an end on the twenty-fourth day after birth.

DISCUSSION

Pyloric atresia is a rare surgical emergency in new-born with incidence has been reported as 1 in 100.000 new-born. Anatomic subgroup of pyloric atresia is divided into pyloric membrane alone, obstruction pyloric canal by a solid cord, and atrophic pyloric with gap between gaster and duodenum (1). It also can be associated with other malformations such as epidermolysis bullosa as seen in our patient.

Pyloric atresia can occur secondary to an intrauterine complication of epidermolysis bullosa where sloughing pyloric mucosa leads to obstruction and fibrosis of pyloric canal (3). which is characterized by fragility of the skin and mucous membrane, then manifesting as spontaneous or post traumatic blister formation and skin peeling. The skin shows localized or extensive areas of blistering or peeling with little or no trauma (2).

A new-born with pyloric atresia may have gastric outlet obstruction symptoms which are recurrent nonbilious vomiting and abdominal distention. Plain radiographs of the abdomen may show single gastric gas bubble.1 Polyhydramnios usually found by third trimester in pregnancy (4).

Skin lesions can be seen at birth or as late as 2 days after birth. The surgical treatment of pyloric atresia depends on anatomic variety. The recommended surgical treatment for a pyloric web is excision of the web in combination with a pyloroplasty. In short type of solid pyloric atresia, the treatment is Heineke-Mikulicz pyloroplasty while in long type solid cord and pyloric atresia with gap, the choice of treatment is gastroduodenostomy (5). In our case we decided to undergo Heineke-Mikulicz pyloroplasty.

Skincare is principal treatment to protect the skin from poorly fitting clothing and using of atraumatic sterile dressings can prevent further skin peeling. Supportive measures including antibiotics and antiseptics to prevent wound infections. To improve outcome, the patient should have adequate nutritional support and be prevented form fluid and electrolyte imbalance condition. Genetic counselling is crucial for families with strong history of pyloric atresia and epidermolysis bullosa. Molecular genetic testing of the parents will

establish heterozygous carrier status. Assuming that the parents are obligate carriers, there is 1 in 4 chance at conception for each new-born of the affected parents of getting the disease (4).

Combination of pyloric atresia and epidermolysis bullosa is highly lethal and death is almost a universal result (2). The prognosis of this disease is bad due to prematurity, respiratory distress syndrome, malnutrition, and associated significant genitourinary disease (4). Neonates were dying from extensive defoliation of skin leading to dehydration, protein loss, electrolyte imbalance, and septicaemia. Very few patients had manageable skin blisters may have longer survival (2). Despite successful surgical correction of pyloric atresia, our patient was died on the 24th day of life. Our patient had severe sepsis induced by infected skin lesion of epidermolysis bullosa.

CONCLUSION

The combination of pyloric atresia and epidermolysis bullosa is exceptionally rare and often results in fatal outcomes. Although surgical treatment for pyloric atresia can temporarily improve survival, the long-term prognosis remains poor. Despite the rarity and poor outlook of this condition, early referral to specialized care could lead to better outcomes due to the complex nature of the disease. Effective management necessitates a team of specialists working together to optimize treatment strategies and potentially improve future patient outcomes.

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