CASE REPORT

PYLORIC ATRESIA ASSOCIATED WITH EPIDERMOLYSIS BULLOSA- A CASE REPORT

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Pyloric atresia is a rare congenital condition marked by obstruction of the gastric outflow because the pylorus is absent or severely narrowed. Blistering and fragility of the skin and mucous membranes are symptoms of the hereditary condition epidermolysis bullosa (EB). It is highly uncommon for pyloric atresia and epidermolysis bullosa to co-occur, and this presents substantial diagnostic and treatment difficulties. We describe a case of a newborn who was born with pyloric atresia and epidermolysis bullosa, focusing on the clinical presentation, the diagnostic procedure, and the surgical therapy. The complex interactions between these two dissimilar illnesses highlight the value of a multidisciplinary approach combining neonatologists, dermatologists, and paediatric surgeons for precise diagnosis and thorough care. By sharing this case report, we hope to add to the limited literature on this particular set of congenital defects and highlight the importance of increased clinical awareness and team-based treatment approaches when dealing with cases this complicated.

Keywords: Pyloric atresia; Epidermolysis bullosa; Carmi syndrome; Heineke-Mikulicz pyloroplasty

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INTRODUCTION

The inherited, variable group of uncommon genetic dermatoses known as epidermolysis bullosa (EB) is characterised by mucocutaneous fragility and blister development and is frequently brought on by minor trauma. Traditionally divided into EB simplex (EBS), junctional EB (JEB), Kindler EB, and dystrophic EB (DEB) according to the degree of basement membrane zone separation.[1] The narrowing of the pylorus and skin blistering are the predominant symptoms of Pyloric Atresia Associated with Junctional Epidermolysis Bullosa (PA-JEB), which is often severe and sometimes fatal in the neonatal era.[2] Only 91 cases of Epidermolysis Bullosa with Pyloric Atresia were reported up until 2012, the first case being reported in 1968.[3,4] Congenital localised absence of skin (aplasia cutis congenita) affecting the extremities and/or head, milia, nail dystrophy, scarring alopecia, hypotrichosis, and corneal abnormalities are additional characteristics shared by Epidermolysis Bullosa with Pyloric Atresia and the other primary forms of epidermolysis bullosa[5]. Surgery for pyloric atresia is only successful for short-term survival. The long-term prognosis of pyloric atresia associated with epidermolysis bullosa is nearly always poor, and the majority of patients die from epidermolysis bullosa related complications.[4] This paper's goal is to describe a severe case of pyloric atresia linked to epidermolysis bullosa.

CASE PRESENTATION

A three-day old baby girl was brought to the paediatric unit with multiple skin lesions (shown in figure-1) and difficulty with respiration for the last 24 hours. Antenatal history revealed that mother had polyhydramnios and gestational diabetes prenatally and emergency caesarean section at 37 weeks of pregnancy done due to foetal distress. Baby was doing well after delivery and was discharged to mother care. No skin lesions noted by parents or doctor at that time.

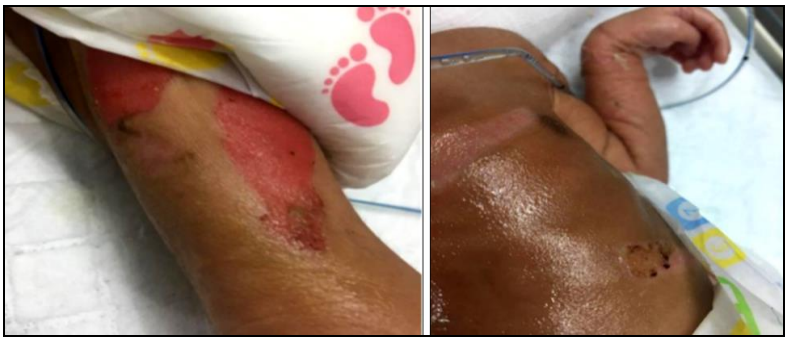

Figure-1: showing multiple skin lesions

On examination, the infant appeared sick, subcostal and suprasternal recessions were noted, bilateral creps with predominance to the right side noted. There were four blistering lesions on the lower limbs, one on the nose tip, and two on the scalp. It did not affect the oral mucosa. The dermis beneath was red, but there was no bleeding or discharge. The Nikolsky sign was positive with blistering in areas of friction. Lesions on the scalp and lower limbs were more prevalent, measuring 2-3 cm in diameter and blistering. The remainder of the exam was unremarkable. Her anthropometric data fell below the 10th percentile, with weight of 2.5 kg, FOC of 13.3 inches and length of 47 cm.

Initial management started with baby kept nil per oral, NG tube passed, oxygen support was given, intravenous maintenance fluids started and labs and chest x-ray ordered. Antibiotics started on injectable Cefotaxime and Amikacin as per local NICU initial protocol. Labs were grossly normal, but the chest x-ray revealed multiple bilateral infiltrates with marked right upper zone involvement. Baby also noted to have a large single gas bubble in the stomach area with no gas shadow distally. (Figure-2)

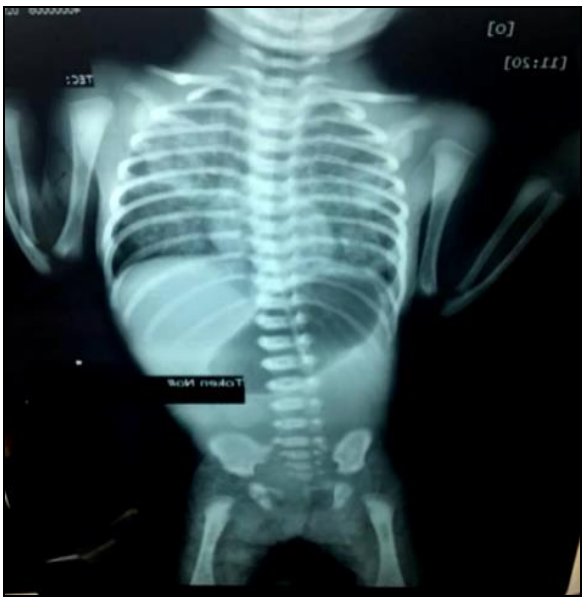

Figure-3: A large single gas bubble in epigastric region

Opinion from Paediatric Surgery and dermatology department sorted. Dermatology opinion was in favour of Epidermolysis Bullosa Junctional Variety and advised skin biopsy once child is stable, local application of fucidin cream and daily dressing. A paediatric surgeon's opinion was in favour of pyloric atresia. Antibiotic regime changed to IV Cefotaxime, Vancomycin and Metronidazole. Surgery for pyloric atresia planned.

Surgery done on day 10 of admission. During the procedure pyloric atresia-web type was noted with an underutilised small bowel; as a result, Web excision and pyloroplasty were performed on the baby. She was released from the hospital seven days after the surgery with an uneventful postoperative course. Parents were called for the follow up but there was no response from them. However, they came to the dermatology OPD post-surgery with complaints of multiple skin lesions. She was admitted in ICU with sepsis but unfortunately, she expired during her stay at the hospital. Skin biopsy was a part of the plan but it was never done.

DISCUSSION

Swinburne and Kohler provided the first description of Pyloric atresia and Epidermolysis Bullosa coexistence in 1968. Since Carmi proposed the pathophysiology of the condition, it is also known as "Carmi syndrome".[6] Histopathologic analyses and observation of recognisable clinical signs are used to make the diagnosis of Epidermolysis Bullosa. It is possible to use ultrasonography to diagnose pyloric atresia in utero. Starting at week fourteen of pregnancy, anatomical stomach traits can be investigated. The connection of polyhydramnios with stomach expansion is usually used to make the diagnosis. Our patient also experienced polyhydramnios during pregnancy. Additionally, patient with this condition also presents with clinical signs such as non-bilious vomiting, respiratory distress, dehydration, and abdominal distension.[7]

Patients with Epidermolysis Bullosa associated with Pyloric Atresia may also experience gastrointestinal, urinary, lung, ocular, and kidney difficulties (dysplastic/multicystic kidney, hydronephrosis/hydroureter, ureterocele, duplicated renal collecting system, missing bladder),[4–8] while our patient presented with pyloric atresia only. The junctional group is where Pyloric Atresia is most typically connected to the bulk of CEB types. Despite surgical treatment, this relationship is a diverse disease with significant fatality rates that approach 100%,[9] that, despite surgical repair, can cause mortality within a few months of birth.[7]

A literature search turned up just one case report in Pakistan describing the connection between Pyloric atresia and Epidermolysis Bullosa. (The Aga Khan University's Surgery Department)[10] A history of gestational diabetes is reported in many case reports, as well as in our patient, but its importance is unknown[11]. There have been a number of familial examples of pyloric atresia recorded, however our patient has no known substantial family history of the condition.[12] Epidermolysis Bullosa with pyloric atresia is an autosomal recessive trait and our patient

was a product of consanguineous marriage too. ITGA6, ITGB4, and PLEC gene mutations are among the potential causes of this. These genes give instructions on how to produce proteins that are essential for the skin and digestive system. About 80% of instances of this condition are caused by mutations in the ITGB4 gene, while only 5% of cases are caused by mutations in the ITGA6 gene.[13]

The management of EB with pyloric atresia lacks established treatment options. The therapies are primarily symptomatic and include conservative measures including the use of the proper dressing, infection prevention, and dietary supplements. Steroids used topically may be used to treat localised inflammation.[14] Treatment for pyloric atresia includes correcting dehydration and biological irregularities, followed by a pyloroplasty procedure that removes the diaphragm. The most widely utilised procedure is the Heineke-Mikulicz pyloroplasty.[15] The prognosis of this condition is poor despite surgical therapy for concurrent pyloric atresia due to nutritional disruption, absorption disruption, and in many cases, the advancement of sepsis. As a result, aggressive surgical treatment is frequently postponed in EB patients who also have pyloric atresia. According to a recent study, however, four out of every five patients who presented with stable vital signs tolerated treatment well following surgery.[14]

Without timely and appropriate treatment, pyloric atresia (PA) and EB are a fatal combination, often leading to death. Severe skin denudation can cause complications such as septicaemia, electrolyte imbalance, protein loss, and dehydration, which contribute significantly to poor outcomes. Even with treatment, the prognosis remains guarded due to the risk of complications, including urologic issues like ureterovesical blockage, underscoring the importance of routine follow-up.[4]

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