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Bart Syndrome Associated with Pyloric Atresia: An Uncommon Case Report

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Abstract

Background: Aplasia cutis congenital type VI (Bart's syndrome) has been known as an extremely rare genetic disorder in which there is localized absence of skin, epidermolysis bullosa and nail deformities.

Case report: Here, we present a rare case of Bart's syndrome in a female newborn diagnosed with congenital loss of skin over upper and lower limbs, trunk, neck and face as well as some bullae on them. Moreover, a dilated stomach was observed in radiographic examination. We treated the baby with TPN, systemic antibiotics, and also her wounds were covered by topical ointments. Laboratory tests, along with liver and renal function analyses were normal, and also serologic tests for infection were negative but she died at the age of 4 days.

Conclusion: The association between Bart's syndrome and pyloric atresia is a highly fatal combination and there is no treatment option to rescue the patients.

Key Words: Absence of skin, Bart's syndrome, Epidermolysis bullosa, Pyloric atresia.

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1- INTRODUCTION

Bart's syndrome (BS) is a rare dermatological disorder, first introduced by Bruce J. Bart in 1966 (1). BS is clinically characterized by Congenital Localized Absence of Skin (CLAS), formation of blisters (epidermolysis bullosa), and nail dystrophy (2).It is commonly inherited via an autosomal dominant pattern with complete penetrance (3, 4). The cutaneous symptoms of BS usually appear on the limbs as sharply demarcated, shining red lesions which stretch upward from the medial and/or dorsal portions of the foot to the shin (1).

In the first line of treatment, it is critical to prevent secondary infections and have a good prognosis but the treatment fundamentally depends on the healing of skin ulcerations (5). Epidermolysis bullosa is rarely associated with gastric outlet obstruction disorder, and it is still reported in a few cases (6).

2- CASE REPORT

A baby girl was born by cesarean section at 35 weeks' gestation to a 25-year-old gravida 3, para 2, abort 1 mother. Her birth weight, length and head circumference were 2kg, 44 cm and 33 cm, respectively. Apgar scores were 5 and 6 at 1 and 5 minutes.The baby was admitted to neonatal intensive care unit (NICU) due to her extensive dermal lesions. The mother denied any drugs or radiation exposure during her pregnancy. The parents were not relatives and there was no history of such conditions or skin diseases in their family. On detailed physical examination, we observed cutaneous aplasia on the upper and lower limbs, trunk, face, and neck with multiple vesicles and bullae on them, which were of varying shapes and sizes (**Fig 1**).

The initial chest/abdomen/pelvis radiograph revealed a dilated stomach. No air was seen in the bowel distally and there was no double-bubble; only the air-filled stomach was observed (Figure 2). From the first day, she was given TPN, systemic antibiotics, and also her wounds were covered with topical ointments. At time of admission, laboratory tests were conducted: the baby's CBC, electrolytes, liver and renal function tests were within normal limits, and serologic tests for infection were also negative. The patient was transferred to the service of pediatric surgery, but she died at the age of 4 days after the surgery.

3- DISCUSSION

Congenital pyloric atresia constitutes less than 1% of all upper gastrointestinal atresia, and its incidence is approximately 1: 100,000 newborns (7–12). CPA can be divided into three main types: 1- simple membrane or web (type A), 2- replacement by a solid cord (type B) which was observed in our patient, and 3- a gap with complete separation between the stomach and duodenum (type C). CPA has been known as an uncommon disorder with a probable autosomal recessive pattern and also in 50% of the diagnosed cases, it is connected with other syndromes (11, 12).

The autosomal recessive Epidermolysis Bullosa (EB) is commonly associated with Aplasia Cutis Congenita (ACC) (13). The skin defects in ACC are likely due to rapid growth of the brain which subsequently causes tension in the skin and underlying tissue; as a result skin lesions are induced at 10-15 weeks of prenatal period and also amniotic fluid disorders can be considered as a less relevant hypothesis (14, 15).

The distinctive manifestation of CPA-EB and/or ACC is recurrently observed in certain ethnic groups including middle-eastern Lebanese, Turks, and American Indians (11, 12). CPA rarely occurs with hereditary multiple intestinal atresia syndrome, a lethal autosomal recessive disorder which has been reported with high incidence in residents of northern Quebec (16).

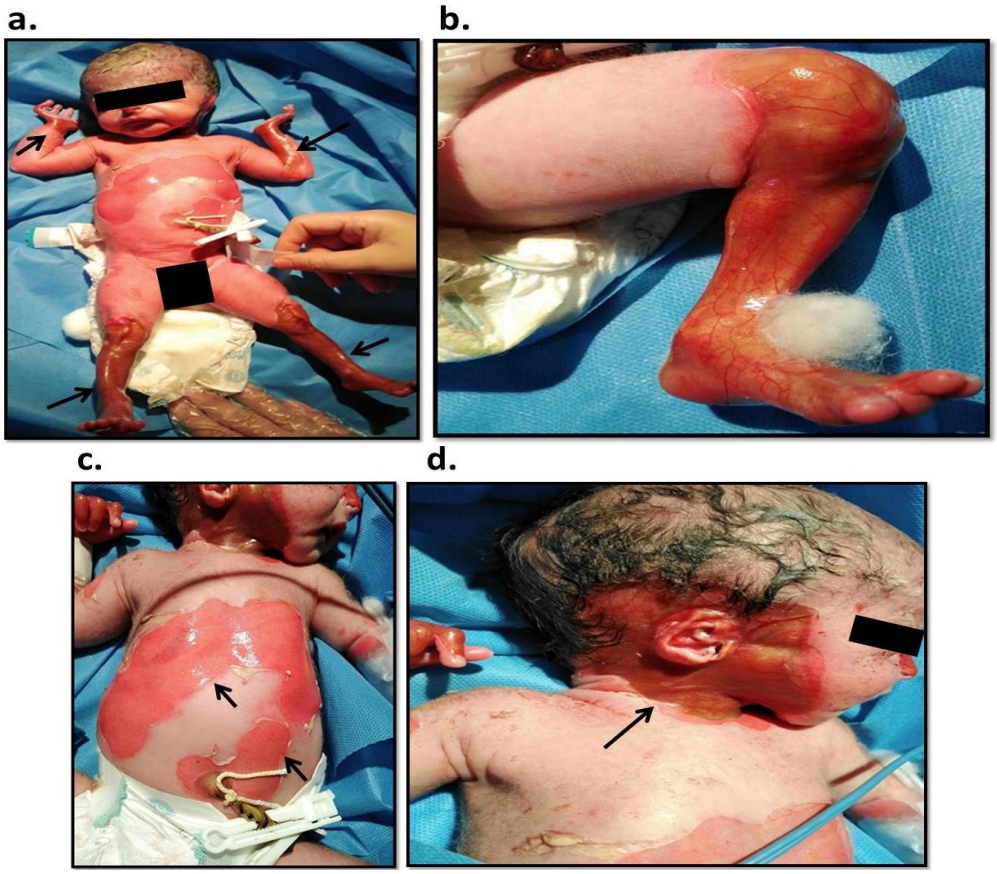

Fig. 1: Clinical presentations of Bart syndrome. a) Bilateral absence of skin on lower and upper extremities. b) Loss of skin over legs from knees to the toes. Lesions have demarcated limits covered by a translucent red membrane, and blood vessels under skin are clearly visible. c, d) Denuded areas on abdomen and neck. Lesions are marked with dark arrows.

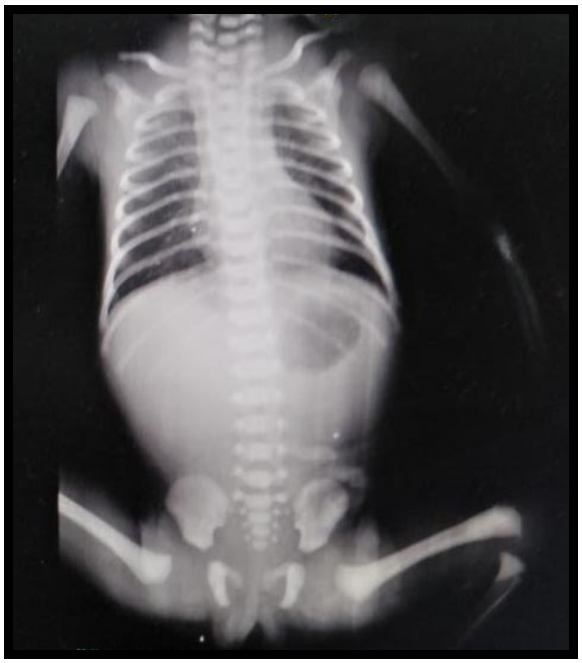

Fig. 2: The radiograph shows a dilated stomach in our patient. No air was observed in the bowel and there was no double-bubble; only the air-filled stomach was observed.

The association between CPA and EB was first defined by Swinburne and Kohler in 1968, and Carmi described the pathophysiology of this coexistence; thus, it is also called ‘Carmi syndrome’ (17–19). Both diseases are exceedingly rare and have an autosomal recessive pattern. The concomitant presence of CPA is due to an intrauterine complication of EB rather than a coincidental association that can lead to sloughing pyloric mucosa, fibrosis and also obstruction of the pyloric canal (20, 21). Indeed, any epithelial-covered surfaces, such as the uroepithelium, may be involved in EB (22).

EB is classified into the three subtypes: EB simplex, junctional EB, and dystrophic EB in which the lesions develop within the epidermis location, the lamina lucida, and lamina dense of the basement membrane, respectively (6).

Shortly after birth, non-bilious vomiting is one of common symptoms in infants with CPA and also radiographs indicate the stomach distension by air and otherwise a gasless abdomen (23). They may show no skin lesions until 48 hours after birth. Furthermore, disruption of the intestinal mucosa in CPA disorder can cause malabsorption, higher sensitivity to food antigens, hemorrhagic diarrhea, and loss of serum protein from intestinal tract (protein losing enteropathy). Patients with PA/EB may have different problems including gastrointestinal, urinary, pulmonary, and eye complications (24).

In addition to CPA, EB disorder is associated with other manifestations such as nail dystrophy, enamel hypoplasia, ACC or congenital localized absence of skin, eye symptoms, ear or nose hypoplasia or atrophy, complications of urinary and respiratory tract, oral feeding difficulty, and diarrhea (in many of the cases). However, the high mortality rate of this disease is due to the extensive loss of skin and as a result lack of barrier function, fluid and electrolyte imbalance, as well as sepsis (25).

Depending on the anatomic location, different surgical procedures are needed to treat CPA. The recommended approach for a pyloric web includes removal of the web in combination with a pyloroplasty (26). For solid type of CPA, the Heineke-pyloroplasty should be considered as a treatment of choice if the atresia size is short; however, in the cases of long atresia, excision of the atretic part by gastroduodenostomy is preferred. Gastroduodenostomy is recommended as a novel type of surgical interventions in CPA with gap (27). In this technique, it is necessary to perform a longitudinal cut on the gap and dissection the two stumps of atresia (28).

The association between EB and pyloric atresia is a highly fatal disorder and so death has become almost a universal outcome. Hayashi et al. reported the survival between 17 months to 16 years in four patients who might carry the alleles with less lethality. The majority of neonates die from extensive loss of skin, resulting in septicemia, electrolyte disturbance, protein leakage, and also dehydration. Furthermore, chronic blood and protein loss from the skin and mucosa barriers leads to anemia and hypoalbuminemia complications (29).

Diagnosis of Bart syndrome is based on the clinical presentation. In some cases, skin biopsy may be used to define the type of epidermolysis bullosa and genetic analysis can also be implemented to determine the gene mutations which may approve the final diagnosis.

In our case, Bart syndrome was presented with localized absence of skin over the lower and upper limbs, trunk, face and neck and also a dilated stomach. From the first day, she was given TPN, systemic antibiotics, and her wounds were also

covered by topical ointments, but after the surgery, she died at the age of 4 days.

There are no treatment options for EB with complications of pyloric atresia. The treatments commonly include conservative modalities with symptomatic effects such as suitable dressing, prophylactic control of infection and dietary supplementations. Topical steroids usually are used to reduce local inflammatory conditions. Also, genetic analysis may be useful in confirming the diagnosis of EB, while efforts on prenatal screening tests, genetic counseling and the treatment options are currently under investigation (19). Nevertheless, surgical procedures are applied to treat the associated pyloric or duodenal atresia. In general, EB has a poor prognosis due to malabsorption and development of sepsis infection in the majority of patients (30).

4- CONCLUSION

In neonates with congenital loss of skin and overlying blisters, Bart's syndrome must be checked and also other systems such as CNS should be evaluated for the congenital malformations. Prenatal genetic counseling and follow-up ultrasound to obtain evidence of pyloric atresia during pregnancy are also necessary to diagnose this uncommon genetic disorder.

5- REFERENCE

1. Rajpal A, Mishra R, Hajirnis K, Shah M. Bart's syndrome. Indian J Dermatol. 2008; 53:88–90.

2. Bart BJ, Garlin RJ, Anderson VE LF. Congenital localized absence of skin and associated abnormalities resembling epidermolysis bullosa: A new syndrome. Arch Dermatol. 1966; 93:296–304.

3. Smith SZ CD. A mechanobullous disease of the newborn : Bart's syndrome. Arch Dermatol. 1978; 114(1):81–4.

4. Sirota L, Dulitzky F MA. Bart's syndrome: a mechanobullous disease of the newborn. Report of five cases and review. Clin Pediatr (Phila). 1986; 25(5):252–4.

5. Quinteros DA, Ferreira LM, Schaffazick SR, Palma SD, Allemandi DA CL. Novel Polymeric Nanoparticles Intended for Ophthalmic Administration of Acetazolamide. J Pharm Sci. 2016; 105(10):3183–90.

6. Shaw DW, Fine JD, Piacquadio DJ, Greenberg MJ, Wang-Rodriguez J EL. Gastric outlet obstruction and epidermolysis bullosa. J Am Acad Dermatol. 1997; 36(2):304–10.

7. Dang N, Klingberg S, Rubin AI, Edwards M, Borelli S RJ Relic J, Marr P, Tran K, Turner A, Smith N, Murrell DF. Differential expression of pyloric atresia in junctional epidermolysis bullosa with ITGB4 mutations suggests that pyloric atresia is due to factors other than the mutations and not predictive of a poor outcome: three novel mutations and a review of the li. Acta Derm Venereo. 2008; 88:438–448.

8. Okoye BO, Parikh DH, Buick RG LA. Pyloric atresia: five new cases, a new association, and a review of the literature with guidelines. J Pediatr Surg. 2000; 35:1242–1245.

9. Puvabanditsin S, Garrow E, Samransamraujkit R, Lopez LA LW. Epidermolysis bullosa is associated with congenital localized absence of skin, fetal abdominal mass, and pyloric atresia. Pediatr Dermatol. 1997; 14:359–362.

10. Toma P, Mengozzi E, Dell'Acqua A, Mattioli G, Pieroni G FG. Pyloric atresia: report of two cases (one associated with epidermolysis bullosa and one associated with multiple intestinal atresias). Pediatr Radiol. 2002; 32:552–555.

11. AH A. Congenital pyloric atresia and associated anomalies. Pediatr Surg Int. 2007; 23:559–563.

12. Ilce Z, Erdogan E, Kara C, Celayir S, Sarimura N SO Ilce Z, Erdogan E, Kara C, Celayir S, Sarimurat N, Senyüz OF, Yeker D. Pyloric atresia: 15-year review from a single institution. J Pediatr Surg. 2003; 38: 1581–1584.

13. Maman E, Maor E, Kachko L CR. Epidermolysis bullosa, pyloric atresia, aplasia cutis congenita: histopathological delineation of an autosomal recessive condition. Am J Med Genet. 1998; 78:127–133.

14. Sandru, F. et al. A Case Report of Aplasia Cutis Congenita Type VI: Bart Syndrome. Mod Med. 2019; 16:213–7.

15. Blionas A, Giakoumettis D, Antoniades E, Drosos E, Mitsios A, Plakas S, Sfakianos G, Themistocleous SM. Aplasia cutis congenita: Two case reports and discussion of the literature. SurgNeurolInt. 2017; 8:273.

16. Conrad C, Freitas A, Clifton MS DM. Hereditary multiple intestinal atresias: 2 new cases and review of the literature. J Pediatr Surg. 2010; 45:21–4.

17. Birnbaum RY, Landau D, Elbedour K, Ofir R, Birk OS CR. 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.

18. Sarin YK NN. Carmi syndrome is complicated by pharyngo-esophageal perforation. Indian Pediatr. 2006; 43:61–4.

19. Carmi R, Sofer S, Karplus M, Ben-Yakar Y, Mahler D, Zirkin H Bar-Ziv J. Aplasia cutis congenita in two sibs discordant for pyloric atresia. Am J Med Genet. 1982; 11:319–28.

20. Hayashi AH, Galliani CA G 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.

21. Maman E, Maor E, Kachko L CR. Epidermolysis bullosa, pyloric atresia, aplasia cutis congenita: histopathological delineation of an autosomal recessive disease. Am J Med Genet. 1998; 78:127–33.

22. Reitelman C, Burbige KA, Mitchell ME HT. The urological manifestations of epidermolysis bullosa. J Urol. 1986; 136:1320–2.

23. Okoye BO, Parikh DH, Buick RG LA. Pyloric atresia: five new cases, a new association, and a review of the literature with guidelines. J Pediatr Surg. 2000; 35:1242–5.

24. Lestringant GG, Akel SR QK. The pyloric atresia-junctional epidermolysis bullosa syndrome:Report of a case and review of the literature. Arch Dermatol. 1992; 128:1083–6.

25. Dang N, Klingberg S, Rubin AI, Edwards M, Borelli S, Relic J Marr P, Tran K, Turner A, Smith N, Murrell DF. Differential expression of pyloric atresia in junctional epidermolysis bullosa with ITGB4 mutations suggests that pyloric atresia is due to factors other than the mutations and not predictive of a poor outcome: three novel mutations and a review of the li. Acta Derm Venereol. 2008; 88:438–448.

26. CM M. Congenital gastric outlet obstruction. J Pediatr Surg. 1998; 24:12416.

27. Al-Salem A, Nawaz A, Matta H JA. Congenital pyloric atresia: the spectrum. Int Surg. 2002; 87:147–51.

28. Dessanti A, Iannuccelli M, Dore A, Meloni GB NP. Pyloric atresia: an attempt at anatomic pyloric sphincter reconstruction. J Pediatr Surg. 2000; 35:1372–4.

29. Hünefeld C, Mezger M, Kern JS, Nyström A, Bruckner-Tuderman L, Müller

I Handgretinger R, Röcken M. One goal, different strategies--molecular and cellular approaches for the treatment of inherited skin fragility disorders. Exp Dermatol. 2013; 22(3):162–7.

30. Dank JP, Kim S, Parisi MA, Brown T, Smith LT, Waldhausen J Sybert VP. 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(10):1243.