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Bart's syndrome Associated with Pyloric Atresia: Case Report

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***Abstract*:-** In literature Bart's syndrome is presented as a genetic disorder of localized absence of skin accompanied with any type of epidermolysis bullosa (EB), but mostly with dominant dystrophic epidermolysis bullosa (DEB dominant), Bart's syndrome is one of the rare presentations of epidermolysis bullosa. It also known as congenital transient mechano-bullous dermatosis, and has tendency for skin and mucous membranes blistering, and may associated with nail deformities. Diagnosis is mainly clinically, but microscopy is required for classification. Palliative treatment and infection control are the main methods of treatment. The association of Bart's syndrome with other congenital anomalies is very rare.Here we reported a very rare interesting condition of a newborn girl baby of Bart's syndrome associated with pyloric Atresia, surgical repair done, together with palliative treatment and good infection control, she died 100 days after birth because of Fulminant sepsis due to Multi microbial infection. To the best of our knowledge, this is the first case of Bart's syndrome associated with Pyloric Atresia reported in Saudi Arabia.

Key words: - *Bart's syndrome, Epidermolysis Bullosa, Pyloric Atresia, Surgical repair, Multi organisms' infection,Fulminant sepsis.*

IINTRODUCTION

In 1966 Bart described three congenital skin manifestation disorders; congenital absence of skin, blistering and associated nail abnormalities.(1) the absence of skinlater known as epidermolysis bullosa (EB).(2)The mode of transmission is suggested to occurs through an autosomal dominant gene with complete penetrance but variable expression(3). (4),It is still one of the lesser presentations of epidermolysis bullosa(5). Today there is general consensus that the term Bart's syndrome should be used to identify patients with any type of epidermolysis bullosa who present with localized congenital absence of the skin on the extremities.(6), So Bart's syndrome is a subtype of dominantly inherited dystrophic epidermolysis bullosa.(7)It is believed that congenital skin absence follows the patterns of Blaschko's lines(8). In the first line treatment aimsto prevent secondary infection and prognosis is favorable but basically depends on the healing of skin lesions.(8).The association of Gastric outlet obstruction disorder with epidermolysis bullosa is rare but is not un common observation, still cases are individually reported.(9)

Here we are introducing an interesting newborn girl baby had congenital large skin free areas appears as red plaques on nose, ears, neck, abdomen, hands, arms, legs and feet, with congenital pyloric Atresia. The diagnosis was clinically Bart's syndrome. Light microscopy confirmed epidermal blistering. Further investigations to classify it was not done. The patient treated surgically for pyloric Atresia, and with other palliative systemic and local measures for skin careand infection control.

IICASE REPORT

Our baby was an outcome of emergency cesarean section delivery because of obstructed labour. This was due to fix breach presentation at Baljurashi General Hospital in KSA. Mother follow up was G3P2A0 and booked at 36 weeks of gestation.Baby was quite normal and cried immediately after birth without any assistance. The attending staffhas observed immediately the multiple localized rich reddish areas of skin defects in both arms, hands, legs,neck, abdomen, chest,nose and ears Fig (1 and 2). Baby initially

was stable and maintaining saturation at room air, skin was peeled and blisters appeared in the next day. We started septic precautions with highly wound sensitive antibiotics eg.fucidinic acid skin ointment, to avoid infections.Baby does not passed stool and later on developed persistent yellowish vomiting with abdominal distension. Imaging investigations revealed Pyloric Atresia, baby referred to the pediatric surgeon andunderwent a successful repair surgery, gradually the intubation removed and baby built up oral feeding and passed stool normally. Sadly, later on during course of admission, she developed recurrent attacks of sepsis spite of aggressive systemic and topical treatment, first sepsis was with Candida, then with enterococcus, and last one was fulminant multi microbial sepsis, which was the cause of death. Our patient, died after 100 days of delivery.

Figure 1: picture of the skin defects

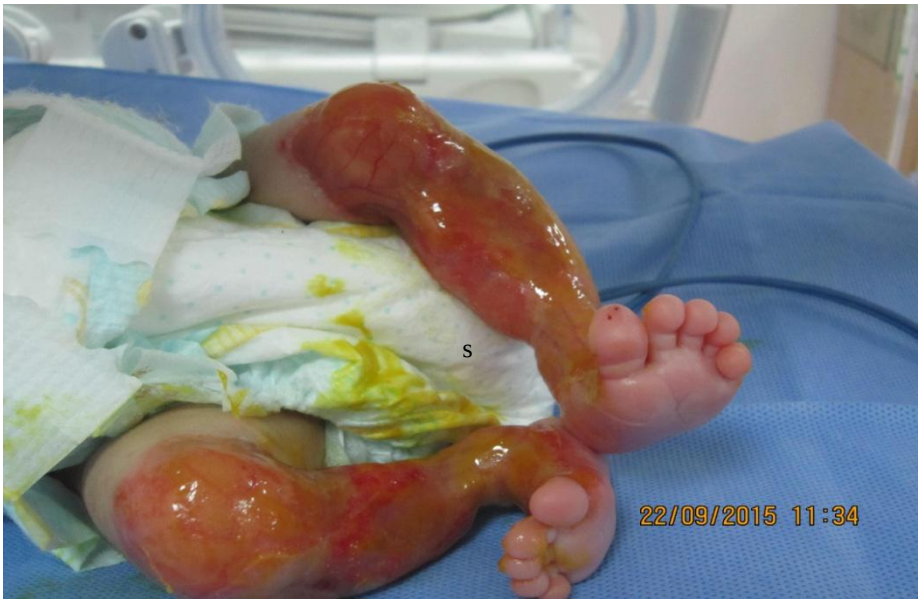

Figure 2: picture of the skin defects of the lower limp

III DISCUSSION

In theliterature, Bart's syndrome is arare congenital disorder,about half a million globally. Bart'sSyndrome occurs just one in a million. On the other hand, The coexistence of pyloric atresia and junctional epidermolysis bullosa (PA-JEB syndrome) has been repeatedlyobserved (10). Some still think it is arare association(11). Again, our case is the first one in KSA and is a valuable addition to this observation. This syndrome can be differentiated clinically from Herlitz junctional epidermolysis bullosa (JEB) by absence

of prominent granulation tissue formation and increased frequencies of genitourinary tract involvement and ear anomalies (10). Other associated congenital anomalies reported with Bart's syndrome is mandibulofacial dysostosis (12). Others reported Gastric outlet obstruction and gastric infarct in junctional epidermolysis bullosa(13) and others with pyloric and concomitant choanal atresia(14) Corbus callosum agenesis is also reported.

Someassociated disorders are more complicated, like a case report of double gastric outlet obstruction, rectovaginal fistula with epidermolysis bullosa. She recovered after excision of diaphragms, pyloroplasty and pelvic colostomy. (15)The variety of congenital disorders, and wide range of clinical presentation raising the question , is Bart's syndrome a specific syndrome?(16).Skin care and infection control were essential parts in plan ofmanagement, but despite of all efforts baby developed frequent attacks of different microbial infections, last one was post operative multi microbialsepsis. The skin care in patients of Bart's syndrome is essential and treatment should be directed towards prevention of sepsis and secondary infection(8)

REFERENCES

[1] Bart BJ, Gorlin RJ, Anderson VE. Congenital localized absence of skin and associated abnormalitiesresembling epidermolysis bullosa: A new syndrome. ArchDermatol. 1966;93:296-303.Amichai B, Metzker A. Bart's syndrome. International journal of dermatology. 1994 Mar;33(3):161- PubMed PMID: 8169011. Epub 1994/03/01. eng.

[2] Butler DF, Berger TG, James WD, Smith TL, Stanely JR, Rodman OG. Bart's syndrome: microscopic, ultrastructural, and immunofluorescent mapping features. Pediatric dermatology. 1986 Feb;3(2):113-8. PubMed PMID: 3513144. Epub 1986/02/01. eng.

[3] Smith SZ, Cram DL. A mechanobullous disease of the newborn. Bart's syndrome. Archives of dermatology. 1978 Jan;114(1):81-4. PubMed PMID: 339847. Epub 1978/01/01. eng.

[4] Sirota L, Dulitzky F, Metzker A. Bart's syndrome: a mechanobullous disease of the newborn. Report of five cases and review. Clinical pediatrics. 1986 May;25(5):252-4. PubMed PMID: 3698444. Epub 1986/05/01. eng.

[5] Kanzler MH, Smoller B, Woodley DT. Congenital localized absence of the skin as a manifestation of epidermolysis bullosa. Archives of dermatology. 1992 Aug;128(8):1087-90. PubMed PMID: 1497364.

[6] Zelickson B, Matsumura K, Kist D, Epstein EH, Jr., Bart BJ. Bart's syndrome. Ultrastructure and genetic linkage. Archives of dermatology. 1995 Jun;131(6):663-8. PubMed PMID: 7778916. Epub 1995/06/01. eng.

[7] Almeida NA, Serafini F, Marchiori J, Moro JG. [Do you know this syndrome? Bart's syndrome]. Anais brasileiros de dermatologia. 2010 Jan-Feb;85(1):119-21. PubMed PMID: 20464103. Epub 2010/05/14. Voce conhece esta sindrome? Sindrome de Bart. por.

[8] Quinteros DA, Ferreira LM, Schaffazick SR, Palma SD, Allemandi DA, Cruz L. Novel Polymeric Nanoparticles Intended for Ophthalmic Administration of Acetazolamide. J Pharm Sci. 2016 Aug 9. PubMed PMID: 27519647. Epub 2016/08/16. Eng.

[9] Shaw DW, Fine JD, Piacquadio DJ, Greenberg MJ, Wang-Rodriguez J, Eichenfield LF. Gastric outlet obstruction and epidermolysis bullosa. Journal of the American Academy of Dermatology. 1997 Feb;36(2 Pt 2):304-10. PubMed PMID: 9039206.

[10] Akar M, Erdeve O, Gokmen T, Oguz SS, Dilmen U, Sirvan L. Visceral bullae: A new finding in Bart's syndrome. Fetal and pediatric pathology. 2010 Jan;29(2):63-8. PubMed PMID: 20334480. Epub 2010/03/26. eng.

[11] Martin RW, 3rd, Myers T, Shehata BM, Zemtsov A, Olson JL. Congenital absence of skin and blistering in a neonate. Bart's syndrome and mandibulofacial dysostosis. Archives of dermatology. 1995 Oct;131(10):1197, 200. PubMed PMID: 7574841. Epub 1995/10/01. eng.

[12] Zirn JR, Scott RA, Aronian JM, 3rd, Lin AN. Gastric outlet obstruction and gastric infarct in junctional epidermolysis bullosa. Pediatric dermatology. 1995 Jun;12(2):174-7. PubMed PMID: 7659647.

[13] Narter F, Buyukbabani N, Yararli H, Ozturk S, Erguven M. Bart's syndrome associated with pyloric and choanal atresia. The Turkish journal of pediatrics. 2013 Mar-Apr;55(2):214-7. PubMed PMID: 24192685. Epub 2013/11/07. eng.

[14] Rawat JD, Sinha SK. Double gastric outlet obstruction with epidermolysis bullosa in a neonate. Indian journal of gastroenterology : official journal of the Indian Society of Gastroenterology. 2006 May-Jun;25(3):157. PubMed PMID: 16877835.

[15] Amichai B, Metzker A. Is Bart's syndrome a specific syndrome? Archives of dermatology. 1993 Jul;129(7):907-8. PubMed PMID: 8323317. Epub 1993/07/01. eng.