EUROPEAN ACADEMIC RESEARCH
Vol. VI, Issue 5/ August 2018

Impact Factor: 3.4546 (UIF)
DRJI Value: 5.9 (B+)

ISSN 2286-4822
www.euacademic.org

Case Report

Junctional epidermolysis bullosa (JEB)

YUNIS HAMODA
ELSHARIF A. BAZIE
AMAL GUBRA
AHMED YAYA ALZHRANI
King Fahad Hospital-NICU-ALBAHA

Abstract:

Second twin delivered by emergency caesarian section , developed blisters in the right leg after delivery and after NICU(Neonatal Intensive Care Unit) admission developed features of intestinal obstruction.

Hereditary epidermolysis bullosa (EB) constitutes three main types which were identified according to electron microscopy study as: simple, junctional, and dystrophic type.

Key words: Epidermolysis Bullosa, King Fahad Hospital, Al-Baha.

Second twin to gravida six, para five mother delivered by emergency caesarian section as she present in labor pain with history of previous caesarian section. Mother on regular follow up during pregnancy and normal antenatal follow-up and scan (Ultrasound).

Mother 38 year's old, house wife had no chronic illness, father 45 years old, they had four siblings and one boy who died at 28 days of age after admission to NICU and diagnosed as Epidermolysis Bullosa. No family history of skin diseases.

Baby delivered active, vigorous, APGOR score 8, 9 at 1, 5 minutes respectively. Birth weight 1720gram. No dysmorphic

features, normal cardiovascular, respiratory, neurological and abdominal examination.

Shortly after delivery baby develop blisters in the anterior aspect of right leg so shifted to NICU immediately after that new lesions developed in the right and left legs, right upper limb , mouth and abdomen ,figure[1&2] showed blisters distribution. Baby did not pass meconium but he pass urine.

Investigations shows:

Normal complete blood count and renal function

Normal abdominal and head ultrasound

Normal echocardiography (ECHO)

Radiological investigations: figure [3] X-ray showed dilated stomach with no air in lower part of abdomen. Figure [4] shows barium swallow with esophageal stricture and pyloric atresia.

Figure [1] Blisters in the right leg.

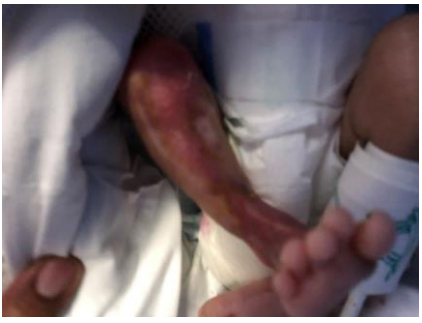


Figure [2]: Blisters in the foot:

Figure [3] Chest and Abdomen X-Ray;

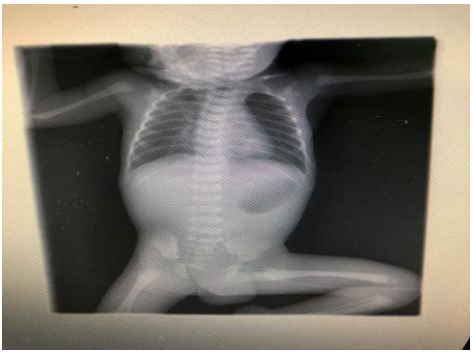
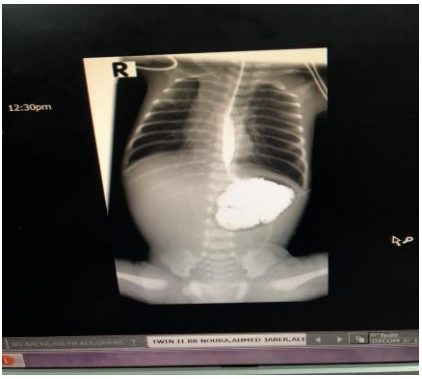


Figure 4: Barium swallow.



DISCUSSION:

To our knowledge this is the first reported case from king Fahad Hospital –Albaha.

Hereditary epidermolysis bullosa (EB) constitutes a genodermatosis group with variable clinical severity. Three main types were identified according to electron microscopy study as follows: simple, junctional, and dystrophic type. [1]
Epidermolysis bullosa simplex (EBS), accounting for 75 to 85 percent of all cases of EB in western countries [2]. In most of cases, EBS is caused by mutations in the keratin genes, resulting in the formation of a cleavage plane at the level of the basal keratinocytes.[2]

Junctional epidermolysis bullosa (JEB) comprises a group of autosomal recessive disorders characterized by blistering of the skin and mucosae that heal with scarring. [1]

Dystrophic epidermolysis bullosa (DEB) is characterized by blistering of the skin and mucosal membranes that heal with scarring. DEB is caused by mutations in the *COL7A1* gene encoding the alpha-1 chain of type VII collagen.[3]

In patients with JEB or recessive DEB, any portion of the gastrointestinal tract may be injured [4]. Gastrointestinal manifestations include esophageal strictures, gastroesophageal reflux, rectal tears, anal fissures and stenosis, and constipation. Esophageal strictures, resulting from recurrent mucosal blistering and scarring, are the most frequent and often the most disabling complication. The majority of strictures occurs in the upper third of the esophagus, but they may arise anywhere, leading to progressive dysphagia initially with hard or bulky foods, then with softer foods, and eventually with liquids.[4]

In Saudi Arabia, only few studies were done concentrating more on clinical features [5]. In Abahussein AA et al study[5]., sixteen patients were found to have EB. Ten (62.5%) had recessive dystrophic EB, four (25%) had epidermolysis bullosa simplex (EBS), one had dominant dystrophic epidermolysis bullosa (DDEB), and one had recurrent bullous eruptions of the hands and feet (Weber-Cockayne) , also in Alhumidi [6] study, 50% of the patients were of junctional type EB, 28% were epidermolytic type while 22% had dystrophic EB.

Acknowledge

We would like to thanks all our NICU staff, pediatric surgery team and our college and nursing staff for their help.

REFERENCES :

1-Fine JD, Eady RA, Bauer EA, et al. The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB. J Am Acad Dermatol 2008; 58:931.

2-Abu Sa'd J, Indelman M, Pfendner E, et al. Molecular epidemiology of hereditary epidermolysis bullosa in a Middle Eastern population. J Invest Dermatol 2006; 126:777.

3-Uitto J, Richard G. Progress in epidermolysis bullosa: genetic classification and clinical implications. Am J Med Genet C Semin Med Genet 2004; 131C:61.

4-Fine JD, Mellerio JE. Extracutaneous manifestations and complications of inherited epidermolysis bullosa: part I. Epithelial associated tissues. J Am Acad Dermatol 2009; 61:367.

5- Abahussein AA, al-Zayir AA, Mostafa WZ, Okoro AN. Epidermolysis bullosa in the eastern province of Saudi Arabia. Int J Dermatol. 1993;32(8):579-81

6- Alhumidi; BJMMR, 8(3): 238-244, 2015; Article no.BJMMR.2015.44