SKINmages

Epidermolysis Bullosa with Pyloric Atresia in a Premature Infant

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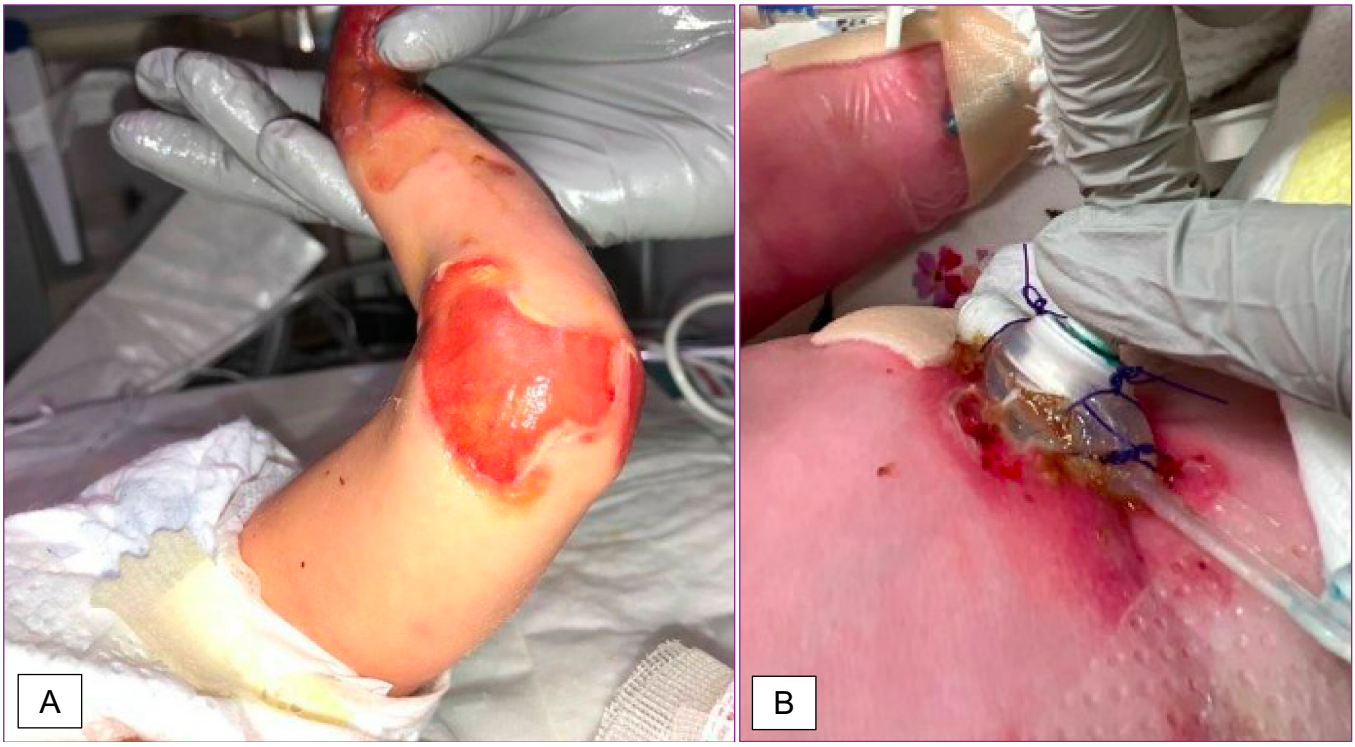

Figure 1. (A) Skin examination on arrival to tertiary care center revealed skin fragility with visible veins on foot and flaccid bullae present on medial knee and thigh (B) Delayed wound healing and purulence evident at site of gastrostomy tube placement

CASE REPORT

A 9-hour-old female infant noted to have skin fragility was transported from a community hospital to a tertiary care center for respiratory distress and tachycardia suspicious for sepsis. The patient's gestational course was complicated by maternal diabetes mellitus, polyhydramnios, and malpresentation necessitating a cesarean section at 35 weeks' gestation. Family history is limited to psoriasis in the patient's father. Initial skin examination revealed widespread abnormalities (**Figure 1A**) concerning for aplasia cutis congenita,

and wound care was initiated. On day 25 of life, genetic testing resulted with novel mutations in both PLEC gene alleles, suggestive of autosomal recessive (AR) Epidermolysis Bullosa with Pyloric Atresia (EB-PA). Initial renal ultrasound (RUS) was unrevealing. Repeat RUS revealed grade 3 hydronephrosis and calculi. Ophthalmic evaluation revealed lesions limited to the lateral eyelid, without corneal involvement. She was also diagnosed with pyloric atresia, prompting pyloric and proximal duodenal resection with gastroduodenostomy. The patient experienced apneic spells secondary to increased respiratory secretions and developed protein-losing enteropathy, prompting tracheostomy and gastrostomy tube placement (**Figure 1B**). She remained inpatient due to continued difficulties with maintaining skin integrity and avoiding infection, respiratory distress, hypothermia, and malnutrition. Currently, she is in stable condition receiving close follow-up outpatient, although she has been hospitalized five times since her initial discharge secondary to infections, primarily of the urinary tract.

DISCUSSION

A rare dermatologic condition occurring in 1 out of every 53,000 live births in the US, Epidermolysis Bullosa (EB) is attributable to mutations in structural proteins of the skin, resulting in fragility and blistering often noted at birth.[1] EB-PA is associated with AR mutations in α6β4 integrin or plectin, a regulatory protein most abundant in the skin and colon that regulates signaling between integrins and laminins in the basal membrane.[2] Most commonly, EB-PA impacts the ocular, gastrointestinal, and genitourinary tracts, although any plectin-containing tissue can develop pathology.[3] Independently, extracutaneous manifestations can be life-threatening, but in the setting of a weakened immune system and compromised skin integrity, morbidity is heightened.

The classic bullae of EB allow for a prompt clinical diagnosis confirmed by immunofluorescence antigen mapping, transmission electron microscopy, or next generation sequencing genetic panel. Conversely, diagnosis of EB in the neonatal period is more challenging due to confounders: congenital diseases, infections, or birth trauma.[5] Notably, EB-PA is mostly diagnosed in premature infants, but genetic panels for confirmation may take weeks to result, so clinical recognition is essential.[5]

Treatment of all EB subtypes is largely symptomatic, begins with wound care, and focuses on maintaining patient hemodynamic stability and comfort. Although promising, the field of gene therapy remains limited to specific mutations, thus not available for EB-PA. Because of the varied presentation of EB, multidisciplinary teams formulate treatment plans, often performing intense risk-benefit analyses with a low threshold for intervention to improve comfort and survival. As evidenced by our patient, intensive care does not preclude serious challenges, but multi-disciplinary teams improve outcomes.

In conclusion, EB-PA is a rare congenital blistering skin condition that can cause life-threatening complications in the neonatal period. Prompt diagnosis, wound care, surveillance for potential systemic manifestations, and multidisciplinary healthcare teams are essential for infection control, patient comfort, and long-term survival.

Conflict of Interest Disclosures: None

Funding: None

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