Epidermolysis Bullosa-Pyloric Atresia with Obstructive Uropathy in a Newborn: A Case Report

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Abstract

Epidermolysis bullosa (EB) is a group of rare inherited medical conditions resulting in easy blistering of the epithelial tissues in response to mechanical trauma. EB is occasionally associated with systemic presentations.

A newborn was referred to Shahid Rahimi Hospital in Khorramabad, Iran, on 11/2/2021 due to skin lesions and poor feeding. The patient was later diagnosed with EB and coexisting pyloric atresia and underwent pyloric resection and gastroduodenostomy. Due to reported hydronephrosis on the ultrasound, the diagnosis of epidermolysis bullosa-pyloric atresia-obstructive uropathy (EB-PA-OU), a multisystem variant of EB, was made.

High phenotypic variability has been found among EB cases. One of the variants is EB-PA, which is an autosomal recessive genodermatosis. In some cases, ureteral and renal abnormalities coexist with EB-PA, leading to a more complex variant named EB-PA-OU.

Given the known association between EB and multiorgan abnormalities, a systemic evaluation is required to detect these disorders and prevent them from progressing to irreversible stages.

Keywords

- Epidermolysis bullosa
- Pyloric atresia
- Gastroduodenostomy
- Renal abnormality

Introduction

Epidermolysis Bullosa (EB) is a rare group of inherited connective tissue disorders characterized by mucocutaneous fragility and blister formation following minor mechanical trauma.[1] Multiple genes have been identified to be responsible for various EB subtypes, resulting in distinct clinical phenotypes and severities.[2-3] The three primary types of EB classified by the level of skin cleavage are EB simplex (EBS), junctional EB (JEB), and dystrophic EB (DEB).[4-5] Some forms of EB are associated with extracutaneous manifestations leading to significant morbidity and mortality. One such rare variant is EB with pyloric atresia (EB-PA), which presents with characteristic skin fragility alongside congenital pyloric obstruction.[1] This case report aims to describe the diagnosis and management of

EB-PA complicated by hydronephrosis in a neonate.

Case presentation

A 5-day-old female newborn was admitted to Shahid Rahimi Hospital in Khorramabad, Iran, on 11/2/2021. She was born to a mother of G4P4Ab1 at 36 weeks of pregnancy with an Apgar score of 9 out of 10. Progressive erythematous skin lesions were observed in the perinasal, periorbital, and periauricular areas and on the extremities. Also, she experienced drooling after attempts at breastfeeding but had no signs of respiratory distress. The mother reported no history of medical conditions or taking medications before or during pregnancy. Echocardiography was performed and revealed a small atrial septal defect. Also, the ultrasound showed hydronephrosis, a 4mm kidney stone on the left side, and biliary sludge. The laboratory data were reported to be within the normal range considering the patient's age. Due to her cutaneous lesions, the newborn was examined by a dermatologist and the diagnosis of EB was made. She was also suspected to have concurrent PA; hence, chest radiography was performed and revealed a single bubble sign, which is the pathognomonic sign of PA. Therefore, she was prepared for surgery. Under general anesthesia in the supine position, an upper abdominal transverse incision was made. The stomach was distended, and the loops of the small intestine were completely converse. Adjacent to the pylorus, the stomach was cut with a transverse incision, and a 10-gauge catheter was inserted but did not pass the distal part of the pylorus. However, the proximal part of the pylorus was open. Atresia was evident on the distal part of the pylorus, thus pyloric resection and gastroduodenostomy were performed. Given the presence of hydronephrosis on ultrasound, along with skin lesions and surgical findings, the diagnosis of an even rarer variant of EB-PA called EB-PA-obstructive uropathy (EB-PA-OU) was suggested. After discharge from the hospital, the patient attended regular follow-up sessions. On one-year follow-up, she was alive but had slow weight gain.

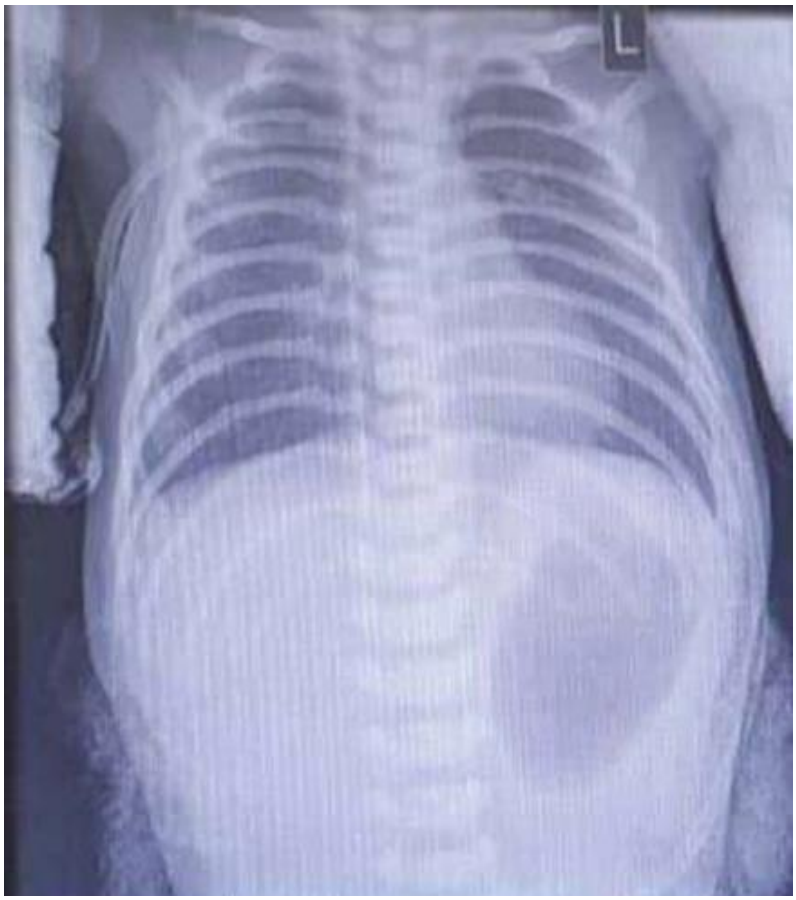

Figure 1: The single bubble sign can be seen on the patient's chest X-ray representing a dilated stomach with an air-fluid level

Discussion

A high phenotypic variability has been found among EB cases. One of the variants is EB-PA, which is an autosomal recessive genodermatosis caused by mutated ITGB4, PLEC1, and ITGA6 genes that encode a subunit of the α6β4 integrin. Ureteral and renal abnormalities occasionally coexist with EB-PA forming a variant named EB-PA-OU.[3] The pathology of urinary complications of EB is the same as skin involvement, with the urothelium being

separated from the underlying tissue at the level of the basement membrane.[6] Urinary disorders can occur in any subtype of EB, presenting with a wide range of manifestations such as dysuria, hematuria, and hydronephrosis. Although genetic analysis is available for EB, it is not necessary for diagnosis.[3]

EB-PA can also be diagnosed prenatally by observing polyhydramnios with a dilated stomach on ultrasound.[7] Recent studies have suggested complete chorioamniotic membrane separation as a novel ultrasonographic sign in prenatally diagnosed EB cases.[8-9] Contemporary case reports of EB-PA-OU highlight severe renal involvement; for instance, a 2025 report described multisystem progression to obstructive uropathy despite early intervention, including cases leading to chronic kidney disease despite ureteral reimplantation.[10-11]

EB frequently causes mortality within the first year of life, with rates exceeding 70% due to sepsis, malnutrition, and organ failure. However, select patients may survive to adulthood with manageable symptoms. For example, a 2023 review of EB-PA outcomes included long-term adult survivors presenting with isolated urinary complications, such as recurrent retention from urethral stenosis, underscoring the variable prognosis.[12-13]

Mortality is high in infancy (~80-90%), often from infection, respiratory failure, or renal complications, though survivors may reach adulthood with minimal skin issues. Recent case reports underscore variable outcomes: a 2023 preterm case with ITGB4 frameshift and missense mutations developed sepsis post-diagnosis without surgery and died shortly after, highlighting refusal of intervention as a barrier. A 2024 lethal Carmi syndrome case (JEB-PA with aplasia cutis) emphasized ethical challenges in aggressive care for severe variants. In 2025, a literature review of 49 EB-PA cases reported >50% postoperative mortality from sepsis or organ failure, but long-term survivors (up to 36 years) with mild blistering.[9,14-16]

Multidisciplinary approaches have improved prognosis, with emphasis on prenatal genetic counseling and cesarean delivery to minimize trauma. A 2025 case revealed a novel papillomatous bladder phenotype in ITGB4-related JEB-PA, with focal wall thickening and chronic inflammation causing urinary difficulty, expanding the uropathy spectrum and urging early cystoscopy. Experimental therapies like gentamicin for nonsense

variants and topical Oleogel-S10 show promise in JEB models. A 2025 review highlights gene-based (e.g., beremagene geperpavec for COL7A1-related) and cell therapies as emerging options to address symptoms, though supportive care remains core for EB-PA. Routine renal surveillance and nutritional supplements are standard to prevent malnutrition.[17-18]

Conclusion

Given the high mortality and morbidity of EB and its known association with multiorgan involvement, a systemic evaluation must be considered to detect the feasible extracutaneous disorders and prevent them from progressing to irreversible stages.

Ethical Consideration

Informed consent was obtained from all individual participants included in the study.

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Conflict of interests

There is no conflict of interest

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