Severe junctional epidermolysis bullosa in a rural infant: Challenges of diagnosis and management in primary care

Flecia Kundayis, Dr.Fam.Med (Fam Med), Erna Sazrina Binti Sahirin, MBBS

Klinik Kesihatan Lohan, Ranau, Sabah, Malaysia

SUMMARY

Epidermolysis Bullosa (EB) is a rare inherited bullous disorder that results in skin fragility and blister formation following minimal trauma. Infants with severe forms of EB are at high risk of developing recurrent infections, nutritional compromise, metabolic derangements, and life-threatening complications. Optimal management requires multidisciplinary support, specialised wound care, and access to dermatology expertise resources that may not be readily available in rural healthcare settings. We describe the case of an infant from rural Sabah who developed extensive blistering shortly after birth and subsequently experienced repeated infections, failure to thrive, electrolyte imbalance, and bronchiolitis complicated by sepsis. Despite ongoing care in the primary setting with consultation from dermatology paediatric specialists, the patient's condition deteriorated and succumbed to death at approximately two months of age. This report highlights the complexity of diagnosing and managing severe EB in a resource-limited environment and emphasises the necessity for improved referral pathways, caregiver education, and system-level support for infants with rare dermatological disorders.

INTRODUCTION

Epidermolysis Bullosa represents a group of inherited blistering disorders characterized by structural defects within the skin that lead to mechanical fragility. The condition is broadly classified into EB simplex, junctional EB, dystrophic EB, and Kindler syndrome, each defined according to the level of dermo-epidermal separation and mutations in specific structural proteins such as keratins, laminins, type VII collagen, and integrins.[1] Severe subtypes, particularly junctional EB and recessive dystrophic EB, often present in the neonatal period with widespread erosions, denuded skin, mucosal involvement, nail deformities, and chronic wounds, and are associated with high mortality.[1] The disease is associated with recurrent infections, malnutrition, growth failure, and multisystem complications.[2] Effective management requires coordinated input from dermatologists, paediatricians, dietitians, wound care nurses, and genetic counsellors. Accessing multidisciplinary care can be difficult in low-resource settings. In Malaysia, cases of EB have been reported sporadically, but the true incidence remains unknown because of diagnostic limitations and the absence of a national EB registry. Rural communities face geographical, specialist-access, and socioeconomic barriers that hinder diagnosis and management. This case illustrates the natural progression of suspected severe junctional EB in an infant from a rural community, emphasizing the clinical, logistic, and systemic challenges encountered in primary care management.

CASE PRESENTATION

The patient was a 2-month-old infant born to a young couple living in a remote rural village in Ranau, Sabah, Malaysia. Both parents were Malaysian with no formal education and were distantly related, raising the possibility of consanguinity between them. The mother had an uncomplicated pregnancy and attended regular antenatal checkups. No abnormalities were detected on routine antenatal ultrasound, and maternal screening tests, including infectious screening and a 75 g oral glucose tolerance test, were normal. The infant was delivered at term through spontaneous vaginal delivery without immediate postnatal complications.

The infant's skin appeared fragile shortly after birth, with spontaneous blistering of the limbs and pressure points observed on day 16 of life. The parents reported that even gentle handling or repositioning resulted in the formation of new bullae and erosions. Over the next few weeks, the lesions became more extensive and involved the elbows, buttocks, hands, and feet. Many of these areas developed erythema, crusting, and delayed healing. Several fingernails and toenails were dystrophic. The infant exhibited irritability and difficulty feeding, resulting in inadequate weight gain.

The family sought care at Klinik Kesihatan Lohan on Day 16 of life, where the child was assessed. The clinical presentation raised a strong suspicion of severe EB, likely junctional or recessive dystrophic type. Due to the rural setting, diagnostic tools such as skin biopsy and genetic testing were unavailable. The patient was subsequently referred to a pediatric dermatology specialist, and a skin biopsy confirmed the diagnosis of junctional epidermolysis bullosa.

Wound care consisted of daily dressings performed at the clinic using white, soft paraffin and non-adhesive gauze. Parents were educated on gentle handling techniques and instructed to avoid adhesive materials that could aggravate the skin. Consistent home wound care was challenging owing to the family's socioeconomic limitations. Pharmacological management included oral cephalexin for secondary bacterial infection of extensive skin erosions, oral nystatin for oral candidiasis contributing to feeding difficulties, and oral itraconazole for suspected fungal superinfection of chronic skin lesions following specialist consultation. Paracetamol

This article was accepted: 09 June 2026
Corresponding Author: Flecia Kundayis
Email: fleciakundayis@gmail.com

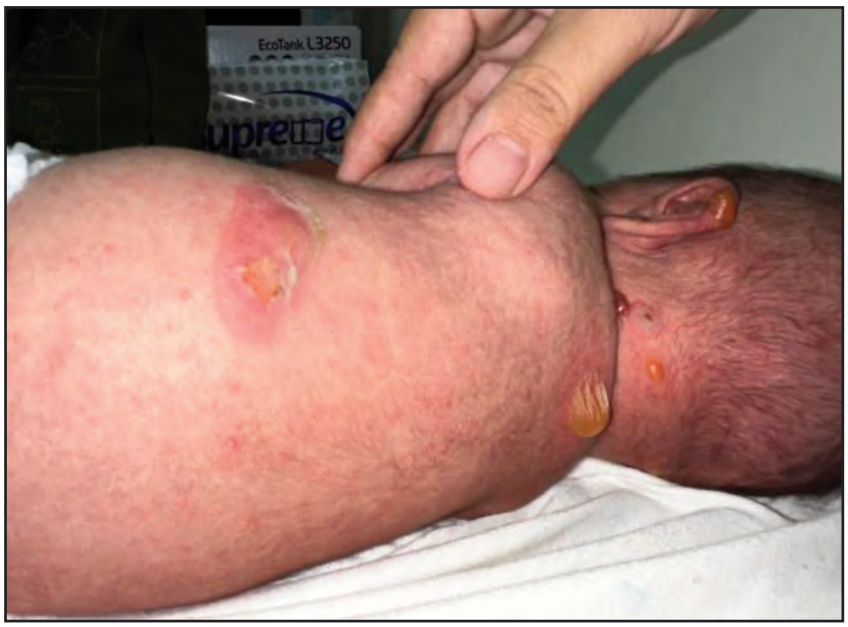

Fig. 1: Extensive tense and flaccid bullae over the earlobe, upper limbs, and trunk on Day 16 of life, consistent with severe bullous disease

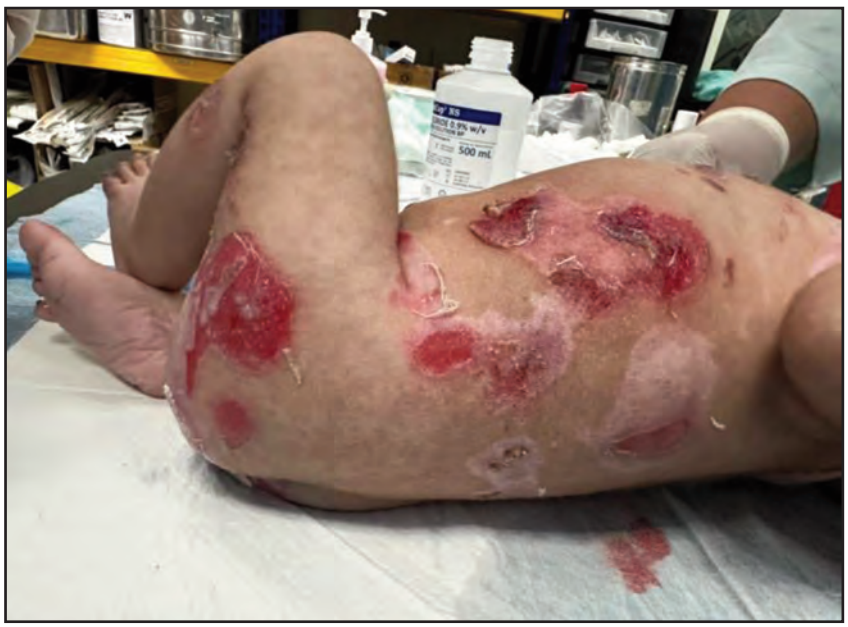

Fig. 2: Multiple erosions, crusted plaques, and healing ulcerations over pressure points at approximately 2 months of age, demonstrating chronic fragility of the skin in severe epidermolysis bullosa

was administered for pain control during routine wound care and dressing changes, and multivitamin supplementation was provided to support nutritional status and wound healing. Despite these interventions, the infant's condition remained fragile, and growth was suboptimal during subsequent follow-up visits.

At approximately two months of age, the infant developed cough, rapid breathing, lethargy, and poor feeding. On examination, he appeared tachypnoeic and had transmitted chest sounds suggestive of acute bronchiolitis. The patient was referred to the nearest district hospital. Investigations revealed severe biochemical derangements, including hyponatremia (serum sodium 126 mmol/L), renal impairment (creatinine 126 µmol/L), and transaminitis (ALT 65.2 U/L, AST 358 U/L). There was marked leukocytosis (total white cell count 25 $\times 10^9$/L) and hyperkalemia, with a potassium level of 10.2 mmol/L, alongside a significantly elevated C-reactive protein level of 196 mg/L. Serum albumin was reduced to 37 g/L, reflecting poor nutritional status and ongoing protein loss from chronic wounds. Venous blood gas analysis revealed profound metabolic acidosis, with a pH of

6.51 and bicarbonate level of 4.5 mmol/L. These findings indicate systemic infection, dehydration, and multi-organ dysfunction. Despite intensive medical management and resuscitative efforts, the infant's condition deteriorated rapidly, progressing to septic shock, severe metabolic acidosis, electrolyte derangements, and multi-organ dysfunction. He subsequently developed cardiac arrest and could not be resuscitated.

During subsequent follow-up, the mother was emotionally well and able to accept the loss with good family support. Screening for depression and anxiety using the Whooley questions and the Generalized Anxiety Disorder (GAD) screening tool was unremarkable. Genetic testing was discussed but declined because of financial constraints. She is currently not keen to conceive and has opted for intramuscular Depo-Provera for contraception. She was advised on early antenatal booking and offered a referral to a dermatology clinic for prenatal assessment should she become pregnant in the future.

DISCUSSION

This case represents a typical presentation of severe junctional epidermolysis bullosa (EB) manifesting in the neonatal period.[1,3] The early onset of blistering, widespread erosions, dystrophic nails, poor wound healing, and feeding difficulties are characteristic of severe junctional EB, which is associated with high morbidity and mortality.[1,2] In the absence of confirmatory genetic testing at the rural clinic, the diagnosis was made clinically, consistent with guidelines that recognize the value of clinical phenotyping in resource-limited settings.[1,3]

EB results from defects in proteins responsible for dermal-epidermal adhesion, leading to skin fragility and blister formation following minimal trauma. Severe subtypes are frequently associated with oral involvement, growth failure, and nutritional compromise.[2,5] The infant in this case demonstrated impaired feeding and failure to thrive, which likely contributed to the significant reduction in total protein levels.[5]

Recurrent infections are a major cause of morbidity and mortality in EB.[2] Chronic wounds act as portals of entry for microorganisms, particularly when access to wound care supplies is limited. Despite daily clinic-based wound care, the infant developed sepsis complicated by metabolic derangements and multi-organ failure. Bronchiolitis further increases infant vulnerability. Chronic inflammation, malnutrition, and fluid loss increase the risk of deterioration during acute illness.[2,5]

This case highlights the substantial challenges in managing severe junctional EB in a rural primary care setting. Access to pediatric dermatology services is limited by geographical barriers and the need for referral to distant tertiary centers. Advanced wound care products recommended for EB, such as silicone-coated mesh dressings and silver-impregnated dressings, were not readily available in the local healthcare setting. Furthermore, the family's socioeconomic limitations affected their ability to obtain specialized wound care supplies and maintain optimal wound care at home. Similar challenges have been reported in other resource-limited settings, where poverty and limited specialist services contribute to poorer outcomes.[2]

This case provides several important lessons for primary-care practitioners. Neonates presenting with extensive blistering should raise suspicion of inherited bullous disorders and warrant early referral for specialist assessment.[3] Infants with severe EB require close monitoring for infection, malnutrition, electrolyte imbalance, and poor growth, as these complications can rapidly become life-threatening.[3,5] Early multidisciplinary involvement, including dermatology, pediatrics, dietetics, nursing, and social support services, is essential to optimize outcomes.[3]

The challenges encountered in this case highlight the opportunities to improve the management of similar patients in rural communities. Early use of teledermatology may facilitate timely specialist input and reduce delays in diagnosis and treatment.[3] Standardized referral pathways for suspected EB could improve coordination between primary care facilities and tertiary centers. Access to appropriate wound care materials and structured caregiver training on dressing techniques, infection prevention, nutrition, and gentle handling may reduce complications and improve home-based care.[3] Medical social workers may help address financial barriers.

In addition to its medical complications, severe EB places a considerable psychosocial burden on caregivers.[4] Parents often experience emotional distress related to chronic wound care, frequent healthcare visits, uncertainty regarding prognosis, and the risk of life-threatening complications. In rural settings, these challenges may be compounded by financial hardship, transportation difficulties, and limited access to specialist support services. Following the infant's death, bereavement support, psychological assessment, and counselling regarding future reproductive options were important components of ongoing care. Given the possibility of parental consanguinity in this case, genetic counselling should also be considered to facilitate informed reproductive decision-making in future pregnancies.[4]

In conclusion, Epidermolysis Bullosa is a complex and debilitating condition that requires comprehensive multidisciplinary management. This case demonstrates the difficulties in diagnosing and managing severe EB in a rural Malaysian health clinic setting, where limited access to pediatric dermatology subspecialty care, socioeconomic constraints, and logistical challenges significantly hinder timely and optimal management. The rapid deterioration and fatal outcomes underscore the need for enhanced rural health resources, improved caregiver education, and strengthened referral systems to support early intervention and continuity of care for infants with EB.

REFERENCES

1. Fine J, Bruckner-Tuderman L, Eady R, Bauer E, Bauer J, Has C, Heagerty A, Hintner H, Hovnanian A, Jonkman M, Leigh I, Marinkovich M, Martinez A, McGrath J, Mellerio J, Moss C, Murrell D, Shimizu H, Uitto J, Woodley D, Zambruno G. Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. Journal of the American Academy of Dermatology 2014; 70(6): 1103-1126.
2. Bardhan A, Bruckner-Tuderman L, Chapple I, Fine J, Harper N, Has C, et al. Epidermolysis bullosa. Nature Reviews Disease Primers. 2020; 6(1): 78.
3. Saad R, Duipmans J, Yerlett N, Plevey K, McCuaig C, Woolfe W, et al. Neonatal epidermolysis bullosa: a clinical practice guideline. British Journal of Dermatology 2024; 190(5): 636-56.
4. Martin K, Geuens S, Asche J, Bodan R, Browne F, Downe A, et al. Psychosocial recommendations for the care of children and adults with epidermolysis bullosa and their family: evidence-based guidelines. Orphanet Journal of Rare Diseases 2019; 14(1): 133.
5. Dos Santos ML, Monteiro A, Nascimento A, Barbosa S, Delgado A, Zamberlan P. Nutritional Profile and Topic Management for Wound Healing in Children with Epidermolysis Bullosa: What Is the Evidence? A Systematic Review. Recent Progress in Nutrition. 2024; 4(3).