ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

Congenital Junctional Epidermolysis Bullosa: Multisystemic Complications in a Pediatric Patient

Rakan Hamad Alajmi[1], MB, BCh, BAO; Ahmed Mohsen Abbas El-Hagrasy[1], MB, BCh, BAO; Amash Hamad Alajmi[2], MD; Ghada Al-Binali[3], MD

[1] School of Medicine, Royal College of Surgeons in Ireland - Medical University of Bahrain (RCSI Bahrain), P.O. Box 15503, Building No. 2441, Road 2835, Busaiteen, Kingdom of Bahrain.
[2] School of Medicine, Jordan University of Science & Technology (JUST), P.O. Box 3030, Irbid, 22110, Jordan.
[3] Department of Ophthalmology, Military Hospital, Royal Medical Services, Kingdom of Bahrain.

Corresponding Author

Rakan Hamad Alajmi
MB, BCh, BAO School of Medicine, Royal College of Surgeons in Ireland - Medical University of Bahrain P.O. Box 15503, Building No. 2441, Road 2835, Busaiteen, Kingdom of Bahrain. Email: 20205967@rcsi-mub.com

Abstract:

Background: Congenital junctional epidermolysis bullosa (CJEB) is a rare inherited blistering disorder caused by defects in dermal–epidermal adhesion, resulting in marked skin and mucosal fragility. Although primarily considered a cutaneous disease, CJEB may also cause significant extracutaneous morbidity requiring multidisciplinary care.

Case Presentation: We report a 14-year-old Bahraini male with CJEB and progressive multisystem involvement. Cutaneous manifestations began in early infancy with recurrent rash, nail dystrophy, and secondary infection. Over time, he developed severe airway disease with laryngeal stenosis requiring emergency tracheostomy and subsequent airway interventions. He also experienced progressive cicatrizing ocular surface disease with recurrent symblepharon, conjunctival fibrovascular overgrowth, corneal scarring, and eventual corneal melt requiring multiple reconstructive ophthalmic procedures. In late childhood, genitourinary involvement emerged with penile inflammation, meatal narrowing, urinary retention, and recurrent urethral stricture requiring repeated dilatations and meatotomy.

Management and Outcome: Management was supportive and multidisciplinary, involving dermatology, otolaryngology, ophthalmology, pediatrics, and urology. Interventions included tracheostomy care, laryngeal surgery, repeated ocular surface reconstruction with amniotic membrane transplantation and mitomycin C, catheterization, urethral dilatation, meatotomy, infection treatment, wound care, and long-term surveillance.

Conclusion: This case highlights the severe multisystem burden of CJEB and emphasizes that it is not solely a skin disorder. Early recognition of extracutaneous complications, close follow-up, and coordinated multidisciplinary management are essential to reduce morbidity and preserve function and quality of life.

Keywords:

Congenital junctional epidermolysis bullosa, CJEB, Epidermolysis bullosa, Airway involvement, Ocular complications, Urethral stricture, Multidisciplinary management

Received : 13-05-2026 **Revised : 20-05-2026** **Accepted: 22-05-2026** **Published : 29-05-2026**

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

Introduction

Congenital Junctional Epidermolysis Bullosa (CJEB) is a rare congenital genetic disorder characterized by extreme fragility of the skin and mucous membranes. Minor trauma frequently results in extensive blistering and ulceration, which are exacerbated by secondary infections, scarring, and multisystem involvement. CJEB arises from mutations in genes responsible for dermal-epidermal adhesion, most commonly *LAMB3*, *LAMC2*, and *COL17A1* [1]. CJEB occurs globally but remains extremely rare, with a prevalence of fewer than 1 in 1,000,000 live births [2]. Current evidence indicates no significant predilection for sex or ethnic group.

Classification of CJEB is based on clinical severity, age of symptom onset, and underlying genetic mutations. The two principal subtypes are Herlitz, which is frequently fatal, and non-Herlitz, which presents with a milder clinical course [3]. Advances in genetic testing have improved early diagnosis, prognostic assessment, and genetic counseling for affected families [4].

While CJEB primarily affects the skin and mucous membranes, it can also lead to significant complications and multi-organ involvement [1]. In the present case, the patient experienced progressive airway disease requiring tracheostomy, recurrent ophthalmologic complications necessitating surgical intervention, and additional extra-cutaneous manifestations. This complexity underscores the necessity for multidisciplinary management in patients with CJEB. Therefore, this case report presents a rare case of CJEB manifesting in a 14-year-old male with multi-organ involvement, aiming to explore the various manifestations of CJEB, the complications that may arise, and potential interventions required to effectively manage this condition.

Case Presentation

This case report presents a 14-year-old Bahraini male, at a height of 139 cm and a weight of 27 kg, diagnosed with Congenital Junctional Epidermolysis Bullosa, and has a reported positive history of Methicillin-resistant *Staphylococcus aureus* (MRSA).

a. Perinatal, Birth History, Family History, and Diagnosis The patient was a term male infant delivered by spontaneous vaginal delivery, with no neonatal intensive care unit admission. The neonatal period was notable only for mild, physiologic jaundice lasting approximately 7 days (reported serum bilirubin 15.9), with normal glucose-6-phosphate dehydrogenase (G6PD) screening and normal hemoglobin electrophoresis.

Cutaneous symptoms began early in life, with a persistent diaper rash from around 2 months of age, followed by progressive facial post-inflammatory hyperpigmentation at sites of nail-induced excoriations and associated nail dystrophy and discoloration by approximately 6 months. At the same age, mucocutaneous fungal infection and eczema with secondary bacterial infection were documented. He was followed in dermatology and was diagnosed at 6 months of age with epidermolysis bullosa simplex.

Early mucosal and ocular involvement was also evident during infancy, including keratoconjunctivitis with purulent discharge from a right scleral flap at approximately 10 months, pyogenic granulomas affecting both eyes by 11 months, and significant right ocular surface injury by 15 months, characterized by right ear pain with inability to open the eye and a large right corneal abrasion; a right conjunctival dermoid was also noted at that time.

The family history was highly suggestive of an inherited blistering disorder: the parents were consanguineous (cousins), one sibling reportedly had a similar condition and died at 4 years of age, and three other siblings were unaffected.

b. Cutaneous Findings Cutaneous manifestations began in early infancy, with a recurrent diaper rash reported from approximately 2 months of age. By around 6

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

months, the patient developed progressive facial hyperpigmented macules at sites of nail-induced excoriations, accompanied by nail abnormalities, including discoloration and dystrophic changes, clinically described as facial lesions with dystrophic nail involvement. Mucocutaneous candidiasis was also documented at approximately 6 months, alongside eczematous dermatitis complicated by secondary infection. On longitudinal follow-up in the dermatology clinic, he was diagnosed at 6 months of age with epidermolysis bullosa simplex.

In July 2012, he re-presented with a 4-month history of an erythematous facial rash associated with nail disorder. In December 2012, at 16 months of age, he presented with an acute onset of bilateral foot erythema and marked swelling with refusal/inability to stand or walk, in the absence of preceding trauma or insect bite, and with documented fever (38–39°C); examination described severe bilateral pedal edema and erythema. Given the recurrent, infective-appearing cutaneous disease, an evaluation was planned to assess immune function and to exclude underlying immunodeficiency, including chronic granulomatous disease (CGD). Cardiovascular examination was otherwise unremarkable, and general activity was reported as preserved.

c. Ear, Nose, Throat & Airway Findings The patient developed significant upper-airway involvement in early childhood, culminating in an emergency tracheostomy at 3 years and 5 months of age for laryngeal stenosis. At 3 years and 9 months, he subsequently underwent excision of a laryngeal lesion with concurrent skin biopsy. In January 2015, he presented with increased work of breathing characterized by tachypnea and bilateral wheeze on auscultation without rhonchi; following nebulized therapy, he became clinically comfortable with only minimal expiratory wheeze, maintaining an oxygen saturation of 100% on room air (respiratory rate 34/min), and was discharged with as-needed medication for episodes of dyspnea or cyanosis.

In June 2015, at one-week post-excision of a supraglottic lesion, he was reported to be clinically well with a patent Shiley 3.5 mm tracheostomy tube in situ. Flexible fiberoptic laryngoscopy visualized the vocal cords, although assessment of mobility was limited at that stage; the cords appeared adducted, and the cough reflex was preserved, with a plan for short-interval reassessment of vocal cord mobility and laryngeal patency. On repeat review two weeks post-operatively, laryngoscopy demonstrated a small interarytenoid lesion not obstructing the supraglottic airway; however, the vocal cords appeared stenotic, and follow-up was arranged for tracheostomy tube change and interval reassessment.

By October 2016, the family reported improved voice loudness, and he was counseled on abdominal breathing techniques and the use of a speech valve. In February 2017, culture from the tracheostomy site grew methicillin-resistant *Staphylococcus aureus* (MRSA) and was treated with trimethoprim–sulfamethoxazole, with subsequent clinical improvement and reduced tracheal secretions; suctioning at review yielded minimal secretions. *(See Figure 1 for Chest Radiograph with Tracheostomy).*

d. Ophthalmologic Findings Ocular involvement was evident from infancy with recurrent conjunctival inflammation and secondary infection. In June 2012, the patient presented with right-eye discharge; examination demonstrated a scleral flap with purulent discharge, and topical antibiotics were prescribed (ofloxacin drops and oxytetracycline ointment). In July 2012, erythematous mucosa was noted at the lateral canthi bilaterally without active discharge. By November–December 2012, he developed episodic right-eye pruritus with swelling and conjunctivitis and was later referred from the emergency department with suspected right corneal abrasion; fluorescein staining was positive, and patching with short-interval review was advised.

In July 2013, as a child with epidermolysis bullosa, he again presented with ocular irritation and discharge (right > left) without an obvious epithelial defect, managed with topical moxifloxacin and frequent lubricants. By October 2016, progressive conjunctivalization

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

was documented, with conjunctival tissue encroaching onto the cornea (right > left) and bilateral conjunctival hypertrophy, consistent with significant cicatrizing ocular surface disease.

Surgical management was planned to address recurrent conjunctival overgrowth and adhesions. In February 2017, he underwent operative intervention, including excision of conjunctival cysts in both eyes, symblepharon release, superficial keratectomy, intraoperative mitomycin C application, and amniotic membrane transplantation (AMT). Early postoperative care included bandage contact lens placement, conformer insertion, and temporary tarsorrhaphy (right eye), followed by further symblepharon correction with repeat amniotic grafting and conformer application (March 2017). Despite these interventions, the disease course was characterized by recurrence, with limbal conjunctival fibrovascular growth extending toward the cornea (right > left) and bilateral discharge documented in November 2017, and persistent bilateral adhesions noted in January 2018. He was later admitted at approximately 5 years of age for conjunctival granuloma excision, reflecting ongoing proliferative and cicatrizing conjunctival pathology.

Ocular surface complications continued into adolescence with recurrent conjunctival fibrovascular growth and symblepharon requiring repeated surgical reconstruction. In February 2022, he underwent excision of a right-eye conjunctival fibrovascular growth, with symblepharon division, mitomycin C, and AMT. In December 2024, he was admitted for bilateral symblepharon release with bilateral AMT and prolonged topical therapy (tobramycin–dexamethasone drops/ointment and long-term hyaluronate lubrication). In early 2025, he exhibited persistent epithelial defects with bandage contact lenses in situ, bilateral corneal scarring, and markedly reduced vision (left eye 6/30; right eye hand motion). By February–March 2025, the right eye developed a recurrent epithelial defect without overt infection, while the left eye deteriorated with staining-positive central thinning and subsequent corneal infiltrate with thinning; examination under anesthesia confirmed corneal melt in the left eye without perforation. Intensive antimicrobial therapy was initiated (fortified vancomycin and ceftazidime for the left eye, with topical fluoroquinolone for the right eye), and further ocular surface reconstruction was performed in March 2025, including a double-layer AMT with fibrin glue for the left eye and AMT for the right eye, underscoring the severe, recurrent cicatrizing ocular surface disease associated with congenital junctional epidermolysis bullosa. *(See Figure 2).*

e. Urological Findings Genitourinary involvement emerged in late childhood with inflammatory penile disease complicated by progressive outlet obstruction, consistent with mucosal fragility and scarring in congenital junctional epidermolysis bullosa. In April 2021, the patient was admitted with a 1-week history of progressive dysuria and penile swelling, associated with initial hematuria. Urinalysis demonstrated marked hematuria and pyuria (RBC ~100/hpf, WBC ~100/hpf) with significant proteinuria (3+), positive leukocyte esterase (4+), and nitrites (2+). Examination noted a ventral penile wound at the coronal level with surrounding edema/cellulitis and functional meatal narrowing, felt likely secondary to edema and EB-related penile involvement with risk of scarring and meatal stenosis.

Ultrasound at that time supported the diagnosis of cystitis, demonstrating a thick-walled, mildly distended bladder containing turbid fluid *(See Figure 3)*. He was treated with intravenous fluids and ceftriaxone and discharged on oral co-amoxiclav with topical fusidic acid/betamethasone (Fucicort) for local penile inflammation.

In September 2021, he re-presented with acute urinary retention requiring urethral catheterization and intravenous antibiotics, and subsequently underwent urethral dilatation prior to discharge. Thereafter, the course was characterized by recurrent urethral narrowing requiring repeated procedures: urethral stricture dilatation (February 2022), urinary meatal

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

dilatation with meatotomy and short-term catheterization (April 2022), and further planned/interval urethral dilatations (August 2022), with the last documented meatotomy in September 2022. At follow-up in October 2023 for urethral stricture disease, ongoing maintenance urethral dilatation (1–2 times weekly) was advised to reduce the risk of recurrent stenosis and urinary tract infections. Overall, the pattern suggests chronic cicatricial genitourinary disease with recurrent meatal/urethral stenosis and episodes of cystitis/retention as clinically important urological complications in this CJEB phenotype.

f. Gastrointestinal or Nutritional Issues Gastrointestinal symptoms were intermittently reported in mid-childhood without evidence of chronic malabsorptive or obstructive complications documented in the available records. In September 2020, at approximately 9 years of age, the patient was admitted under the surgical team for observation due to abdominal pain with vomiting, clinically managed as acute gastroenteritis. Supportive treatment was provided, including intravenous fluids (Ringer's lactate at 60 mL/hour) and symptomatic therapy (intravenous paracetamol and metoclopramide as needed). He improved without requiring procedural intervention, was discharged home in stable condition, and was advised on dietary modification, including avoidance of spicy foods. Symptoms were also noted to improve with acid suppression, and he was discharged on a 6-week course of omeprazole.

g. Hospital Admissions Hospitalizations in this patient with congenital junctional epidermolysis bullosa demonstrated a progressive, multisystem course dominated by recurrent mucocutaneous inflammation/infection and cicatricial sequelae affecting the airway, ocular surface, and genitourinary tract. In infancy (9–16 months), admissions and emergency presentations were driven by severe cutaneous disease with secondary infection and early ocular involvement, including purulent keratoconjunctivitis and corneal epithelial injury. By 3 years 5 months (January 2015), airway disease became clinically significant, culminating in emergency tracheostomy for laryngeal stenosis, followed by operative management of supraglottic/laryngeal lesions and serial endoscopic reassessment. From 5 years onward, the ophthalmologic course was characterized by severe, recurrent cicatrizing ocular surface disease requiring repeated surgical interventions (symblepharon release, superficial keratectomy, mitomycin C, and amniotic membrane transplantation), later complicated by persistent epithelial defects, corneal scarring, and corneal thinning/melt necessitating examination under anesthesia, intensive fortified antimicrobials, and repeat amniotic membrane grafting in adolescence.

Genitourinary admissions began at 9 years 8 months with UTI/cystitis and penile involvement with evolving meatal narrowing, progressing to recurrent urinary retention and urethral stricture disease requiring repeated urethral dilatations and meatotomy, with ongoing maintenance dilatation advised to reduce recurrence and UTI risk. A single gastrointestinal admission at 9 years 1 month was for observation of abdominal pain and vomiting and resolved with conservative management without surgical intervention. *(See Figure 4 for Multisystem Timeline).*

Discussion

Epidermolysis Bullosa (EB) includes a wide range of genodermatoses affecting cutaneous membranes and is characterized mainly by fragility and blistering of skin, and is usually associated with other extracutaneous manifestations. EB subtypes include simplex, junctional, and dystrophic; histopathological confirmation is required to determine the subtype [5]. The prevalence of all EB subtypes is estimated at approximately 11 to 45 per million live births, based on data from the United States, England, Wales, Germany, and Slovenia, with an estimated prevalence of junctional EB at 1-14.23 per million [6–8].

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

The subtype of junctional EB present from birth is termed CJEB to describe the congenital nature of the condition. The main underlying pathophysiological mechanism involves a defect in attachment between or within the epidermis and dermis layers of the skin, usually caused by genetic mutations [9]. Further subclassification can be based on the severity of disease and is usually characterized as generalized severe CJEB or generalized intermediate CJEB. Impacted organs may include the skin, eyes, ears, nose, upper airway, as well as the gastrointestinal and genitourinary tracts, which require a multidisciplinary team approach to successfully manage patients as complications arise, as there is no current cure for the condition [9].

Differential Diagnosis JEB generalized severe (formerly known as Herlitz JEB) and JEB generalized intermediate (formerly known as non-Herlitz JEB) are both autosomal recessive genetic mutations in the same genes (*LAMA3*, *LAMB3*, or *LAMC2*), which disrupt the skin anchoring proteins; they differ significantly in clinical severity and prognosis [10]. Herlitz is a more severe and typically lethal form where a near total absence of the laminin-332 results in extensive blistering of the skin and internal mucosal linings from birth. These infants often suffer from severe granulation tissue, respiratory distress, and malnutrition, and rarely survive beyond their first year of life. Whereas non-Herlitz represents a milder disease spectrum in which partial protein function permits a generally normal life expectancy. Although blistering may remain widespread, it is frequently localized to the hands, feet, and joints. Infants are more likely to develop secondary symptoms such as dental enamel defects, nail loss, and hair thinning, rather than life-threatening internal complications characteristic of the Herlitz type [1].

While all forms of epidermolysis bullosa (EB) share the hallmark of mechanical skin fragility, they are primarily distinguished by the specific ultrastructural level where blistering occurs within the skin layers [11]. EB Simplex (EBS) is the most common and generally mildest form, characterized by intraepidermal blistering that usually heals without significant scarring [12]. In contrast, Junctional EB (JEB) occurs within the lamina lucida of the basement membrane and is uniquely associated with dental enamel hypoplasia and, in severe cases, life-threatening periorificial granulation tissue. Dystrophic EB (DEB) involves deeper cleavage below the lamina densa, leading to profound scarring, milia, and severe physical deformities, such as finger fusion (pseudosyndactyly) [13]. Finally, Kindler syndrome is distinct for its mixed nature, as blistering can occur at multiple levels of the skin, and it is the only subtype to feature photosensitivity and progressive poikiloderma (mottled skin pigmentation) [14].

Diagnostic confirmation of epidermolysis bullosa (EB) subtypes depends on identifying the specific protein deficiency and the precise level of tissue separation using specialized laboratory techniques [15]. In EB Simplex, immunofluorescence mapping (IFM) demonstrates an intraepidermal split with generally normal protein expression; however, rare recessive cases may exhibit an absence of keratin 5 or 14 [16]. Junctional EB is defined by a cleavage plane within the lamina lucida. In the Herlitz type, immunoblotting reveals a complete absence of laminin-332 due to null mutations, whereas the non-Herlitz type shows reduced or altered expression of laminin-332 or collagen XVII [14]. Dystrophic EB is characterized by a sublamina densa split, with IFM and immunoblotting indicating a significant reduction or total loss of collagen VII, typically associated with *COL7A1* mutations [16, 17]. Kindler syndrome is distinguished by a unique mixed cleavage pattern at multiple levels, with mutation analysis confirming defects in the *FERMT1* gene and a corresponding loss of kindlin-1 protein [14].

Investigations Diagnosis begins with a skin biopsy of a fresh blister analyzed via transmission electron microscopy (EM), which reveals a definitive cleft at the lamina lucida. This cleavage occurs within the middle layer of the basement membrane due to the structural failure of anchoring proteins. EM findings typically show hypoplastic or absent hemidesmosomes, the rivets that secure the

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

epidermis to the dermis; their total absence is a hallmark of the severe Herlitz type, while reduced numbers often characterize non-Herlitz forms [16, 18–20]. Genetic confirmation is used to pinpoint the underlying molecular defect, typically targeting the *LAMA3*, *LAMB3*, and *LAMC2* genes (laminin-332) or *COL17A1* (collagen XVII). Severe Herlitz JEB is identified by null mutations that result in a complete absence of protein, whereas non-Herlitz types usually involve missense or splice-site mutations that allow for some residual protein function. This genetic blueprint is essential for distinguishing JEB from other subtypes and for determining the long-term prognosis [18, 21, 22].

Relevant laboratory tests are vital for monitoring the systemic impact of chronic skin loss. Patients frequently present with microcytic anemia and hypoalbuminemia due to chronic inflammation and protein loss through blister fluid. Additionally, because the skin barrier is permanently compromised, frequent wound cultures and blood tests are necessary to screen for bacterial infections or sepsis, which are commonly caused by *Staphylococcus aureus* or *Pseudomonas* [16, 21].

General Management Principles & Follow Up Given that there is no cure to date for the condition, general management principles include a multidisciplinary, expectant, watchful waiting approach to manage complications as they arise, while providing necessary preventive measures to minimize morbidity [9]. However, there have been many advances in treatment methods, including the potential use of gene therapy, protein replacement therapy, cell therapy (allogeneic fibroblasts, mesenchymal stromal cells), bone marrow stem cell transplant, culture/vaccination of revertant mosaic keratinocytes, gene editing/engineering, and the clinical application of inducible pluripotent stem cells [23]. Although not yet in routine clinical practice, these advances offer greater hope for patients with CJEB to modify the disease process and improve their quality of life while battling the condition.

The main pillars of management include wound care, controlling pain, minimizing risk of infection, providing adequate nutritional support, and supportive treatment for complications [9]. The primary methods of wound prevention are protecting fragile skin from friction, carefully draining blisters to prevent extension, and using dressings as needed [24]. Wound care involves the use of advanced non-adherent, absorbent, antimicrobial dressings that reduce pain, the risk of infection, exudate, and the risk of increased trauma during dressing changes. These dressings include hydrogel/solidifying agents, chitosan antibacterial dressings, honey-based dressings, Cutimed Sorbact, polyhexanide dressings, PolyMem, and ibuprofen-containing dressings [23]. Antibiotic use in these patients is not considered routine practice but is usually reserved for supportive treatment of recurrent infections and secondary wound infections caused by organisms such as MRSA and *Pseudomonas aeruginosa* [24]. Beyond routine dressings, skin grafts or tissue-engineered skin substitutes are promising, with products such as Apligraf, Biobrane, OrCel, and BIOOPA becoming important options for chronic or difficult wounds [23]. Gene therapy and stem-cell–based treatments are highly promising for future disease modification, but these are not yet standard wound-care therapy.

Complication-directed management through an MDT approach is required by dermatologists, otolaryngologists, ophthalmologists, and pediatric urologists, who provide supportive measures as needed to help improve the patient's quality of life. Tracheostomy care and airway surveillance are required in selected patients with laryngeal granulation tissue or airway compromise; ophthalmic surgery, including amniotic membrane transplantation (AMT) with or without mitomycin C (MMC), may be needed for severe ocular surface disease to promote epithelial healing and limit cicatricial scarring; urological management is important when genitourinary defects or obstruction are present; and because CJEB is a multisystem disorder, patients should have regular multidisciplinary follow-up with dermatology, ENT, ophthalmology, and pediatric surgery to monitor and treat evolving

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

skin, airway, eye, and visceral complications [16, 25, 26].

In CJEB, supportive care extends beyond wound treatment and is essential for preserving function and quality of life. Because blistering and hyperkeratosis can impair walking, appropriate footwear and physical therapy are important to maintain ambulation and prevent further disability. Psychosocial support, including social services and psychological counseling, is also a key component of care, given the chronic, burdensome nature of the disease. In addition, chronic pain management should be addressed proactively, with specialist referral when pain is difficult to control, while regular dental follow-up is necessary because enamel abnormalities are common and can lead to significant oral morbidity. Together, these measures form an important part of comprehensive multidisciplinary management in CJEB [16].

Outcome and Prognosis Prognosis in CJEB is variable and depends largely on disease severity and extracutaneous involvement. Survival has improved in severe JEB, with one recent cohort showing a trend toward longer survival over 19 years and a median survival of 12.7 months in JEB-S during the most recent 5-year period, likely reflecting better multidisciplinary care [7]. In survivors with milder or intermediate JEB phenotypes, growth and long-term function may be more favourable; in a 200-child cohort, growth in JEB-generalized intermediate resembled that of healthy children, although anaemia could still occur from the second year of life onward [27]. Functional burden remains substantial: in a registry-based study of children with EB, only 31% of children with JEB were fully independent in walking, and independence in other activities of daily living ranged from 39% to 73%, highlighting the impact on mobility and daily function [28]. Quality of life is also significantly affected, with worse QoL reported in more severe EB subtypes, particularly when skin involvement is extensive [9, 29]. School participation may also be impaired; although CJEB-specific attendance data are limited, a recent EB education study found frequent absences in 51.6% of students, and students with junctional EB reported more negative school experiences [30]. Overall, these findings show that outcomes in CJEB are determined not only by survival but also by growth, mobility, pain, school participation, and psychosocial burden, underscoring the importance of sustained multidisciplinary follow-up [9].

Learning Points

CJEB extends far beyond the skin. The most severe complications in this case were emergency tracheostomy, corneal melt, and urinary retention, all of which were extracutaneous. Always screen for systemic involvement at every visit.

Airway disease can be sudden and life-threatening. Laryngeal stenosis developed silently and required emergency tracheostomy at age 3. Any change in voice, stridor, or breathing warrants urgent ENT referral.

Ocular disease is progressive and vision-threatening. Despite repeated surgical interventions, this patient developed bilateral corneal scarring and eventual corneal melt by adolescence. Early ophthalmology involvement and aggressive lubrication are essential.

Genitourinary involvement is underrecognized. Mucosal scarring can affect the urethra. Routinely ask CJEB patients about urinary symptoms, as meatal and urethral stenosis may develop insidiously.

Multidisciplinary care is the cornerstone of management. With no current cure, coordinated longitudinal care across specialties is the only means of preserving function and quality of life.

Consanguinity should prompt earlier, more aggressive surveillance. A family history of inherited blistering disorders in a consanguineous family warrants prompt genetic confirmation and anticipatory subspecialty referral.

Resistant organisms are a real and recurring threat. Frequent wound cultures and targeted antimicrobial therapy, rather than routine broad-spectrum antibiotics, are essential, given the risk of MRSA and polymicrobial infections.

Conclusion

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

This case highlights the severe multisystem burden of congenital junctional epidermolysis bullosa, extending beyond cutaneous fragility to significant airway, ocular, and genitourinary complications. It emphasizes the importance of early recognition of extracutaneous involvement, close longitudinal surveillance, and coordinated multidisciplinary care to reduce morbidity and preserve function and quality of life.

References

1. MedlinePlus Genetics. Junctional epidermolysis bullosa [Internet]. Bethesda (MD): National Library of Medicine (US); [cited 2026 Mar 9]. Available from: https://medlineplus.gov/genetics/condition/junctional-epidermolysis-bullosa/
2. Baardman R, et al. Novel insights into the epidemiology of epidermolysis bullosa (EB) from the Dutch EB Registry: EB more common than previously assumed? J Eur Acad Dermatol Venereol [Internet]. 2021 [cited 2026 Mar 9]. Available from: https://onlinelibrary.wiley.com/doi/10.1111/jdv.17012
3. Genetic and Rare Diseases Information Center (GARD). Junctional epidermolysis bullosa [Internet]. Gaithersburg (MD): National Institutes of Health; [cited 2026 Mar 9]. Available from: https://rarediseases.info.nih.gov/diseases/2152/junctional-epidermolysis-bullosa
4. Baardman R, et al. Evolution of genome diagnostics in epidermolysis bullosa: Unveiling the power of next-generation sequencing. J Eur Acad Dermatol Venereol [Internet]. 2025 [cited 2026 Mar 9]. Available from: https://onlinelibrary.wiley.com/doi/full/10.1111/jdv.19938
5. Kao CH, Chen SJ, Hwang B, Yang AH, Hsu CY, Huang CH. Junctional Epidermolysis Bullosa. J Chin Med Assoc. 2006;69(10):503-506.
6. Has C, Hess M, Anemüller W, et al. Epidemiology of inherited epidermolysis bullosa in Germany. J Eur Acad Dermatol Venereol. 2023;37(2):402-410.
7. Petrof G, Papanikolaou M, Martinez AE, et al. The epidemiology of epidermolysis bullosa in England and Wales: data from the national epidermolysis bullosa database. Br J Dermatol. 2022;186(5):843-848.
8. Štublar A, Dragoš V, Dolenc-Voljč M. Inherited epidermolysis bullosa: epidemiology and patient care in Slovenia with a review of the updated classification. Acta Dermatovenerol Alp Pannonica Adriat. 2021;30(2):63-66.
9. Hon KL, Chu S, Leung AKC. Epidermolysis Bullosa: Pediatric Perspectives. Curr Pediatr Rev. 2022;18(3):182-190.
10. Khanna D, Bardhan A. Epidermolysis Bullosa [Internet]. Treasure Island (FL): StatPearls Publishing; 2026 [cited 2026 Mar 9]. Available from: http://www.ncbi.nlm.nih.gov/books/NBK599531/
11. Genetic Clinics [Internet]. [cited 2026 Mar 9]. Available from: https://iamg.in/genetic_clinics/full_text7520.html?id=284
12. Naqvi E. Types of Epidermolysis Bullosa [Internet]. Epidermolysis Bullosa News. [cited 2026 Mar 9]. Available from: https://epidermolysisbullosanews.com/types-of-epidermolysis-bullosa/
13. Sait H, Srivastava S, Saxena D. Integrated Management Strategies for Epidermolysis Bullosa: Current Insights. Int J Gen Med. 2022;15:5133-5144.
14. MSD Manual Professional Edition. Epidermolysis Bullosa - Dermatology [Internet]. [cited 2026 Mar 10]. Available from: https://www.msdmanuals.com/professional/dermatologic-disorders/bullous-diseases/epidermolysis-bullosa
15. Medscape. Epidermolysis Bullosa: Background, Pathophysiology, Etiology [Internet]. 2025 Sep 25 [cited 2026 Mar 10]. Available from: https://emedicine.medscape.com/article/1062939-overview
16. Pfendner EG, Lucky AW. Junctional Epidermolysis Bullosa. In: Adam MP, et al, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington; 1993-

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

2026 [cited 2026 Mar 10]. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1125/

17. Barzegar M, Asadi-Kani Z, Mozafari N, Vahidnezhad H, Kariminejad A, Toossi P. Using immunofluorescence (antigen) mapping in the diagnosis and classification of epidermolysis bullosa: a first report from Iran. *Int J Dermatol*. 2015;54(10):e416-e423. doi:10.1111/ijd.12804
18. Chen F, Wei R, Wang Y, et al. Identification of deep intronic variants in junctional epidermolysis bullosa using genome sequencing and splicing assays. Npj Genomic Med. 2025;10(1):8.
19. Fine JD, Bruckner-Tuderman L, Eady RAJ, et al. Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. J Am Acad Dermatol. 2014;70(6):1103-1126.
20. Yuen WY, Lemmink HH, van Dijk-Bos KK, Sinke RJ, Jonkman MF. Herlitz junctional epidermolysis bullosa: diagnostic features, mutational profile, incidence and population carrier frequency in the Netherlands. Br J Dermatol. 2011;165(6):1314-1322.
21. IVAMI. Genetic testing - Junctional epidermolysis bullosa – COL17A1, LAMA3, LAMB3 and LAMC2 genes [Internet]. [cited 2026 Mar 10]. Available from: https://www.ivami.com/en/genetic-testing-human-gene-mutations-diseases-neoplasias-and-pharmacogenetics/6066-genetic-testing-junctional-epidermolysis-bullosa-i-col17a1-lama3-lamb3-i-and-i-lamc2-i-genes
22. Sproule TJ, Bubier JA, Grandi FC, et al. Molecular Identification of Collagen 17a1 as a Major Genetic Modifier of Laminin Gamma 2 Mutation-Induced Junctional Epidermolysis Bullosa in Mice. PLOS Genet. 2014;10(2):e1004068.
23. Nita M, Pliszczyński J, Kosieradzki M, Fiedor P. Review of the Latest Methods of Epidermolysis Bullosa and Other Chronic Wounds Treatment Including BIOOPA Dressing. Dermatol Ther. 2021;11(5):1469-1480.
24. Namboothiri SP. Junctional epidermolysis bullosa in children: an overview. Clin Res Commun. 2023;6(1):2.
25. Swarup A, Ta CN, Wu AY. Molecular mechanisms and treatments for ocular symblephara. Surv Ophthalmol. 2022;67(1):19-30.
26. Eslani M, Baradaran-Rafii A, Movahedan A, Djalilian AR. The Ocular Surface Chemical Burns. J Ophthalmol. 2014;2014(1):196827.
27. Reimer A, Hess M, Schwieger-Briel A, et al. Natural history of growth and anaemia in children with epidermolysis bullosa: a retrospective cohort study. Br J Dermatol. 2020;182(6):1437-1448.
28. Fine JD, Johnson LB, Weiner M, Suchindran C. Assessment of mobility, activities and pain in different subtypes of epidermolysis bullosa. Clin Exp Dermatol. 2004;29(2):122-127.
29. Tabolli S, Sampogna F, Di Pietro C, et al. Quality of life in patients with epidermolysis bullosa. Br J Dermatol. 2009;161(4):869-877.
30. Alheggi A, Bin Shlhoob R, Alharthi R, Bukhari A, Alkhodair R. Addressing the Unmet Educational Needs of Students with Epidermolysis Bullosa in Saudi Arabia. Risk Manag Healthc Policy. 2025;18:2565-2573.

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

Figures

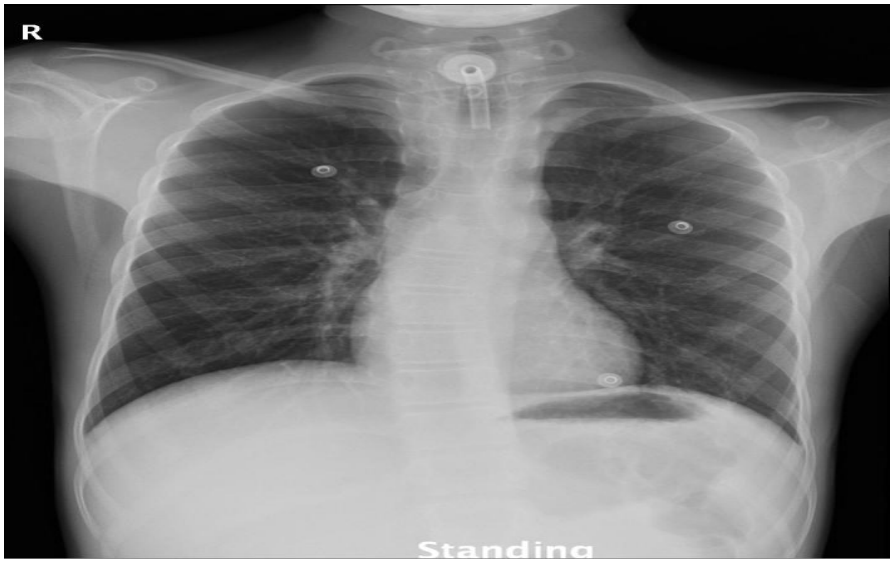
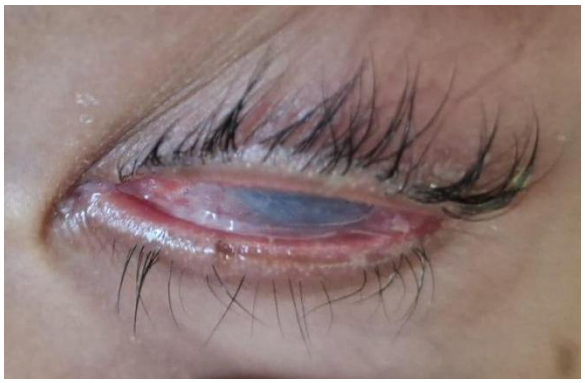


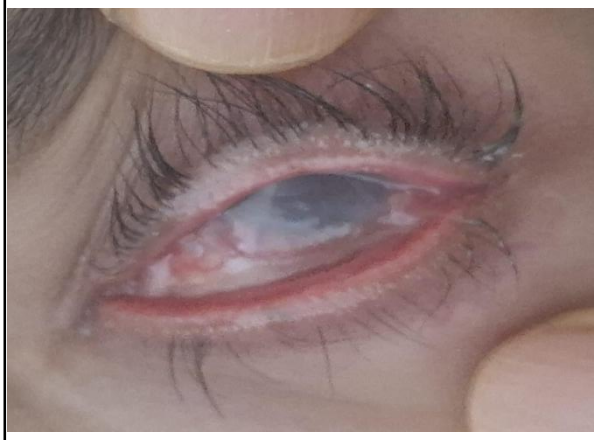
Figure 1: Chest radiograph with tracheostomy tube in situ. Standing AP chest X-ray demonstrates a tracheostomy tube in the upper trachea, with an otherwise unremarkable cardiomediastinal silhouette and lung fields. External ECG monitoring leads are seen overlying the chest.

	
A: Right eye – conjunctival hyperemia with corneal surface opacity	**B:** Left eye – conjunctival hyperemia with corneal surface opacity

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

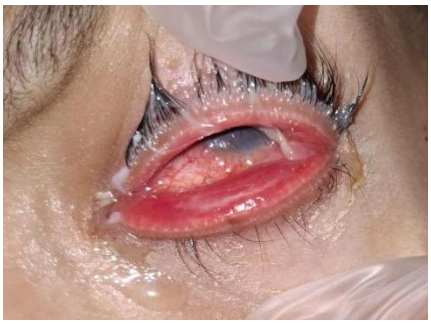
Journal of Dermatological Case Reports

C: Right eye – fibrovascular conjunctival tissue encroaching onto the cornea

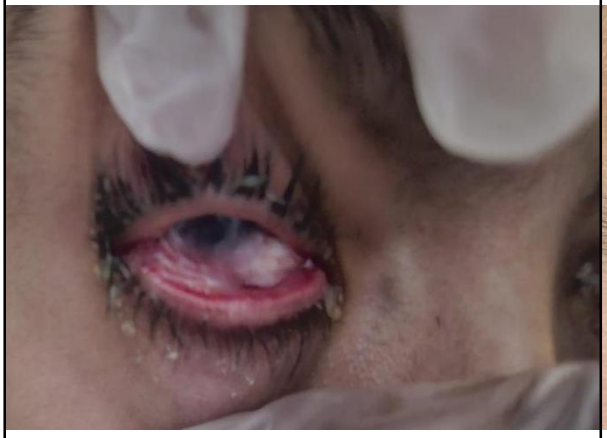


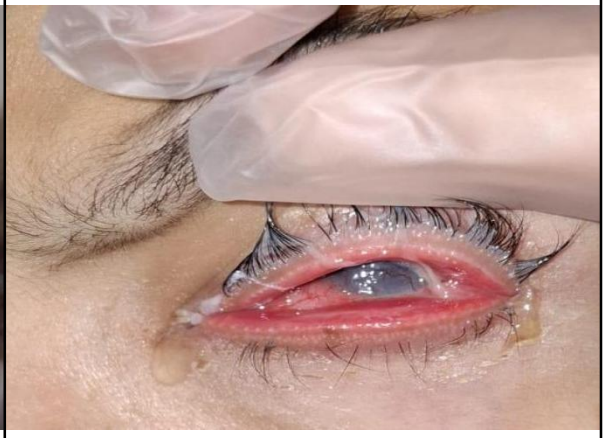
D: Left eye - fibrovascular conjunctival tissue encroaching onto the cornea

E: Right eye - additional view showing the severity and extent of cicatrizing ocular surface changes bilaterally



F: Left eye - additional views showing the severity and extent of cicatrizing ocular surface changes bilaterally





ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

G: Right eye - Additional view showing the severity and extent of cicatrizing ocular surface changes bilaterally	**H: Left eye - additional view showing the severity and extent of cicatrizing ocular surface changes bilaterally**

Figure 2. External ocular photographs demonstrating severe cicatrizing ocular surface disease. Composite images of the right eye (A, C, E, G) and left eye (B, D, F, H) show marked conjunctival inflammation with fibrovascular conjunctivalization extending onto the corneal surface, associated with corneal opacity/scarring and cicatricial ocular surface changes (suggestive of forniceal shortening/symblepharon).

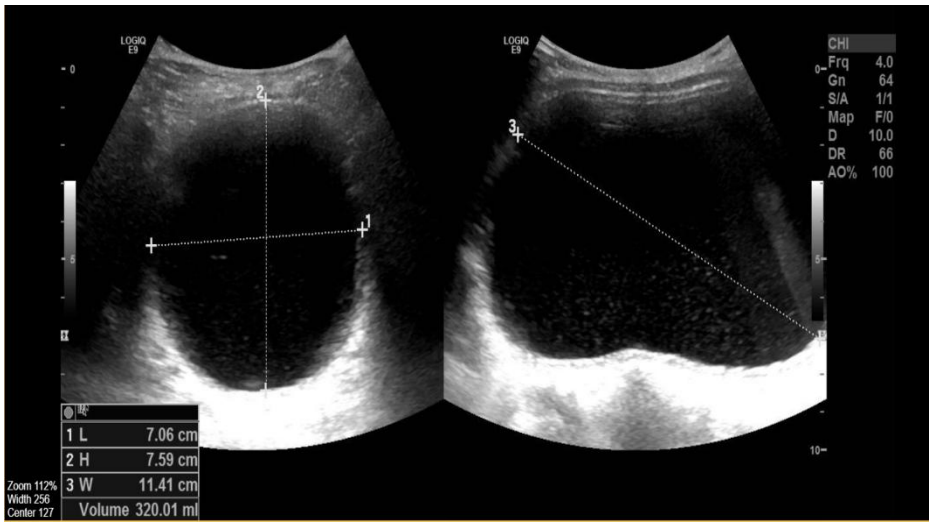


Figure 3: Urinary bladder ultrasound demonstrating cystitis and retention. Ultrasound performed in September 2021 for urinary retention/meatal stenosis shows a distended urinary bladder (volume ~320 mL; dimensions L 7.06 cm × H 7.59 cm × W 11.41 cm) with turbid intravesical fluid, consistent with cystitis in the context of recurrent obstructive uropathy (meatal/urethral stenosis) associated with congenital junctional epidermolysis bullosa.

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.132.145

Journal of Dermatological Case Reports

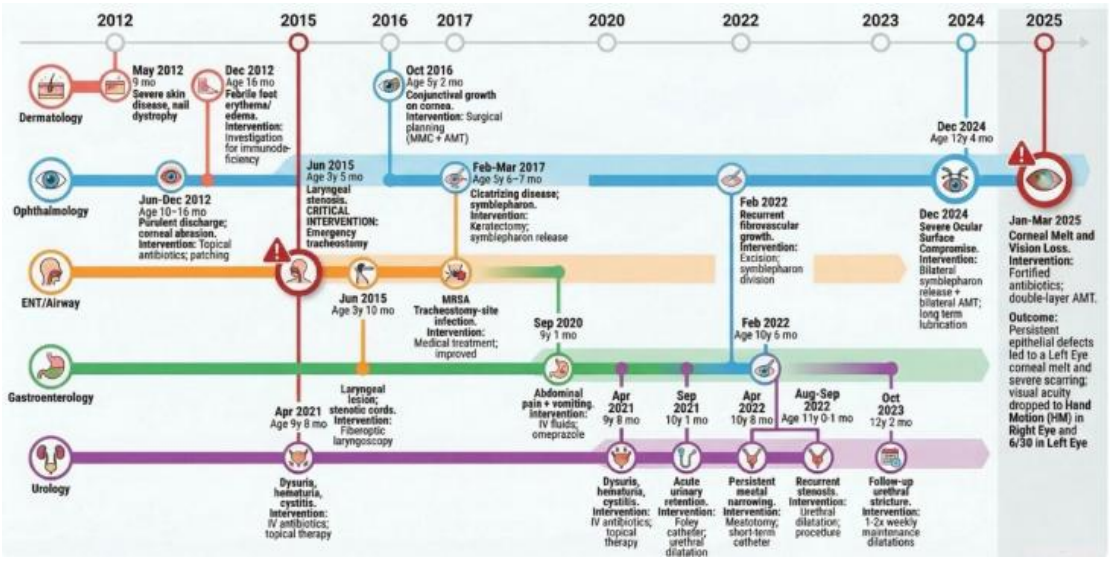


Figure 4: Multisystem timeline of major CJEB-related complications, admissions, and interventions. Chronological overview of key dermatologic, ophthalmologic, ENT/airway, gastrointestinal, and urological events from infancy through adolescence, highlighting early severe cutaneous disease, progression to laryngeal stenosis requiring emergency tracheostomy, recurrent cicatrizing ocular surface disease necessitating repeated surgical reconstruction (including AMT/MMC) with subsequent corneal melt and vision loss, and evolving obstructive uropathy with recurrent meatal/urethral stenosis requiring catheterization, dilatations, and meatotomy