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A novel bladder phenotype in junctional epidermolysis bullosa: a case report

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Background: Junctional epidermolysis bullosa (JEB) is a rare inherited blistering disorder, and its urological spectrum remains poorly defined.

Case Presentation: A 19-month-old boy carrying compound heterozygous *ITGB4* mutations (p.R252C, p.P305L) had a 17-month history of intermittent voiding. Ultrasound demonstrated focal papillomatous bladder-wall thickening, and cystoscopy showed scattered follicular mucosal changes without masses. Biopsies revealed mild oedema and chronic lymphocytic inflammation; no malignancy. Urine cultures were negative.

Conclusion: This case broadens the reported urological spectrum of *ITGB4*-related JEB by illustrating a papillomatous–follicular bladder phenotype. Early urological evaluation in patients with JEB presenting with unexplained urinary symptoms may facilitate timely, targeted management and help prevent chronic complications.

KEYWORDS

junctional epidermolysis bullosa, *ITGB4* mutation, bladder involvement, papillomatous bladder thickening, follicular mucosal changes, rare disease

Introduction

Epidermolysis bullosa (EB) comprises a group of inherited mechanobullous disorders with an overall incidence of $\approx 1: 50,000$ live births, of which junctional EB (JEB) accounts for 5%–10% (1, 2). According to the 2020 consensus reclassification (3), EB is subdivided into EB simplex, JEB, dystrophic EB and Kindler EB, each defined by the ultrastructural level of tissue cleavage and its causative genes. In addition to *ITGB4*-related JEB, bladder involvement has also been reported in JEB caused by laminin-332 gene variants (*LAMA3*, *LAMB3*, *LAMC2*), manifesting as urethral strictures, meatal stenosis or severe inflammatory cystitis (4, 5). JEB involves tissue separation at the lamina lucida within the basement membrane zone (6). It typically arises from mutations in genes encoding hemidesmosome components or associated integrins, compromising dermal-epidermal adhesion and causing widespread skin fragility, mucosal involvement, and multisystem complications (1).

The *ITGB4* gene, encoding the $\beta 4$ subunit of the $\alpha 6\beta 4$ integrin complex (a vital hemidesmosome component anchoring basal keratinocytes), is particularly significant (7). *ITGB4* mutations are classically linked to JEB with pyloric atresia (JEB-PA) (7), but phenotypic variability exists. *ITGB4* mutations also occur in patients with JEB without pyloric atresia or with milder/late-onset skin issues alongside significant extracutaneous involvement, including severe uropathy (8).

Urological complications occur in various EB forms, and patients with JEB are particularly susceptible. Documented issues include meatal (urethral) stenosis (up to

11.6% in JEB-Herlitz), urinary retention (9.3%), hydronephrosis (7.0%), bladder-wall hypertrophy (4.6%), and cystitis (9). The urothelium, like epidermis, relies on robust cell-matrix adhesion; $\alpha 6 \beta 4$ integrin disruptions can predispose it to damage (10). While skin and gastrointestinal JEB manifestations are well-documented, and general bladder pathologies (mucosal blistering, wall thickening, fibrosis, polypoid masses) are anecdotally noted in EB (4), specific bladder phenotypes linked to *ITGB4* mutations are largely uncharacterized.

We report a 19-month-old boy with junctional epidermolysis bullosa carrying compound-heterozygous *ITGB4* variants who fulfils the diagnostic criteria for JEB with pyloric atresia (JEB-PA); the pyloric atresia was surgically corrected in the neonatal period. Here we present the clinical, imaging and genetic findings of this case and discuss their implications. We discuss the potential pathophysiological relevance of these changes, their relationship to the underlying *ITGB4* deficiency, and how they further broaden the recognised genitourinary spectrum of *ITGB4*-related disease.

Case presentation

Patient history and clinical findings

Skin fragility had been mild and episodic since birth; consequently, a definitive diagnosis of epidermolysis bullosa was not established until comprehensive genetic testing at 19 months of age. A 19-month-old boy presented with a 17-month history of difficult, interrupted urine streams noted by caregivers, accompanied by occasional discomfort. Congenital pyloric atresia

was corrected surgically neonatally. At 2 months, he developed intermittent urinary difficulty with white flocculent material in his urine. A basic urinalysis at that age showed mild pyuria and hematuria; urine culture and crystal analysis were not performed. Antibiotics provided temporary relief, but episodes recurred. An ultrasound at 6 months revealed bilateral ureteral dilation; voiding cystourethrography showed no vesicoureteral reflux. Ureteral dilation resolved by 12 months. At 19 months, ultrasound revealed a focal bladder wall thickening (~0.7 cm) that protruded into the lumen, exhibiting a papillomatous appearance (Figure 1).

He is the son of nonconsanguineous, asymptomatic parents; his older sister is unaffected. Physical examination showed partial nail loss/thickening, occasional subungual blistering, and multiple oral mucosal ulcers, consistent with EB. Genital/external urinary exams were normal. Cardiorespiratory and abdominal examinations were unremarkable.

Laboratory tests

Urinalysis performed at 19 months revealed mild hematuria (red blood cells $306.5 \text{ cells } \mu\text{l}^{-1}$) and pyuria (white blood cells $12.6 \text{ cells } \mu\text{l}^{-1}$), with trace proteinuria; urine culture was negative.

Imaging

Bladder ultrasound showed localized wall thickening protruding into the lumen, with a papillomatous appearance (Figure 1). Color Doppler confirmed vascular flow.

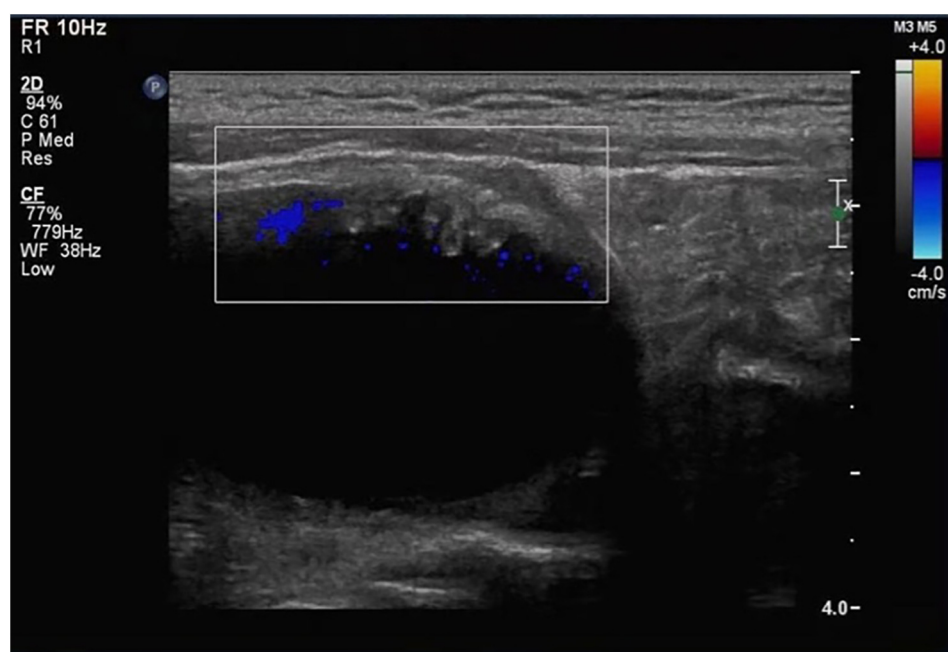


FIGURE 1

Ultrasound findings of the bladder wall. Localized papillomatous thickening of the bladder wall protruding into the bladder lumen, with visible vascular flow signal (color Doppler).

Cystoscopy and histopathology

Cystoscopy revealed scattered, raised lesions exhibiting follicular mucosal changes on the bladder mucosa. Surrounding mucosa appeared pale and edematous. No solid masses were identified (Supplementary Figure 1). These projections were distinct from typical diffuse inflammatory changes. Bladder dome biopsies showed mild edema and chronic lymphocytic inflammatory infiltrate with vascular congestion. The urothelium was largely intact and no malignant cells were identified (Figure 2). Immunohistochemistry: Cytokeratin (+), Vimentin (+), PAX-8 (+), PAX-2 (+), few CyclinD1 (+) cells, SMA (–), BCL-2 (–), Ki67 <1%. Findings were consistent with chronic mucosal inflammation and no neoplasia; viral immunostains for human papillomavirus (p16) and polyomavirus SV40 were not performed owing to limited tissue.

Genetic testing

Genetic analysis identified compound heterozygous ITGB4 mutations [c.754C>T (p.R252C) and c.914C>T (p.P305L)] (Supplementary Figure 2). Each mutation was inherited from an asymptomatic parent, consistent with autosomal recessive inheritance (Figures 3A,B; Supplementary Figures 3A,B). Bioinformatic tools (SIFT, PolyPhen-2, MutationTaster) suggested these variants were likely damaging. According to the American College of Medical Genetics and Genomics/Association for Molecular Pathology (ACMG/AMP) criteria, c.754C>T (p.Arg252Cys) meets evidence codes PVS1, PM1, PM2 and PP3 and is therefore classified as pathogenic, whereas c.914C>T

(p.Pro305Leu) meets PM2 and PP3 and is classified as likely pathogenic. Both variants were queried against multiple population and clinical databases. They are absent from gnomAD v3.1 and v4.0 (allele count = 0). c.754C>T (p.Arg252Cys) is recorded in ClinVar and HGMD as pathogenic, whereas c.914C>T (p.Pro305Leu) is not listed in either database. The complete absence of population frequency, together with existing clinical annotations, further supports their pathogenicity. In-silico pathogenicity was further assessed with SIFT v6.2 (J. Craig Venter Institute, Rockville, USA); PolyPhen-2 v2.2 (Harvard University, Cambridge, USA); and MutationTaster (Charité—Universitätsmedizin Berlin, Germany). Structural modelling suggested p.Arg252Cys might disrupt hydrogen bonds, destabilising the integrin $\alpha 6\beta 4$ complex, and p.Pro305Leu might affect protein flexibility.

Structural modeling suggested p.R252C might disrupt hydrogen bonds, destabilizing the integrin $\alpha 6\beta 4$ complex (Figures 4A,B), and p.P305L might affect protein flexibility (Figures 5A,B).

Discussion

This report describes a patient with JEB who developed a previously undescribed papillomatous–follicular bladder phenotype associated with compound heterozygous ITGB4 variants. These findings expand the current understanding of urological manifestations in JEB. Similar papillomatous changes, described as “papilla bullosa”, were reported in an 11-year-old girl (parental origin documented) with ITGB4-related JEB by Ilmiers et al. in 2016 (11). In addition, phenotypic heterogeneity associated with $\beta 4$ -integrin mutations has been highlighted and commented on by Bruckner-Tuderman in an editorial comparing two siblings with divergent JEB manifestations (12).

Contextualization with existing literature

Urological involvement in JEB is known. Previous reports focus on urethral strictures, hydronephrosis, urinary retention, general bladder wall hypertrophy, and cystitis (9). General bladder pathology in EB includes mucosal blistering, diffuse wall thickening, fibrosis, and occasional polypoid masses (4). A recent systematic review summarised 13 epidermolysis bullosa cases with vesicular or papillomatous cystitis, underscoring bladder mucosal vulnerability (13). The current case’s papillomatous protrusion and accompanying follicular mucosal changes are morphologically distinct. Whereas “wall thickening” is a non-specific descriptor, a papillomatous projection denotes an organised outgrowth rather than simple hypertrophy. The follicular mucosal changes likely represent small, lymphoid-aggregate-related lesions that differ from larger polypoid formations (4). To our knowledge, this combination has not previously been described in the JEB literature.

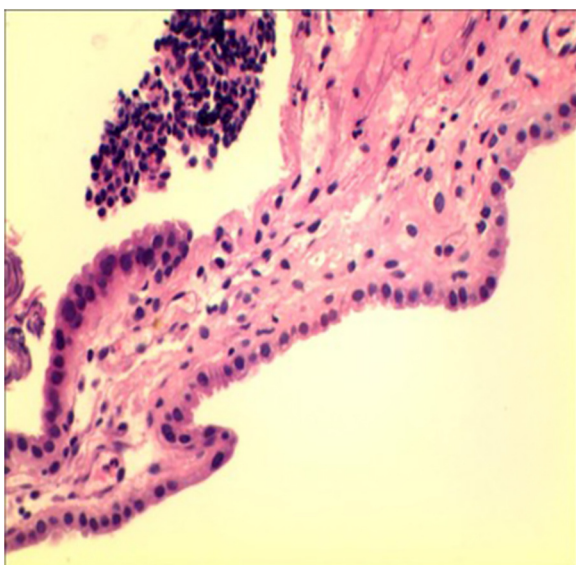


FIGURE 2
Histopathological examination of the bladder dome biopsy. Mild inflammation is observed, with edema and lymphocytic infiltration.

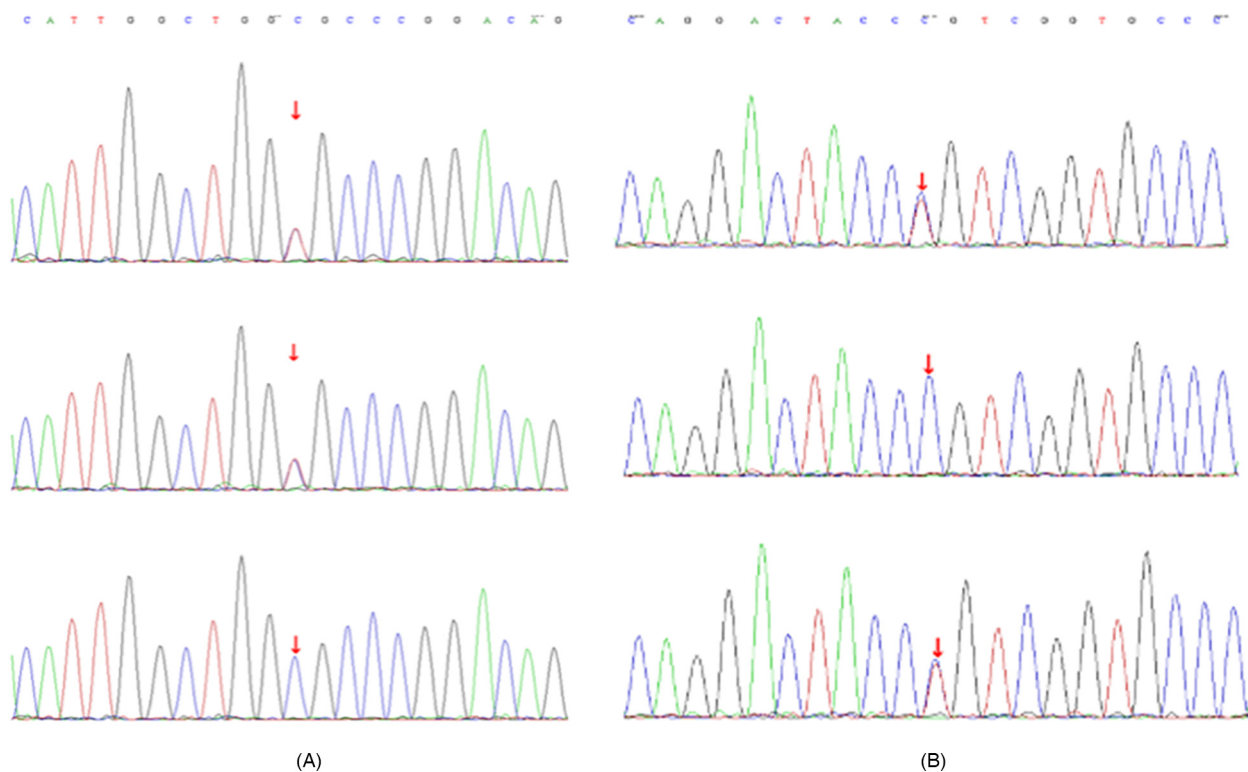


FIGURE 3
Sanger sequencing results of *ITGB4* mutations. (A) Sanger sequencing showing the *ITGB4* c.754C>T (p.R252C) mutation in the proband (top panel), father (middle panel), and mother (bottom panel). (B) Sanger sequencing showing the *ITGB4* c.914C>T (p.P305I) mutation in the proband (top panel), father (middle panel), and mother (bottom panel).

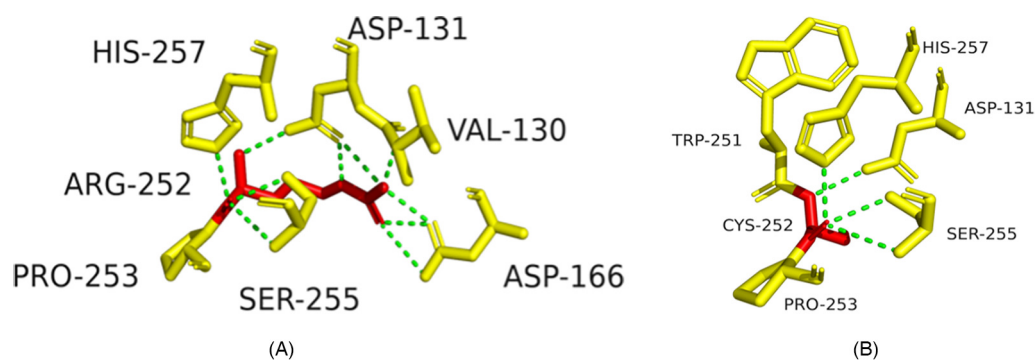


FIGURE 4
Structural impact of *ITGB4* c.754C>T (p.R252C) mutation. (A) Wild-type *ITGB4* structure showing key amino acid interactions. (B) Mutant *ITGB4* structure with c.754C>T substitution, showing loss of hydrogen bonds.

Significance of *ITGB4* mutations (p.R252c and p.P305I)

Compound-heterozygous *ITGB4* variants p.R252C and p.P305I are central to this case. *ITGB4* encodes the $\beta 4$ -integrin subunit, a key component of hemidesmosomes in epithelia such as the urothelium (6). Alterations at residue Arg252 are particularly noteworthy: Ellis et al. reported a patient with p.R252I who had

mild cutaneous JEB-PA but severe obstructive uropathy (8), and Dang et al. documented a lethal JEB-PA neonate with p.R252C (14). These observations suggest that residue R252 is critical and that its alteration predisposes carriers to urological complications. Similar *ITGB4*-PA cases without bladder involvement have been reported (15). Reitelman et al. described an *ITGB4*-mutant patient with severe ureteric obstruction but no bladder lesions, further illustrating phenotypic variability (16). However, the

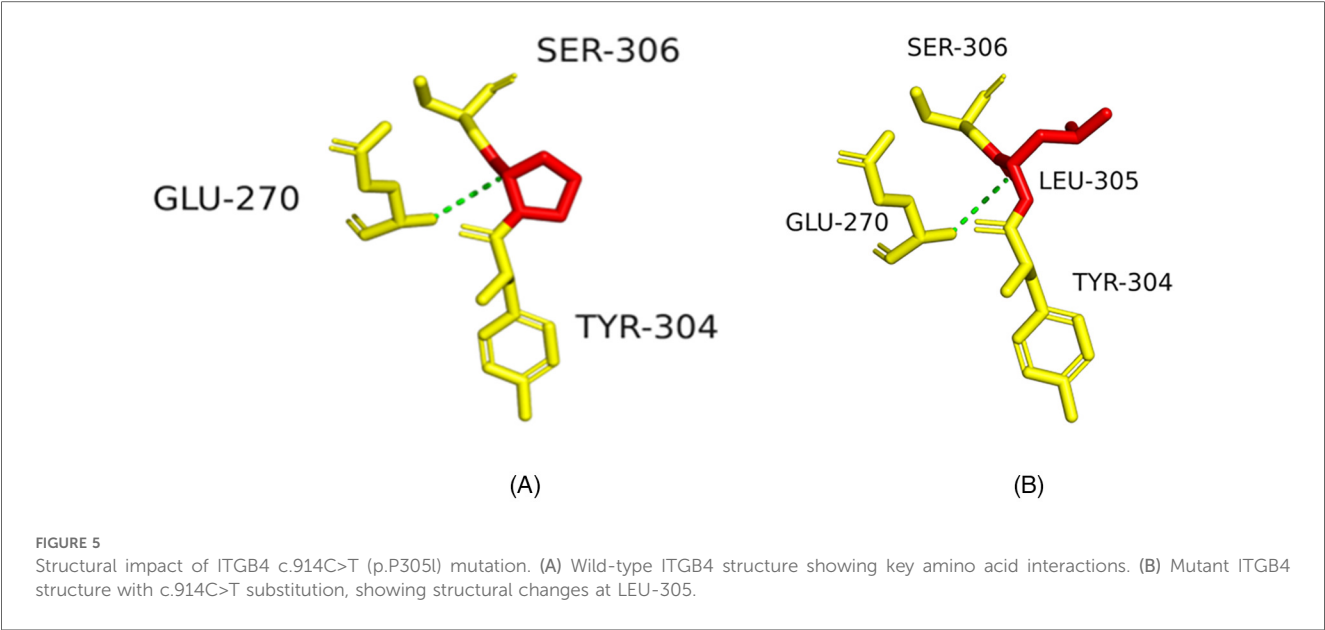


TABLE 1 Comparison of urological phenotypes in reported cases of JEB with ITGB4 mutations^a.

Reference (study/ authors, year)	Patient age/sex	ITGB4 mutations (specific variants)	Pyloric atresia (PA)	Key skin phenotype	Key urological/bladder findings	Outcome/ severity
Current case (present report)	19-month-old M	c.754C>T (p.R252C) & c.914C>T (p.P305I) (Comp Het)	Present (Corrected)	Nail dystrophy, oral ulcers, intermittent skin blisters	Papillomatous bladder wall thickening, follicular mucosal changes; mild hematuria/pyuria; chronic inflammation	Ongoing, managed
Ellis et al. (2021) (8)	6-year-old M	p.R252I & 3793+1G>A (Comp Het)	Present	Mild cutaneous EB (blistering resolved early)	Severe obstructive uropathy; bladder wall hemorrhage/blistering; bilateral ureteric reflux; CKD stage 2	Nonlethal, severe uropathy
Dang et al. (2008) (14)	Neonate (d. 4 m)	658delC (PTC) & p.R252C (Comp Het)	Present	Severe skin blistering, erosions	Not detailed for bladder morphology	Lethal
Mattioli et al. (2022) (10)	10-year-old M	c.320G>C (p.Arg107Pro) & c.542C>T (p.Pro181Leu) (Comp Het)	Absent	Minimal, late-onset skin fragility (from age 6)	Severe obstructive uropathy; thickened bladder wall; exuberant/diseptelialized bladder mucosa; hydroureteronephrosis	Severe uropathy, ileal neo-bladder
Reitelman et al. (2023) (16)	8- year-old F	c.1234A>G; c.2345dup	Absent	Mild skin fragility	Severe ureteric obstruction; no bladder lesion	Managed

^aRBC, red blood cells; WBC, white blood cells; pyuria, elevated WBC in urine.

specific “stalactite-like” bladder-wall thickening and “follicular” mucosal changes documented in our patient were not reported in those cases (8, 14). Ellis et al.’s patient had bladder-wall haemorrhage/blistering (8, 10) reported exuberant, de-epithelialised bladder mucosa in a child carrying different ITGB4 variants without pyloric atresia. This highlights that although R252 mutations are linked to uropathy, our patient’s bladder manifestation is distinct. The p.P305I variant, far less characterised, was bio-informatically predicted as damaging and may further impair α6β4-integrin function. Although pyloric atresia defines the JEB-PA subtype, bladder involvement has been documented only sporadically. The coexistence of surgically corrected PA and a papillomatous–follicular bladder phenotype in our patient broadens the recognised genitourinary spectrum of

ITGB4-related disease and underscores the need for systematic urological surveillance in JEB-PA.

Table 1 compares urological phenotypes.

This comparison shows that while ITGB4 mutations (especially at R252) link to severe uropathy, our patient’s specific bladder phenotypes appear novel.

Mechanistic insights

The novel phenotypes most plausibly arise from impaired urothelial integrity caused by dysfunctional α6β4 integrin. The ITGB4 variants (p.R252C, p.P305I) are predicted to destabilise the integrin complex, rendering the bladder mucosa vulnerable to

mechanical stress, chronic inflammation and recurrent sub-clinical injury. Repeated insults may induce dysregulated repair, leading to chronic oedema, inflammatory infiltration and aberrant remodelling. The observed papillomatous projections could represent focal fibromuscular hyperplasia or organised granulation tissue, whereas the follicular mucosal changes suggest reactive lymphoid hyperplasia. Although urine cultures were negative, occult low-grade colonisation or sterile inflammation cannot be excluded. The widespread expression of $\alpha 6\beta 4$ integrin in urogenital epithelium offers a biologically plausible link between integrin dysfunction, mucosal fragility and these secondary alterations (4).

Clinical implications

Recognizing these novel bladder phenotypes in patients with JEB, especially with *ITGB4* mutations, may aid earlier diagnosis and management. This case reinforces recommendations for systematic urological evaluation (ultrasound, possibly cystoscopy) in patients with JEB with urinary symptoms or unexplained bladder anomalies (13, 17). Early identification could allow timely interventions to mitigate chronic irritation and prevent progressive dysfunction. A multidisciplinary approach is paramount.

Future directions

1. Verification of Phenotypic Specificity

Cohort studies or case series are needed to determine if these bladder changes are specific to certain JEB genotypes (e.g., *ITGB4* p.R252C) or broader manifestations.

2. Molecular Mechanism Studies

Investigating how *ITGB4* mutations lead to these specific bladder phenotypes using patient-derived cells or organoids could identify dysregulated pathways or biomarkers.

3. Exploration of Gene Therapy

Advances in gene therapy for EB, such as Beremagene perpavec (B-VEC) for dystrophic EB and ex vivo LAMB3 correction for JEB (18), provide a rationale for developing *ITGB4*-targeting strategies, in line with recent insights into *ITGB4* genetics and management (19). These approaches could potentially mitigate both cutaneous and systemic complications, including the bladder phenotype described here.

Limitations

This single case report's findings may not be generalizable. Long-term follow-up and additional cases are needed to determine the specificity, prevalence, and clinical relevance of these bladder changes in JEB.

Conclusion

This report describes a novel bladder phenotype in a patient with junctional epidermolysis bullosa (JEB) carrying compound heterozygous *ITGB4* mutations, characterized by focal papillomatous bladder wall thickening and follicular mucosal changes. These findings broaden the recognized phenotypic spectrum of JEB and emphasize the importance of comprehensive urological assessment in patients presenting with urinary symptoms or unexplained bladder abnormalities. Early recognition and targeted intervention may help prevent chronic urological complications. Further investigation into the underlying molecular mechanisms may inform the development of future therapeutic strategies.

Data availability statement

The data underlying this article are available in the article itself; further information can be obtained from the corresponding author on reasonable request.

Ethics statement

The studies involving humans were approved by Ethics Committee of Children's Hospital Affiliated to Shandong University (Jinan Children's Hospital). The studies were conducted in accordance with the local legislation and institutional requirements. Written informed consent for participation in this study was provided by the participants' legal guardians/next of kin. Written informed consent was obtained from the minor(s)' legal guardian/next of kin for the publication of any potentially identifiable images or data included in this article.

Author contributions

QH: Conceptualization, Investigation, Methodology, Writing – original draft, Writing – review & editing. MG: Conceptualization, Writing – review & editing. HW: Investigation, Writing – original draft. LZ: Conceptualization, Data curation, Supervision, Writing – original draft, Writing – review & editing.

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Conflict of interest

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Supplementary material

The Supplementary Material for this article can be found online at: <https://www.frontiersin.org/articles/10.3389/fped.2025.1555599/full#supplementary-material>

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