Case Report

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Genitourinary complications as initial presentation of inherited epidermolysis bullosa

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ABSTRACT

Epidermolysis bullosa (EB) is a rare disorder that presents with urological complications. We present a 6-year-old boy admitted with urological symptoms that revealed an inherited EB misdiagnosed. We also review the literature on this disorder and management of the common urological complications.

Key words: Epidermolysis bullosa, skin, urogenital tract

INTRODUCTION

Inherited epidermolysis bullosa (EB) is a rare dermatological disorder, with an incidence of 20 per million live births.[1,2] It is one of the most devastating chronic diseases always misdiagnosed. Three major groups of diseases, namely EB simplex (EBS), junctional EB (JEB), dystrophic EB (DEB), and 17 subtypes are known to exist. Diagnosis can be made on the basis of ultrastructural and immunohistochemical studies. EBS causes lesions at the basal layer, JEB causes breakage at the level of the basement membrane, and the third type, DEB, causes division at the level of the dermis. It is characterised by marked fragility of epithelial tissues occurring not only on the skin, but also on eyes, trachea, mouth, gastrointestinal and genitourinary tract. Patients presenting with urological complications have been reported rarely in the literature.

We report a case of inherited EB revealed by urological problems. We also review different urological procedures reported in this disease.

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OBSERVATION

A 6-year-old boy presented with 1 month history of voiding difficulty and urinary retention which required repeated catheterisations. Physical examination revealed meatal stenosis with peri-meatal blistering [Figure 1] and difficult voiding with a splayed stream. Cystoscopy showed bullous changes throughout the bladder, with bladder oedema. The child underwent a radiological evaluation, including sonography and voiding cystourethrography which revealed an irregular bladder wall thickening, with urethral stricture interpreted as sphincter hypertonia [Figure 2]. The patient was treated for neurogenic bladder with oxybutynin and clean intermittent catheterisation was done using topical steroids, despite normal results of medullar magnetic resonance imaging and urodynamic evaluation.

During the next week, blisters occluded completely the distal urethra resulting in urinary retention and child presented erosion on the mouth, face, trunk, pelvis, legs, feet [Figures 3 and 4]. Cystoscopy showed urethral stenosis. The patient underwent biopsy of skin lesion and the diagnosis of JEB was made on the basis



Figure 1: Meatal stenosis and peri-meatal blistering

of ultrastructural and immunohistochemical studies. A definitive continent cutaneous urinary diversion was used with a trans-appendicular tube (Mitrofanoff) [Figure 5]. Bladder biopsy was done, and it showed a thickened, fibrotic bladder wall with extensive granuloma formation.

DISCUSSION

The first case report of genitourinary tract involvement in EB was published in 1973 by Kretkowski.[3] He reported a 3-year-old boy with recessive DEB (RDEB), who presented during the second year of life, with pain upon voiding, urethral meatal stenosis, ulcerations of the glans, with distension bladder and hydronephrosis. Since then, such cases have been rarely reported.

The most common modes of presentation of urological disorder include meatal stenosis, dysuria, sepsis and haematuria. In patients with EB who have genitourinary tract involvement, urethra, penis, female genitalia, bladder, ureters and kidney can be affected [Appendix 1].[1,2,4] Most genitourinary complications reported have involved JEB patients associated with congenital pyloric atresia (JEB-pyloric atresia syndrome).[5] The urinary tract may be involved in any subtype of inherited EB, although these complications usually arise in patients with the most severe types, namely JEB and RDEB. Precise frequencies of urological complications have been elucidated only in a study published by the National Epidermolysis Bullosa of the National Institutes of Health throughout the continental United States, which reported 16.6, 30.2, 19.5 and 31.1% of patients with EBS, JEB, DDEB and RDEB, respectively.[1]

In our review of published reports, we found no reported cases of inherited EB, revealed initially by genitourinary complications as in our observation. EB was misdiagnosed and it was revealed by urological symptoms. Skin lesions appeared later and might be triggered by oxybutinin known agent that causes



Figure 2: Cystography showing irregular bladder wall thickening with urethral stricture



Figure 3: Skin lesions



Figure 4: Enamel hypoplasia and lesions on the face and mouth



Figure 5: Catheterizable stoma, skin lesion, keloid and hypertrophic scarring

mucosal and skin dryness as side effects. Furthermore, we detected a generalised enamel hypoplasia [Figure 4]. This allowed us to rectify the diagnosis.

Blisters may arise within the urethral orifice, resulting in painful urination. If erosions occluded the distal urethra completely, urinary retention occurs. Urethral strictures may result if blisters become more chronic. Complete and permanent stenosis of urethra occurs following repeated catheterisations and the use of invasive procedures to the diagnosis. This was the case in our patient.

Bladder involvement in EB is characterised by bladder oedema and thickened bladder wall; bladder may become fibrotic and scarred as a result of mechanical disruption of urothelium and chronic inflammation that was confirmed by an ultrastructural study done in our case.

In reported cases, the cause of hydroureteronephrosis is ureterovesical junction stenosis due to scarring at this site. Hydronephrosis is secondary to ureteral strictures. Only a few cases of cystitis and pyelonephritis complicating inherited epidermolysis bullosa have been reported in the litterature.[1,6,7]

Numerous urological surgical procedures have been reported in inherited EB [Appendix 2], like urethral meatal dilatation with urethral catheterisation and meatotomy for urethral meatal stenosis or stricture (penile and bulbar). Many procedures for urinary diversion have been performed such as ureterostomy, vesicostomy, cystostomy, ureterosigmoidostomy and continent cutaneous urinary diversion.[1,8] Because of metabolic complications, ureterosigmoidostomy has led to its abandonment for the other types of procedures.

Appendix 1: Urological complications in EB

- Scaring of glans penis
- Fusion of the labia
- Urethral stricture (penile and bulbar)
- Urethral diverticuli
- Bladder hypertrophy
- Bladder oedema
- Cystitis
- Bladder extrophy[9]
- Ureteral stenosis
- Obstruction of ureter-vesicular junction
- Hydroureter
- Hydronephrosis
- Pyelonephritis
- Renal insufficiency[10]

Appendix 2: Urological procedures

- Urethral catheterisation
- Meatal dilatation
- Meatotomy
- Cystostomy
- Vesicostomy
- Ureterostomy
- Ureterosigmoidostomy
- Continent cutaneous urinary diversion

CONCLUSION

The kidney, ureters, bladder and lower genitourinary tract may become involved in inherited EB. In any patient with signs or symptoms of genitourinary tract disease associated with skin lesions or signs of skin fragility, EB should be suspected and when diagnosis is confirmed, the patient should be immediately referred for urological evaluation and treatment because if untreated renal deterioration may result.

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