Congenital anterior urethrocutaneous fistula at the penoscrotal junction with proximal penile megalourethra: A case report

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ABSTRACT

Congenital anterior urethrocutaneous fistula and megalourethra are both rare anomalies. These anomalies are commonly associated with other anorectal or genitourinary anomalies and evaluated with voiding cystourethrography. We examined a 34-month-old boy who presented with a fistula at the penoscrotal junction. A voiding cystourethrogram showed a jet of urine coming through the fistula and proximal saccular dilatation of the penile urethra. We present the imaging findings of the first case of an association between a congenital anterior urethrocutaneous fistula at the penoscrotal junction and a proximal penile megalourethra. We also discuss the etiology, management, and differential diagnosis of this entity, and review the literature.

CASE REPORT

CASE REPORT

A 34-month-old boy presented with leakage of urine from the penoscrotal junction. His parents noticed it shortly before approaching a doctor. There was no history of trauma or surgical intervention. Physical examination of the patient showed a tiny hole at the penoscrotal junction with urine leakage (Fig. 1). The preputial skin was intact with a normally placed external urethral orifice and absent chordee. Both testes were located in the scrotum. A voiding cystourethrogram performed with a 3.5 Fr urethral catheter inserted smoothly through the external urethral orifice, showed a jet of urine coming through the fistula at the penoscrotal junction with proximal saccular dilatation of the penile urethra (Fig. 2). The urine passed mostly through the fistula but also through the external urethral orifice, which might be related to the catheter in the distal urethra at micturition. A congenital anterior urethrocutaneous fistula with proximal penile megalourethra was diagnosed. The patient was then referred to a specialist for an optimal surgical correction.

DISCUSSION

Etiology & Demographics:

A congenital anterior urethrocutaneous fistula of the male urethra is a rare anomaly. Only 47 cases have been reported in the literature to date, out of which the fistula was at the distal shaft in 24 patients; at the mid-shaft in 13; at the penoscrotal junction in 3; and at an unspecified location in 7 [1, 2]. It may be associated with other anomalies of the genitourinary tract, such as deficient distal urethra, distal hypospadias, chordee, anorectal malformations, stenosed bulbar urethra, epispadic urethral duplication, and megalourethra, or it may develop in an isolated fashion [1, 2].The etiology of a congenital anterior urethrocutaneous fistula is not clear. A coronal fistula may be

explained by misalignment of the glandular and penile urethra [1]. In this respect, Campbell stated that congenital urethrocutaneous fistulas represent embryonic urethral blowouts behind a distal congenital obstruction [3]. Olbourne suggested that the focal defect in the urethral plate results in arrested distal migration of the urethral plate or a localized deficiency of a portion of the plate [4]. Cook and Stephens suggested that pressure atrophy from the heel of the baby's foot was a potential cause [5].

Megalourethra is a rare congenital anomaly resulting from the faulty development of the corpora cavernosa and corpus spongiosum. The scaphoid type, as in our case, is caused by poor development of the corpus spongiosum. In the more severe fusiform type, the corpora cavernosa is also affected [6]. Megalourethra is often associated with anomalies of the genitourinary, gastrointestinal, or other systems, such as VACTERL association, duplication of the urethra, renal dysplasia, megacystis, polycystic kidney, brachydactylia, unilateral renal hypoplasia, colonic malrotation, hydronephrosis, hydroureteronephrosis, duplication of the ureter, cryptorchidism, vesicocutaneous fistula, and Prune-belly syndrome [7].

The association between congenital anterior urethrocutaneous fistula and proximal megalourethra might be owing to prenatal insults that result in focal defects of the urethral plate and adjacent urethral folds, leading to the appearance of a fistula at the anterior portion of the megalourethra (Fig. 3).

Clinical & Imaging findings:

Congenital anterior urethrocutaneous fistulas have been described mostly in the pediatric age group. Clinically, a fistula with urine leakage is present despite a normal urethral meatus. History is also important for establishing a diagnosis, since any trauma or surgery could result in fistula formation. Only 1 case of a fistula below the coronal sulcus with distal penile megalourethra proximal to the fistula has been reported without image correlation [1]. Penile swelling during urination is noted in patients with megalourethra.

Voiding cystourethrography is an important tool for evaluating congenital anomalies of male urethras. Congenital anterior urethrocutaneous fistulas demonstrate a contrast-filling fistula between the anterior urethra and skin on a voiding cystourethrogram. The dilated urethra of a scaphoid or fusiform megalourethra is clear on a voiding cystourethrogram without definite stenosis or obstruction [8]. During the prenatal ultrasonography, fetuses with megalourethra present with a cystic structure in the perineal region and a distended bladder with or without hydroureter and hydronephrosis or any other associated anomalies [9].

Treatment & Prognosis:

Surgical repair is the main treatment. For isolated cases, the fistula can be circumscribed and then closed using a simple multilayer fistula closure [1]. For more complex cases, closure requires the more complex reconstruction techniques employed in formal hypospadias surgery. Primary repair was successful in 42 out of the 47 cases examined, but 4 patients required 2 operations and 1 patient required 3 closures [1]. Treatment of megalourethra follows the principles of surgery for hypospadias. For the fusiform type, the placement of a penile prosthesis in adulthood could be considered [7]. The prognosis depends on the severity of associated anomalies.

Differential Diagnoses:

Hypospadias

Hypospadias refers to an abnormally positioned urethral meatus, which can be placed proximally and ventrally to its normal position. It is thought to be an abnormality in the normal hormonal signaling pathway, which causes interruption of urethral migration, and thus a ventral location of the urethra [10]. Despite the abnormal location of the urethral meatus, retrograde urethrography or voiding cystourethrography may also reveal meatal stenosis, vesicoureteral reflux, prostatic utricle, or rudimentary vagina [11]. Generally, the more severe the hypospadias, the larger and more distal the utricle [10].

Urethral duplication

Urethral duplication is a rare anomaly with varied clinical manifestations such as a deformed penis, twin streams, urinary tract infection, urinary incontinence, outflow obstruction, and associated anomalies. A dorsally opening accessory urethra is the most common type. Embryogenesis of urethral duplication is not clear. Effmann et al. classified urethral duplication into 3 types [12]. Type 1 is incomplete urethral duplication. Type 2 is complete urethral duplication, and is further subclassified as 2A for 2 meatuses and 2B for 1 meatus. Type 3 is urethral duplication from duplicated or septated bladders. Voiding cystourethrography may demonstrate both the urethral channels, if the duplication is complete. Retrograde urethrography may be used to determine the length of the defect in incomplete duplication [13].

Anterior urethral valve

An anterior urethral valve is a rare congenital anomaly that results in lower urinary tract obstruction in children. The embryologic development of the anterior urethral valve remains unclear. Forty percent of the anterior urethral valves are located in the bulbar urethra, 30% at the penoscrotal junction, and 30% in the pendulous urethra [14]. Voiding cystourethrography shows the dilated urethra proximal to the valve [13]. A valve may appear as a linear filling defect along the ventral wall, or it may be indicated by a dilated urethra ending in a smooth bulge or an abrupt change in the caliber of the dilated urethra [15]. Voiding cystourethrography may also reveal associated anomalies such as the vesicoureteral reflux [15].

Anterior urethral diverticulum

Anterior urethral diverticulum is the second most common cause of congenital urethral obstruction in boys [16]. A diverticulum develops on the ventral surface of the penile urethra as a result of either focally incomplete development of the corpus spongiosum or incomplete fusion of a segment of the urethral plate [17]. Most children are diagnosed in infancy with dribbling-type micturition or infection [13]. On voiding cystourethrography, the saccular diverticulum of the anterior urethra fills with contrast material and appears as an oval structure on the ventral aspect of the anterior urethra [18].

TEACHING POINT

A congenital anterior urethrocutaneous fistula is a rare disease, which can arise without any trauma or surgical history, wherein a voiding cystourethrogram can show a fistula between the anterior urethra and skin. Megalourethra results from the faulty development of the corpora cavernosa and corpus spongiosum and is rarely associated with a congenital anterior urethrocutaneous fistula.

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FIGURES

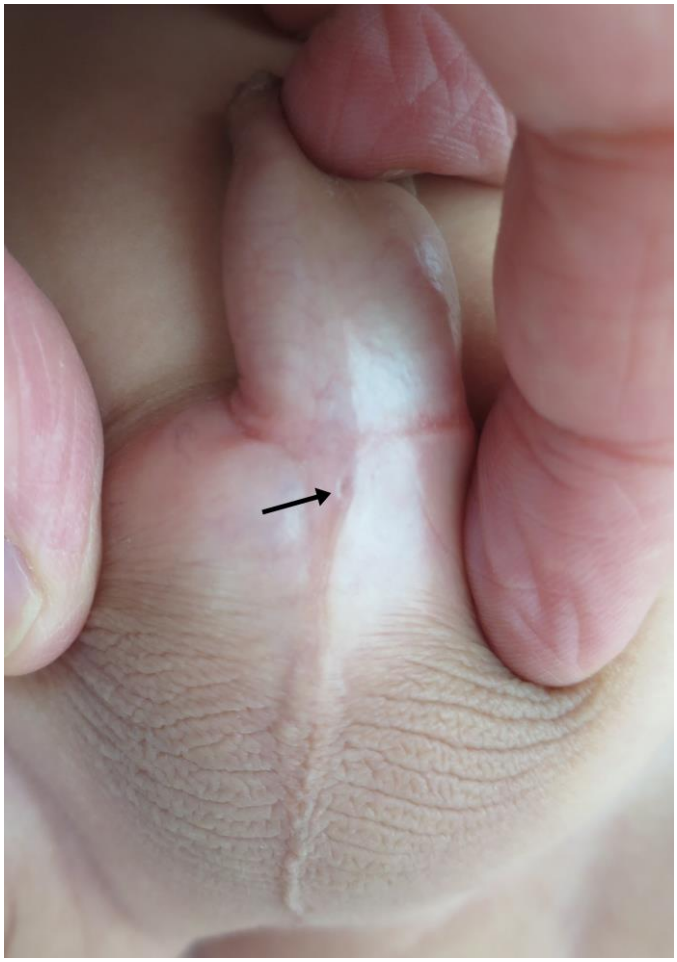

Figure 1: A photograph of a congenital anterior urethrocutaneous fistula in a 34-month-old boy with a tiny hole at the penoscrotal junction (arrow).

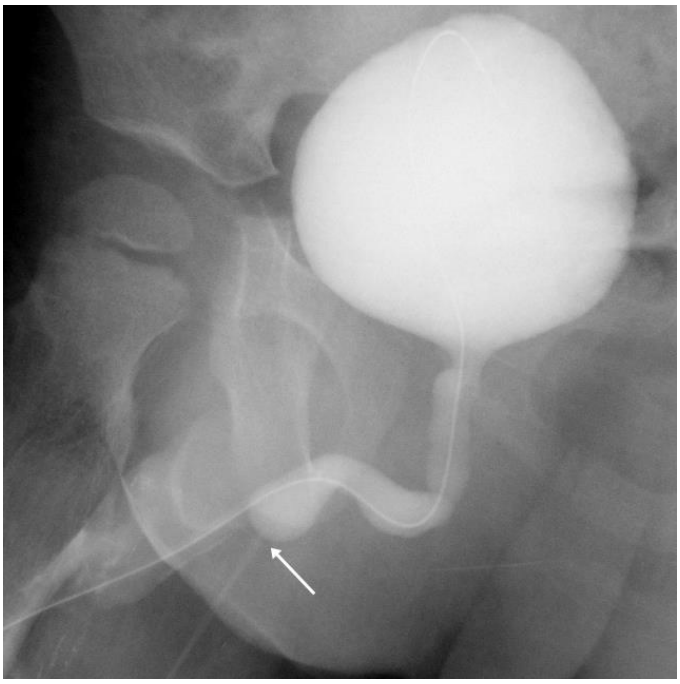

Figure 2: A 34-month-old boy with a congenital anterior urethrocutaneous fistula at the penoscrotal junction. FINDINGS: A voiding cystourethrogram shows a jet of urine (arrow) coming through the fistula with proximal saccular dilatation of the penile urethra. TECHNIQUE: Voiding cystourethrography using diluted Urografin (Urografin® 76%, 370 mg I/mL, 1:5 dilution with normal saline) instilled via a 3.5 Fr urethral catheter inserted smoothly through the external urethral orifice at a right posterior oblique position under fluoroscopy (HITACHI DR1000X, 55 kV, 200 mA).

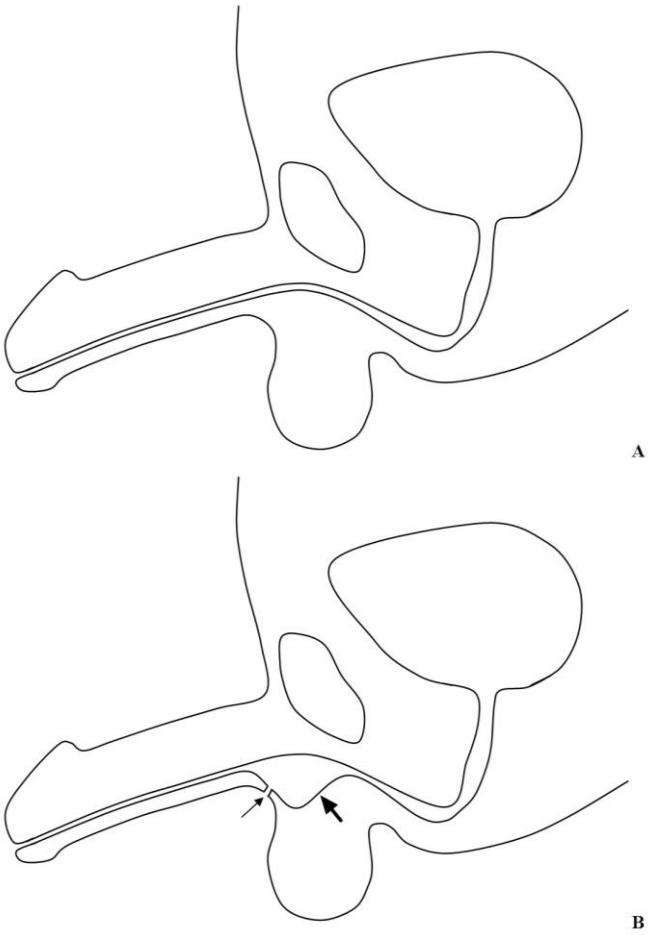

Figure 3: A diagram showing urethral abnormalities. (A) Normal anatomy of the urethra in males. (B) Congenital anterior urethrocutaneous fistula (thin arrow) at the penoscrotal junction with proximal penile megalourethra (thick arrow).

	Congenital anterior urethrocutaneous fistula	**Megalourethra**
Etiology	Unknown	Maldevelopment of the corpora cavernosa and corpus spongiosum
Incidence	Only a few case reports	Only a few case reports
Gender ratio	Unknown, male predominance	Unknown
Age predilection	Congenital	Congenital
Risk factors	Unknown	Unknown
Associated anomalies	Deficient distal urethra, distal hypospadias, chordee, anorectal malformations, stenosed bulbar urethra, epispadic urethral duplication, or megalourethra	VACTERL association, duplication of the urethra, renal dysplasia, megacystis, polycystic kidney, brachydactylia, unilateral renal hypoplasia, colonic malrotation, hydronephrosis, hydroureteronephrosis, duplication of the ureter, cryptorchidism, vesicocutaneous fistula, and Prune-belly syndrome
Treatment	Simple fistulectomy or techniques employed in formal hypospadias surgery	Surgery for hypospadias and/or placement of penile prosthesis in adulthood
Prognosis	Good	Depends on the severity of associated anomalies
Findings on imaging	VCUG shows a contrast-filling fistula between the anterior urethra and skin	VCUG shows scaphoid or fusiform dilatation of the urethra without definite stenosis or obstruction Prenatal ultrasound shows a cystic structure in the perineal region and a distended bladder with or without hydroureter and hydronephrosis

Table 1: Summary table of congenital anterior urethrocutaneous fistula and megalourethra cases.

	Clinical	VCUG or RUG
Congenital anterior urethrocutaneous fistula	A fistula with urine leakage is present despite a normal urethral meatus	A contrast-filling fistula between the anterior urethra and skin
Megalourethra	Ballooning of the penis during micturition and post-void dribbling with redundant skin on the ventral surface	Scaphoid or fusiform dilatation of the urethra without definite stenosis or obstruction
Hypospadias	Abnormally positioned urethral meatus, proximally and ventrally placed compared to its normal position	Meatal stenosis, vesicoureteral reflux, prostatic utricle, or rudimentary vagina in addition to an abnormally positioned meatus
Urethral duplication	Deformed penis, twin streams, urinary tract infection, urinary incontinence, outflow obstruction, and associated anomalies	VCUG may show the two urethral channels if the duplication is complete. RUG may be used to determine the length of the defect in incomplete duplication
Anterior urethral valve	Urinary tract infection, dribbling, hydronephrosis, and abnormal renal function	VCUG shows the dilated urethra proximal to the valve and possible associated vesicoureteral reflux
Anterior urethral diverticulum	Dribbling-type micturition or urinary tract infection	Saccular diverticulum appears as an oval structure on the ventral aspect of the anterior urethra

Table 2: Differential diagnosis table for congenital anterior urethrocutaneous fistula and megalourethra.

ABBREVIATIONS

RUG: retrograde urethrography
VACTERL: vertebral, anal, cardiac, tracheal, esophageal, renal, and limb anomalies
VCUG: voiding cystourethrogram

KEYWORDS

congenital; fistula; hypospadias; megalourethra; penis; urethra; urethrocutaneous; voiding cystourethrogram; megalourethra; urethrocutaneous fistula; penoscrotal junction