Exstrophy variant in a female child

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DESCRIPTION

Bladder exstrophy–epispadias complex is a spectrum of rare congenital malformations involving the urinary, genital and musculoskeletal systems. The condition can be subdivided into classic/typical forms, comprising epispadias, classical bladder and cloacal exstrophy, and variant/atypical forms, including duplicated, covered and pseudoexstrophy. Diagnosis can be made clinically. Incidence varies. However, the occurrence is higher in males. Genetic and environmental components likely contribute to the aetiopathogenesis, which remains unknown.^{1 2} There is a hypothesis that cloacal exstrophy variant could result from an ischiopagus conjoined twinning with blighting of one twin.³

We present a 9-year-old girl with urinary incontinence. She was initially referred at 2-1/2 years for ambiguous genitalia but did not continue follow-up. The mother denied receiving prenatal care or completing postnatal studies. On examination, she has a bifid clitoris, symphysis pubis widely separated and low-placed umbilicus with midline muscle defect where the bladder seemed to herniate (figure 1). Genetic studies reported normal female karyotype and Fluorescence in situ hybridization (FISH) result, and negative FISH result for sex-determining region Y (SRY). Testosterone level and renal ultrasound were unremarkable. Kidney, ureter, and bladder X-ray (KUB) demonstrated widening of the pubic symphysis

Figure 2 Widening of the pubic symphysis.

(figure 2). Voiding cystourethrogram (VCUG) ruled out ureterocele and vesicoureteral reflux and showed foreshortened urethra. Dynamic urography and magnetic resonance imaging (MRI) were pending, anticipating transfer of care to another centre for surgical management.

Treatment focuses on securing abdominal wall closure, achieving urinary continence with renal function preservation and obtaining adequate cosmetic and functional reconstruction. Pelvic osteotomies are often used to approximate the pubic diastasis, with postoperative immobilisation to facilitate a tension-free surgical repair.^{1 2} However, management is challenging, and spontaneous voiding cannot be guaranteed.

Figure 1 Bifid clitoris and umbilicus located below the horizontal line of the iliac crest.

Learning points

- ▶ Bladder exstrophy–epispadias complex is a spectrum of challenging congenital malformations, ranging from classic or typical forms to variant or atypical forms.
- ▶ Widening of the pubic symphysis is a feature consistent with the spectrum of closed bladder exstrophy–epispadias complex.
- ▶ Comprehensive prenatal care, including detailed ultrasound evaluation during pregnancy, could be key in determining the exact aetiology of exstrophy cases and its variants.

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REFERENCES

- 1 Baradaran N, Gearhart JP. Bladder Exstrophy-Epispadias-Cloacal exstrophy complex: a Contemporary Overview. *Neoreviews* 2010;11:e705–e713.
- 2 Ebert AK, Reutter H, Ludwig M, et al. The exstrophy-epispadias complex. *Orphanet J Rare Dis* 2009;4:23.
- 3 Casale P, Grady RW, Waldhausen JH, et al. Cloacal exstrophy variants. can blighted conjoined twinning play a role? *J Urol* 2004;172:1103–7.

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