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Bladder Exstrophy in a Female Neonate: A Case Report

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Abstract

Bladder exstrophy (BE) is a rare birth defect characterised by a spectrum of anomalies involving the ventral body wall, urinary tract, genitalia, bony pelvis, spine and anus. BE is almost exclusive to males and a part of the spectrum of Exstrophy-Epispadias complex that ranges in severity from epispadias to classical bladder exstrophy and cloacal exstrophy being the most severe. We report and discuss the case of a term female newborn, in whom clinical examination revealed genitourinary defect consistent with the classic bladder exstrophy variant of the exstrophy–epispadias complex. The defect was covered with sterile silicon gauzes and transparent waterproof dressing. Baby was then referred to paediatric surgery department for further management.

Keywords: Bladder exstrophy; female; neonate; congenital defect

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Introduction

Bladder exstrophy (BE) is a rare birth defect characterized by a spectrum of anomalies involving the ventral body wall, urinary tract, genitalia, bony pelvis, spine and anus (Ludwig M et al. 2009). BE is part of the spectrum of Exstrophy-Epispadias complex that ranges in severity from epispadias to classical bladder exstrophy and cloacal exstrophy being the most severe (Ebert AK et al. 2009). The estimated prevalence of classical bladder exstrophy is approximately 3.3 per 100,000 live births (Jayachandran D et al. 2011). The disease is more common in boys as compared to girls with a male to female ratio of 6:1 (Ebert AK et al. 2009). It is also less prevalent among the non-white race, high or low socioeconomic status, and Western geographic region (Nelson CP et al. 2005). Maternal smoking, exposure to radiation and valproate especially in the first trimester, has been associated with increased risk of disease occurrence, whilst periconceptional folate intake has been reported to be protective (Reutter H et al. 2011). We report a case of female neonate with bladder exstrophy because of its rarity and term birth.

Case presentation

This is the case of a female neonate of non-consanguineous parents; that was delivered via caesarean section at 37 weeks gestation and had good extra-uterine transition with a birth weight of 2,870 grams. Her mother was regular with her antenatal clinic visits, received only prescribed medications and had unremarkable laboratory results. She reported no exposure to ionizing radiation, no history of smoking and had no known chronic pathology. The neonate presented at birth with a genitourinary defect consistent with the classic bladder exstrophy variant of the exstrophy–epispadias complex (EEC). An

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exposed, everted bladder was clearly visible immediately below umbilical stump; the labia majora and minora are separated and splayed outwards and the clitoris is split and there was no obvious urine leak; the anus was present, normally located and patent with normal sphincter tone. There was no ano-rectal malformation (figure 1). Other physical examination was otherwise unremarkable. The defect was covered with sterile silicon gauzes and transparent waterproof dressing. Point-of-care cerebral, cardiac, abdominal, and kidney ultrasound scans were normal. The baby was then referred to pediatric surgery department for further management.

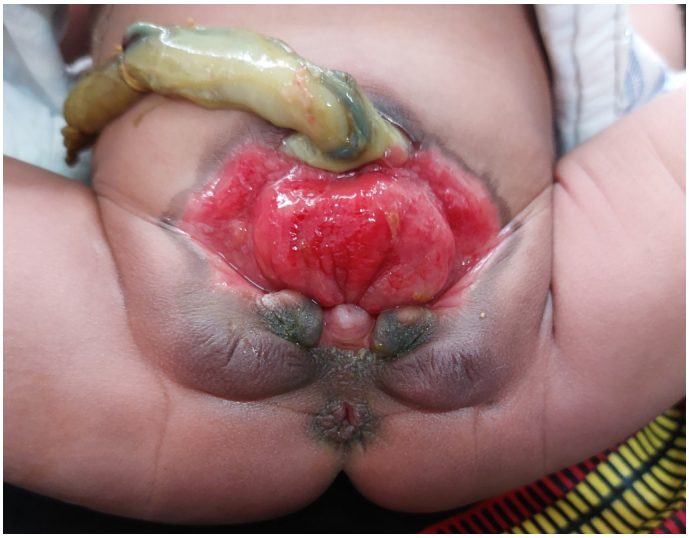

Figure 1: A female infant with classic bladder exstrophy

Discussion

Bladder exstrophy (BE) is a rare birth defect that was first described in 2000 BC (Buyukunal CS et al. 2011). The Classic bladder exstrophy (CBE) is characterized by a defect of the lower abdominal wall with evagination of the bladder plate (Dellenmark-Blom M et al. 2019; Jarosz SL et al. 2022). This embryologic malformation arises from an abnormally large cloacal membrane that causes a wedge effect, preventing the medial

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migration of the mesenchymal tissue. As a result, the lower abdominal wall is not well formed. A subsequent rupture of the cloacal membrane results in herniation of all the contents leading to the clinical picture of an exstrophy bladder (Kulkarni B et al. 2008).

The diagnosis of BE is clinical and often made by the neonatologist or paediatrician who are involved in the immediate management of the baby after birth before referral to the paediatric surgeons. Patients with bladder exstrophy undergo a series of surgeries between infancy and adulthood (Szymanski KM et al. 2019). The primary goals of surgery are to manage urinary continence, preserve renal function, and repair the cosmetic and functional aspects of the genitalia (Szymanski KM et al. 2019). The most important factor in determining the quality of life of these patients is the degree of urinary continence. Those who attain a sufficient bladder size with age can hold urine for 2 to 3 hours during the daytime and remain dry during the night time as well. However, a considerable proportion of children may not be able to do so due to poor bladder growth, leading to psychosocial issues, school dropouts, and delays in their education (Anand S et al. 2025). Therefore, children with BE would require long-term follow-up with the involvement of multiple disciplines in ensuring optimal care.

Conclusion

Bladder exstrophy is a rare birth defect that primarily affecting the urinary and genital tracts. Early detection and optimal surgical interventions helps achieve urinary continence and improved quality of life.

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