The Pitfalls of Imaging in the Antenatal Diagnosis of Bladder Exstrophy-Epispadias Complex in a Resource-low Setting

Theophile Njamen Nana¹,², Paul N. Tolefac²,3,4, Charlotte Nguefack Tchente¹,5, Calypse Ngwasiri⁴, Rita Frinue Tamambang³, Kouam Siegning⁶, Alain Mefire Chichom¹,7

¹Service of Obstetrics and Gynaecology, Douala general hospital, Douala, Cameroon
²Faculty of Health Sciences, University of Buea, Cameroon
³Faculty of Medicine and Biomedical Sciences, University of Buea, Cameroon
⁴Clinical Research Education Networking and Consultancy
⁵Faculty of Medicine and Pharmaceutical Sciences, University of Douala, Cameroon
⁶Faculty of Medicine University of Heidelberg Germany
⁷Service of Paediatric surgery, Douala Gynaeo-Obstetric and Paediatric Hospital, Douala, Cameroon

Corresponding Author: Dr. Theophile Nana Njamennjanatheo@yahoo.fr

ABSTRACT

Background: Bladder exstrophy is a rare congenital malformation of the genitourinary system usually associated with other malformations. Case summary: We report the case of a new born delivered vaginally at term with bladder exstrophy-epispadias complex. Three antenatal ultrasounds scan done all missed the diagnosis. Medical management was done, and the patient was discharged against medical advice 48 hours later with surgical intervention deferred due to lack of consent. Conclusion: The diagnosis and management of bladder exstrophy – epispadias complex remains a challenge in our surroundings. Radiologists should maintain a high index of suspicion during antenatal ultrasounds. Reporting: Care 2016 guideline.

Keywords: Bladder exstrophy, epispadias, antenatal ultrasound, case report

1. INTRODUCTION

Bladder exstrophy-epispadias complex (BEEC) is a complex congenital anomaly of the genitourinary system with a spectrum of congenital malformations ranging from isolated epispadias, to classical bladder exstrophy, to cloacal exstrophy as the most severe—and rarest—presentation. The malformation may extend to involve the genitalia, the abdominal wall muscles, the pelvic floor musculature, and the bony pelvis¹,². It is an extremely rare congenital malformation with an overall incidence estimated by Nelson et al.³ at 2.15 per 100,000 live births with classical bladder exstrophy estimated at 1:10,000 to 1:50,000 live births and epispadias estimated at 1:117,000 live births⁴. BEEC results from an insult during the development of the urogenital system during which there is a disorder leading to cloacal membrane overgrowth preventing medial migration of mesenchymal tissue⁵. Surgery remains the main stay of management with adequate abdominal wall closure, achieving urinary continence with renal function preservation and adequate cosmetic/functional genital reconstruction being the approach considerations. The currently
most utilized strategy is a staged approach. In the standard-staged repair primary closure of the bladder without osteotomy, and without genital reconstruction in males, is attempted in the first 72 hours of life; between 6 and 12 months of age, the epispadias repair is performed in males. Bladder neck reconstruction follows around the age of 5 years if a reasonable bladder capacity is reached[9]. We present the case of a newborn delivered vaginally at 39 weeks and two days of gestation with bladder extrophy – epispadias complex. Three prenatal ultrasounds scan done all missed the diagnosis. Medical management was done, and the patient was discharged against medical advice 48 hours later refusing surgical intervention.

2. CASE REPORT

A 29-year-old gravida 3 para 2 (G3P2) apparently well throughout pregnancy presented at our service in active labour at term (39 weeks, 2 days) which progressed to the delivery of a living male with Apgar 8, 9 and 10 at 1st, 5th, and 10th minute respectively. Prenatal ultrasounds were done at 12 weeks, 23 weeks and 31 weeks of pregnancy were normal. Maternal serologies for HIV, HBsAg, HCV, Toxplasma, and rubella were all negative, and her fasting blood sugar was normal. Apart from routine sulfadoxine – pyrimethamine and iron and folic acid were taken, she denies taking other drugs during pregnancy.

The newborn had a genitourinary defect as shown in figure 1 and 2. There was an abdominal wall defect with exposed and everted bladder draining urine which was clearly visible immediately below the umbilical stump (Fig. 1); a completely dorsally opened urethral plate ran from bladder neck down to the open glans (Fig. 2); left and right corpora cavernosa were clearly visible beneath and alongside urethral plate (Fig. 2); the scrotum was normally developed with both testes present and palpable within, (Fig. 1). The anus was present, normally located and patent. Other aspects of the physical examination were otherwise normal.

Figure 1: Showing bladder everted and scrotum displaced downward

This genitourinary anomaly was consistent with the BEEC. The defect was discovered in the delivery room with sterile silicon gauzes, and transparent waterproof dressing and the baby transferred to the neonatology unit. Trans-fontanel, cardiac, pelvis, and renal ultrasound were then performed and normal. Biological investigations including full blood count, serum electrolytes and urea and creatinine were all normal. His blood group and Rhesus factor were determined as B rhesus positive. The baby spent 48 hours in the neonatology unit, and the parents signed for discharged against medical advice deferring surgical intervention.

3. DISCUSSION

Epidemiology

BEEC is a rare congenital malformation of the genitourinary system with the overall incidence estimated by Nelson et al.\(^{3}\) in 2.15 per 100,000 live births, with an even male-to-female ratio (OR = 0.989; 95% CI = 0.88–1.12), and a significantly increased incidence in white compared with non-white neonates (incidence, 2.63 vs. 1.54 per 100,000; p < 0.0001). This incidence varies depending on the geographical location and socioeconomic status\(^{3}\). The clinical syndrome includes bladder exstrophy, epispadias, and cloacal exstrophy. Classic bladder exstrophy occurs in 1:10,000 to 1:50,000 live births\(^{4}\); epispadias is estimated to occur in 1:117,000 live births\(^{6}\), and cloacal exstrophy in 1:250,000 births\(^{7}\). The incidence is not known in our sub region.

Embryology

The embryology of the BEEC has been long studied, yet debate still exists over the specific origins of the anomaly\(^{8}\). BEEC is thought to be derived from a derangement in the fusion of the mesoderm during the first week of life\(^{1}\). Normally, at the end of the third week of life, intermediate mesoderm invaginate to form the genitourinary system, while the lateral plate mesoderm will contribute in forming the primitive gut tube\(^{1,2}\). A disruption in this interaction, linked to cloacal membrane overgrowth preventing medial migration of mesenchymal tissue, is reported to give rise to BEEC\(^{9}\). The severity of the resulting condition depends on the point at which disturbed mesodermal layers interaction begins. Given the embryological origin of BEEC, it is often associated with other peculiar orthopaedic, musculocutaneous, and gynaecological conditions. Associated upper urinary tract anomalies are rare. Gastrointestinal and spinal/neurological anomalies can be associated in patients with BEEC. Some of the associated malformations include vesicoureteric reflux, megaureter, horseshoe kidney, ureterocele, abdominal wall defect, umbilical hernia, neural tube defects, spinal dysraphism, club feet, congenital hip dislocation, pubic symphysis diastasis, anteriorly displaced anus, imperforate anus and Mullerian anomalies\(^{1,5,10}\). In our indexed case, after a thorough
clinical assessment and morphological investigations, the only associated anomaly was abdominal wall defect as the anus was patent, renal, trans-fontanel and pelvic ultrasounds were normal.

Antenatal diagnosis

The diagnosis of bladder exstrophy can be made before delivery by prenatal ultrasounds. It is based on the non-visualization of the bladder during the first-trimester ultrasound, but in most cases, it is confirmed by the morphological ultrasound of the second trimester. This second trimester morphological ultrasounds also helps in the diagnosis of other associated malformations(13). In one study, the sensitivity of prenatal ultrasound for diagnosis of congenital malformations was estimated at 7/36 (19%) at the 17-18th week of gestation, and 13/36 (36%) overall(12). In another study, forty-three prenatal ultrasounds from 25 pregnancies with bladder exstrophy, where the ultrasound was done between 14-36 weeks of pregnancy, the diagnosis of bladder exstrophy was made before delivery in only three cases. Five factors associated with bladder exstrophy were identified: 1) non visualization of the bladder on ultrasound in 12 of 17 cases (71%); 2) lower abdominal bulge representing the exstrophied bladder in eight of 17 cases (47%); 3) a small penis with anteriorly displaced scrotum in eight of 14 males (57%); 4) low set umbilical insertion in five of 17 cases (29%); and 5) abnormal widening of the iliac crests was seen in three of 17 cases (18%)(13). In our patient, three prenatal ultrasounds were done between 12-31 weeks, and none was diagnostic bladder exstrophy. The radiologists didn’t mention any of the above features. Factors contributing to missed prenatal diagnosis may include a low incidence of the pathology, lack of clinical suspicion, few radiologists and high workload, poor local training and lack of continuing medical education.

Management

Bladder exstrophy is a paediatric urologic emergency. Immediate medical management usually consists of covering the extruding viscera with sterile silicon gauze surrounded by an occlusive dressing to prevent air contact and dehydration, fluid and electrolyte balance and antibiotic prophylaxis if indicated(2). Surgical correction varies depending on the type and severity of the defect. Currently, a staged approach is the strategy most commonly used. In the standard-staged repair, a primary closure of the bladder without osteotomy, and without genital reconstruction in males, is attempted in the first 72 hours of life; between 6 and 12 months of age, the epispadias repair is performed in males. Bladder neck reconstruction follows around the age of 5 years if a reasonable bladder capacity is reached(9). Alternatively, the neonatal primary complete repair and the deferred primary complete repair have been proposed(14). Our patient benefited from the immediate medical management consisting of hospitalization in the neonatology unit, covering the extruded viscera with wet gauze and fluid resuscitation. We didn’t give antibiotics. While the surgical team was mobilized, for the closure of the defect the mother opted to go home against medical advice. This discharge against medical advice can be explained by the low socioeconomic status of the mother, financial constraints, refusal of the surgical intervention, and lack of confidence in the health care system which may all contribute to her leaving the hospital to seek for alternatives (traditional medicine).

4. CONCLUSION

Bladder exstrophy – epispadias complex is a rare congenital malformation. Even in the era of highly advanced information technology, the prenatal diagnosis of congenital malformations remains a challenge. Radiologists and other experts doing prenatal ultrasounds should maintain a high index of suspicion during while doing prenatal ultrasounds. The management of BEEC is multidisciplinary.

DECLARATIONS

Ethics approval and consent to participate: Ethical approval was obtained from the ethical committee of Douala general hospital. A copy of is available for review upon request by the Editor-in-Chief of this journal.
Consent for publication: Obtained from the patient’s mother. A copy of is available for review upon request by the Editor-in-Chief of this journal.
Availability of data and material: The patient’s medical records are available in the services of obstetrics and gynaecology and neonatology of Douala general hospital on reasonable request by the editor – in – chief.
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