Annals of Case Reports and Clinical Studies
Case Report (ISSN: 2834-5673)

Early Prenatal Diagnosis of Isolated Bladder Exstrophy

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Citation: Geya Gottipati, Geeta Kolar, Suseela Vavilala. Early Prenatal Diagnosis of Isolated Bladder Exstrophy. Ann Case Rep Clin Stud. 2025;4(12):1-6.

Received Date: 24 December 2025; **Accepted Date:** 28 December 2025; **Published Date:** 30 December 2025

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ABSTRACT

Introduction: Isolated Bladder Exstrophy is a rare congenital abnormality. Reported incidence is 1 in 30,000 to 1 in 50,000 live births with a significant male preponderance with a male to female ratio of 3:1. It is a midline fusion defect of urinary bladder, anterior pelvis, and genitalia. Prenatal detection rate is only 15% and average gestational age at detection is after 25 weeks. Early prenatal diagnosis helps in providing options of continuation vs termination.

Case report: We report a case presenting at 20 weeks with persistent non visualization of bladder, normal liquor and normal appearing kidneys, and a homogenous solid mass of 3x2x2cm in the lower abdomen. The cord insertion appeared low with the umbilical arteries running alongside the bulging mass. Detailed structural survey revealed no other associated abnormalities confirming the diagnosis of isolated bladder exstrophy. MRI further confirmed the diagnosis. Couple were counselled regarding the diagnosis and need for reconstructive surgery, short and long-term quality of life following reconstruction of bladder exstrophy. Couple opted for termination of pregnancy. The diagnosis of isolated bladder exstrophy was confirmed at autopsy.

Conclusion: Early prenatal diagnosis allows parental counselling, planning birth at tertiary centre and offer better postnatal care involving multidisciplinary team.

Keywords: Bladder; Abdominal wall defect; Termination of pregnancy.

INTRODUCTION

Classic Bladder Exstrophy is a rare congenital abnormality. Reported incidence is 1 in 30,000 to 1 in 50,000 livebirths [1]. It is a part of spectrum of Bladder exstrophy epispadias complex. Mildest form being epispadias and most severe form being cloacal exstrophy. Urinary bladder can be detected as early as 10 weeks. Fetal urinary production starts at 9 weeks of pregnancy and increases significantly beyond 16 weeks. The ultrasound of normal fetal bladder usually demonstrates round or oval echo-free area in the fetal pelvic region.

Identification is easy because of its pelvic location between the umbilical arteries. Physiological emptying and refilling occurs usually within 30-45 minutes. Bladder exstrophy is a midline fusion defect of urinary bladder, anterior pelvis, and genitalia. Prenatal detection rate is only 15% and average gestational age at detection is after 25 weeks. [2] Early prenatal diagnosis helps in counselling and giving options of continuation of pregnancy Vs termination.

CASE REPORT

We report a case of early diagnosis of classic bladder exstrophy with sono-pathological correlation. Routine fetal anomaly scan on a second gravida at 20 weeks revealed persistent non visualization of bladder, normal liquor and normal appearing kidneys, and a homogenous solid mass of 3x2x2cm in the lower abdomen (Figure 1,2). The cord insertion appeared low with the umbilical arteries running alongside the bulging mass (Figure 3). Detailed structural survey revealed no other associated abnormalities confirming the diagnosis of classic bladder exstrophy. Fetal MRI further confirmed the diagnosis (Figure 4,5). Couple were counselled regarding the diagnosis and need for reconstructive surgery, short and long-term quality of life. Couple opted for termination of pregnancy. The diagnosis of bladder exstrophy was confirmed at autopsy with epispadias, short penis and undescended testis and pubic diastasis (Figure 6,7).

DISCUSSION

Urinary bladder can be detected as early as 10 weeks and physiological emptying and refilling occurs usually within 30-45 minutes. Exstrophy-epispadias complex (EEC) represents a spectrum of genitourinary malformations. Severity can range from epispadias (E) to classical bladder exstrophy (CEB) and exstrophy of the cloaca (EC) i.e., OEIS complex(3)

Classic bladder exstrophy needs reconstruction of bladder and genitalia in multiple sittings starting at birth, till puberty. Overall survival prognosis is excellent, but quality of life issues are expected like urinary incontinence(25%),erectile dysfunction(25%),male infertility (90%)female infertility(75%)psychosexual issues and malignancy i.e., bladder or colon carcinoma(17%)women are predisposed to vaginal uterine prolapse [4]

Pathophysiological changes here are bladder is open to the abdominal wall.Hence urine is released immediately into the amniotic cavity and the bladder is never visible as a fluid-filled structure. Both bladder halves are exposed and everted leading to inflammation of the exposed mucosa. This may cause irregular thickening of the posterior bladder wall-visible on ultrasound as an irregular contour of the lower anterior abdominal wall. Ultra sonography features include 1)Non-visualization of the bladder with normal amniotic fluid 2)Presence of parallel umbilical arteries on color Doppler 3)Mass in the suprapubic region on the anterior abdominal wall 4)Low-set insertion of the umbilical cord 5) Genital abnormality 6) Splayed iliac bones (3)(5)

Upon reviewing the literature, newer proposed technique for early detection of bladder exstrophy is “umbilical cord insert to genital tubercle length” A prospective trial was conducted at the Chaim Sheba Medical Center over 10 years from 2005 to 2015 and all cases were evaluated before 18 weeks and distance was measured in mid sagittal plane. All cases with bladder exstrophy had an umbilical cord insertion–to–genital tubercle length below the fifth percentile for gestational age i.e., below 10 mm (6).

Associated malformations can be a part of a complex malformation syndrome like OEIS complex (Omphalocele, Exstrophy of the bladder, Imperforate anus, Spinal defect), limb body wall complex syndrome, associated genital abnormalities like epispadias, short or split penis and undescended testis in males, bifid clitoris and uterine and vaginal abnormalities in females. Hence a detailed fetal structural survey to look for accompanying bowel malformations and genital malformations. [5] "Target sign" by the perianal muscular complex-The fetal anus might be identified as a hypoechoic ring with an echogenic center. Absence of this sign indicates anal atresia / rectal agenesis with imperforate anus.

Following is a table showing differences between bladder and cloacal exstrophy

	Bladder exstrophy	**Cloacal exstrophy**
Bladder	Absent	Absent
Bowel	Bowel loops are never extruded and rectum is normally formed	Extrusion of rectum, bladder and genitourinary tract
Spine	Normal	Associated -tethered cord, lipomeningocele, and lipomyelocystocele
Kidneys	Normal	Seen in 60% of cases Hydronephrosis , renal developmental variants
Genitalia	Can be present	Present
Other features		Omphalocele "Elephant trunk" sign

Genetic counselling should be offered. It is of Sporadic occurrence hence no role for Prenatal karyotyping. Recent genetic studies have identified duplications of chromosomal region 22q11.2 as a rare cause for isolated classic bladder exstrophy. [7] Recurrence rate is around 3-4% in siblings. Amniocentesis for fetal sex determination (outside India) in cases of ambiguous genitalia can be offered. This helps in counselling the couple and planning the postnatal reconstruction.

Antenatal management

Course of pregnancy is not affected, there is no increased risk of IUD or neonatal compromise at birth. Regular sonographic evaluation has to be conducted to exclude deterioration of the fetal status. Counselling by a pediatric urologist and a pediatric surgeon should be done. Cesarean section should be reserved only for

obstetric indications. Delivery should be planned in a tertiary center with a pediatric urologists and anesthesiologists in place.

Postnatal diagnosis and management

Following features help us in establishing postnatal diagnosis. 1)Bladder plate protrudes immediately beneath the umbilical cord.2) Rectus muscles are divergent.3) Outward rotation of innominate bones and eversion of the pubic rami 4) In males-phallus is short, splayed glans and dorsal chordee 5) In females-Mons pubis, Clitoris and labia are separated and Vaginal orifice may be displaced anteriorly 6) Bilateral inguinal hernias are common (15% of females and 56% of males).

At birth, reconstruction of the bladder and primary closure of the abdominal wall with/ without iliac osteotomy should be done. Best if performed within 3 days of life. At 12–18 months, corporeal lengthening and dorsal chordee release with urethroplasty in male, vulvoplasty and clitoroplasty in female patients should be performed. At 4–8 years, bladder neck reconstruction and additional operations for urinary continence, anti-reflux procedures and additional complementary operations as necessary should be performed. After puberty, cosmetic surgery of the female mons area and abdominal wall has to be offered. [8]

CONCLUSION

When there is persistent non visualization of bladder, always look for the presence of fetal kidneys and for fetal growth and liquor. If there is an absent bladder despite above findings being normal, suspect bladder exstrophy. Early prenatal diagnosis allows parental counselling, planning birth at tertiary centre and offer better postnatal care involving multidisciplinary team. Hence visualization of bladder and clear documentation should be a part of anatomical assessment at 11-13+6 weeks scan and not include as an optional structure.

Ultrasound Images

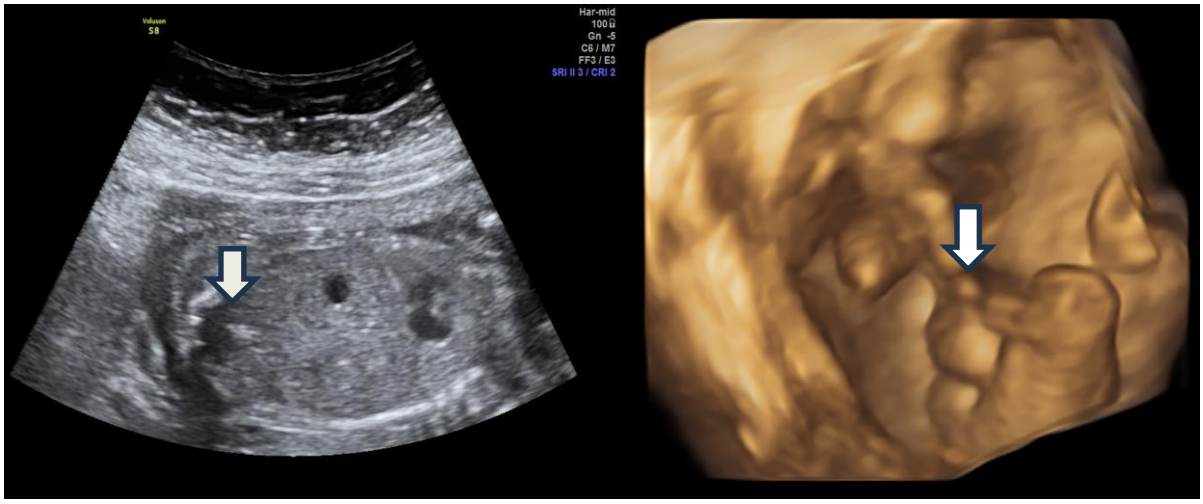

Figure-1: Coronal plane of fetal abdomen showing persistent non visualization of fetal bladder

Figure-2: 3D reconstruction of fetal abdomen showing bulging solid mass with low cord insertion

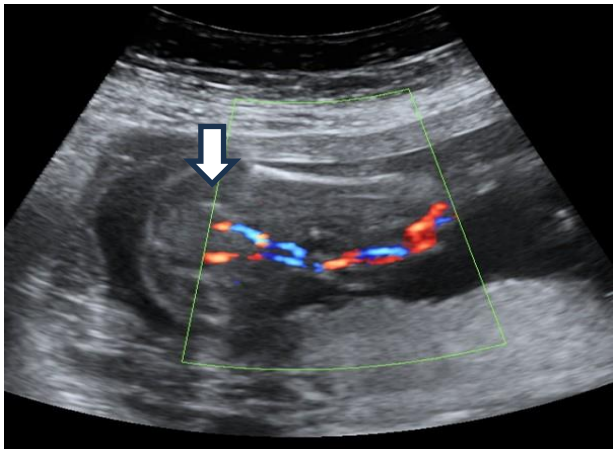

Figure-3: Coronal plane of fetal abdomen, showing absent bladder with umbilical arteries running alongside

Fetal MRI Images

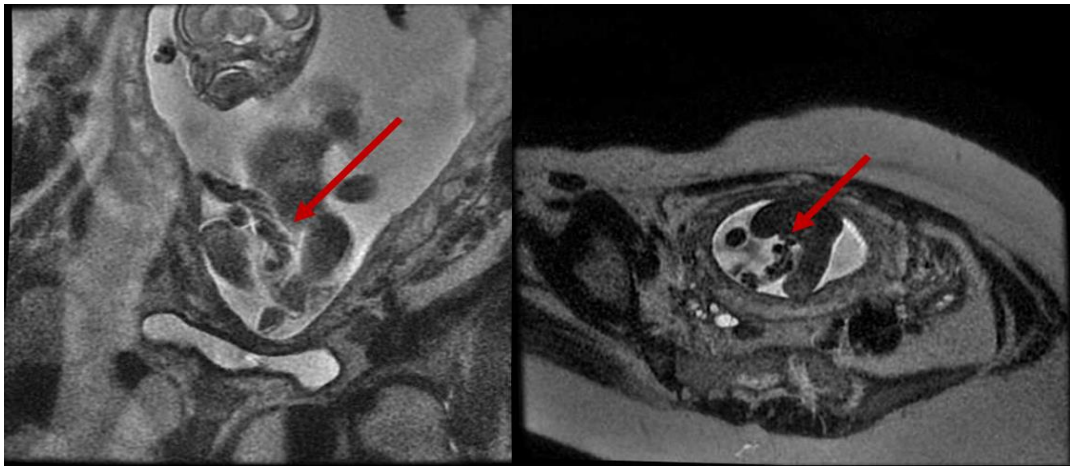

Figure- 4: Coronal MRI image showing bulging mass with low cord insertion

Figure- 5: Axial MRI image showing anterior abdominal wall defect

Autopsy Findings

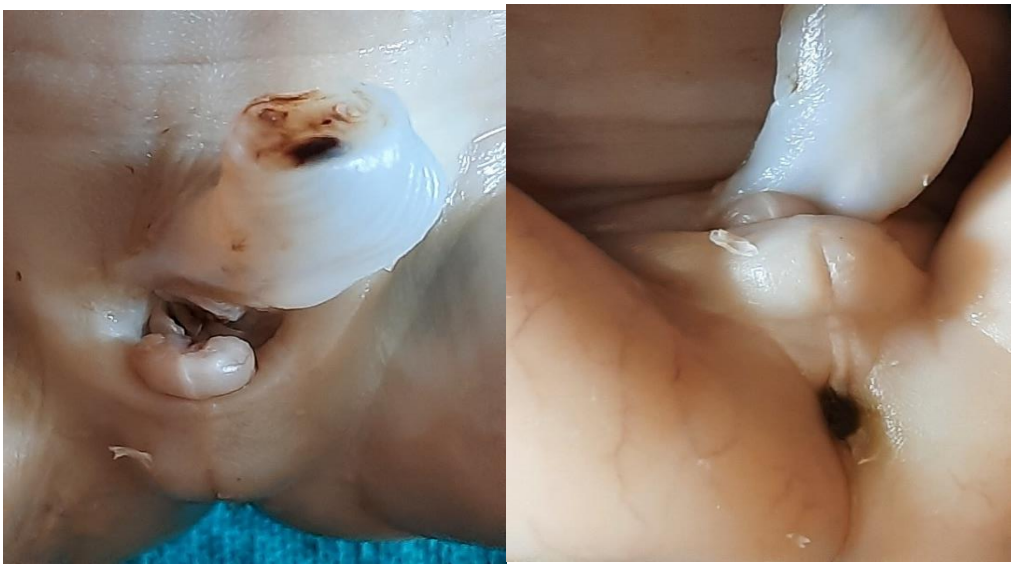

Figure-6: Autopsy image showing anterior abdominal wall defect, low cord insertion and epispadias

Figure -7: Autopsy image demonstrating anal orifice, thus excluding cloacal exstrophy

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