

Title: In Utero Diagnosis of Fetal Bladder Exstrophy

Introduction:

Bladder exstrophy is a rare congenital anomaly affecting 1 in 30,000-50,000 live births. Isolated classic bladder exstrophy is the most common form of bladder exstrophy-epispadias complex: a spectrum of anomalies involving the urinary tract, genital tract, gastrointestinal tract, and musculoskeletal system.

Case Report:

A 27-year-old, G2P1001 at 23 weeks EGA had a fetal anatomy ultrasound that was concerning for absent bladder and abnormal external genitalia. The patient had negative male NIPT results. Detailed obstetrical sonography revealed anomalies involving the lower anterior abdominal wall and external genitalia and no visible bladder. The amniotic fluid volume was normal. These findings raised concern for bladder exstrophy. Further investigation with MRI at 26 weeks EGA revealed a non-visualized bladder, a mass-like protrusion from the anterior abdominal wall, and normal kidneys. Following birth, the prenatal diagnosis of bladder exstrophy was confirmed. Microphallus was also noted with bilaterally descended testes and a low-set umbilical cord.

Discussion:

Exstrophy of the bladder results from failed mesenchymal cell migration leading to incomplete closure of the lower anterior abdominal wall.¹ Delineating bladder exstrophy from other abdominal wall or urogenital defects on prenatal ultrasound may be accomplished by identifying the typical findings of bladder exstrophy. Differential diagnoses may include renal agenesis, omphalocele, gastroschisis, and cloacal exstrophy.

Conclusion:

In utero diagnosis of bladder exstrophy may be challenging and requires a thorough obstetrical sonogram, while allowing sufficient time for the bladder to fill when not initially visualized. Fetal MRI is an important secondary testing modality to further investigate and confirm the diagnosis of fetal bladder exstrophy and exclude other diagnoses.