Delayed recognition of bladder exstrophy and persistent cloaca: An Omphalocele exstrophy imperforate anus spinal abnormality (OEIS) Variant

Itunu O. Arojo MD, Lynn L. Woo MD.
HPI

• Newborn term infant with abnormal prenatal ultrasound on 22-week anatomy scan.
  – Widened pubic diastasis
  – Dilated rectosigmoid colon
  – Mild polyhydramnios
  – Distended bladder in pelvis protruding into the omphalocele
    • Empties during exam

• Mother aged 31 G3P3, otherwise healthy

• Prenatal MRI obtained to confirm findings
Physical Exam

- Omphalocele
- Asymmetric labia/clitoral tissue
- Fistulous connection to the bladder
- Imperforate anus with a single mucosal-lined opening in perineum
Diagnostic evaluation

• Cystoscopy/EUA
  – cloacal malformation
  – Appearance of an intact urethra/bladder neck complex
  – vaginal duplication
  – fistulous tract to the rectum
Diagnostic evaluation

• Cystogram
  - smooth-walled bladder without evidence of reflux

• Spinal ultrasound was normal.
- DOL 4
  - Omphalocele rupture revealed bladder extrophy
Hospital course

- She underwent a diverting colostomy on DOL 4.
- Discharged home with seran wrap coverage and prophylactic antibiotics
- Cloacal repair and closure of bladder extrophy is planned at age 6 months of age.
Discussion

• Cloacal extrophy is a rare and complex congenital anomaly
• Incidence of 1/200,000-400,000 live birth
• Occurs along a spectrum
• Associated omphalocele often contains bowel or liver
• Prenatal diagnosis of extrophy is difficult to make. Often a diagnosis of omphalocele/gastroschisis made and the extrophy overlooked.
Conclusion

• This case highlights the complexity associated with diagnosis of exstrophy-epispadias complex. Despite advances in prenatal imaging, diagnosis of this condition relies upon physical examination with the knowledge that each presentation can be different; not fitting in one box.
References