

[EP05.06] Cloacal anomaly in a male fetus with hydronephrosis and ectopic ureter associated with caudal regression syndrome

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Introduction:
Cloacal anomalies in males are rare, with only a few cases reported in literature. We describe a prenatally suspected case of male cloacal anomaly on the background of caudal regression, hydronephrosis and ectopic ureter.

Case History:
A primigravida in her late 20s with no significant medical or family history was referred for a structural anomaly scan at 19 weeks. First trimester aneuploidy screening and NIPS were low risk.

- Ultrasound Features:**
Fetal growth parameters were normal, mild polyhydramnios noted.
- Genitourinary:** There was right renal pelvicalyceal dilatation (UTD A2/3) with blunted peripheral calyces, ureteric dilatation and a normal left kidney and ureter. The dilated ureter had an ectopic insertion into the posterior urethra below the bladder neck (Fig .A,B).
 - Gastrointestinal:** The rectum joining into a common channel with the posterior urethra. (Fig A)The perianal muscular complex was indistinct, pseudotarget sign seen(Fig. D). The upper GI tract was unremarkable.
 - Skeletal:** The fetal spine was short with lumbosacral defects, fused sacral vertebrae and a tethered cord.(Fig.C)

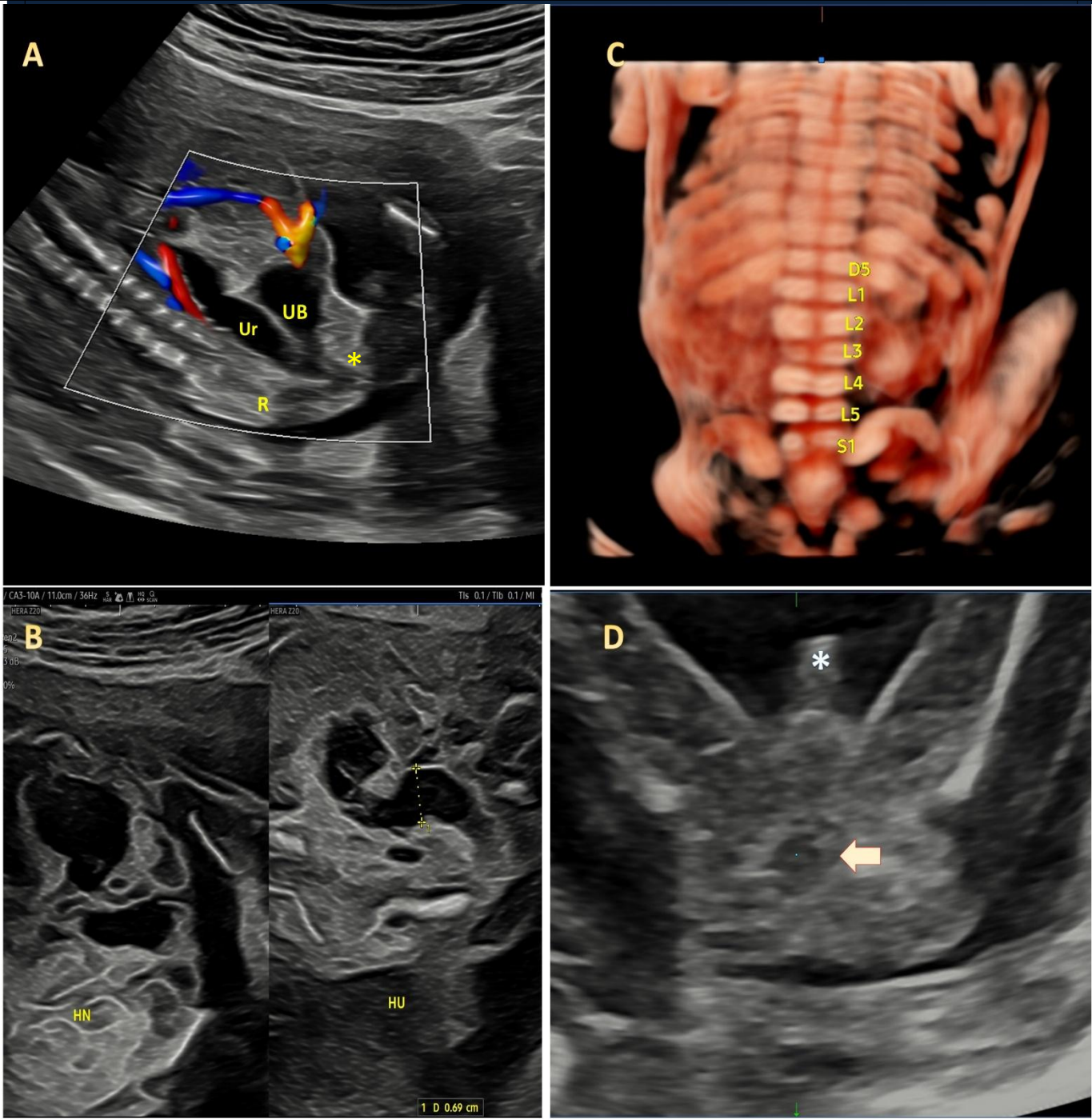
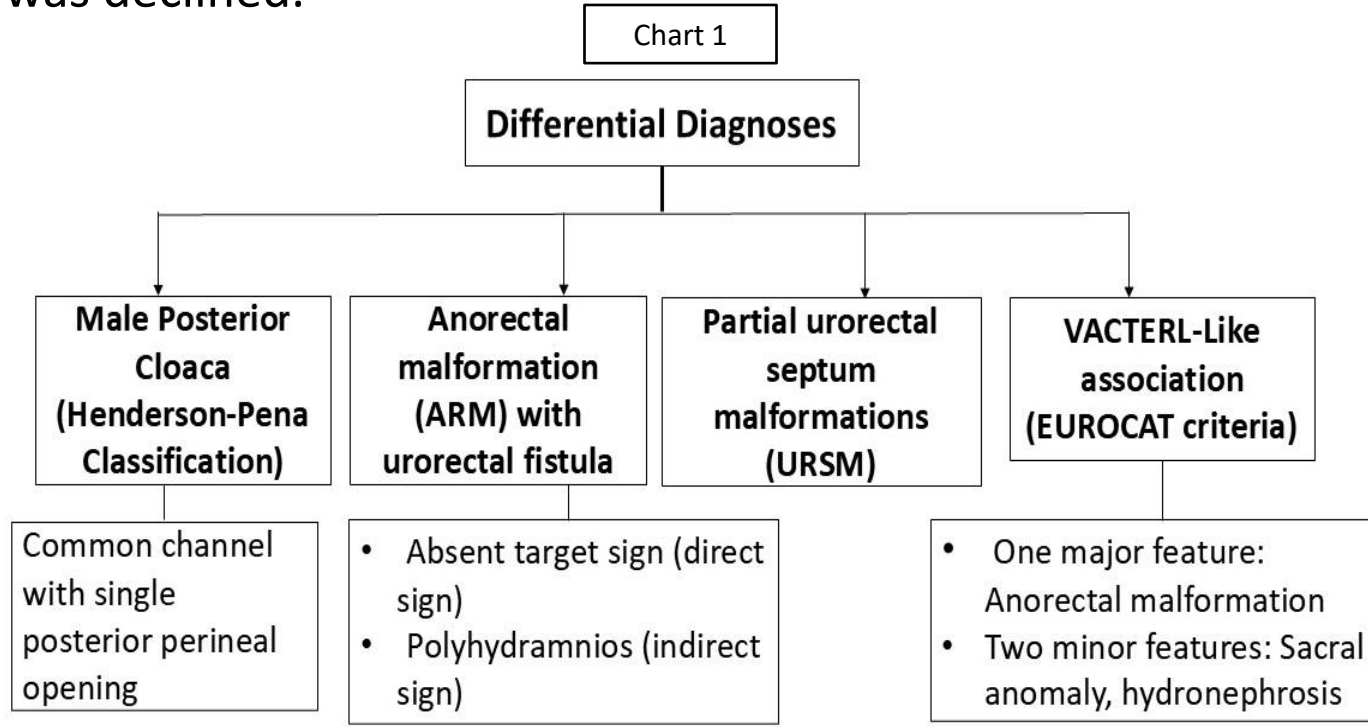


Fig A. Urinary bladder(UB), Ureter(Ur) and Rectum(R) seen opening into a common channel (asterix). Fig B. Hypoplastic sacrum. Fig C. Right hydronephrosis and hydroureter. Fig. D. Pseudo-target sign(arrow) ,: an indistinct perianal muscular complex.

Discussion:
Cloacal anomalies are under-reported in males and a considerable overlap exists amongst various subtypes (Chart 1). Up to 10% of cases have a chromosomal or genetic association. In this case, amniocentesis revealed a normal chromosomal microarray. Further tests were declined and pregnancy terminated at 22 weeks on parental request. On examination, the abortus had a single perineal opening posterior to the scrotum, detailed post mortem examination was declined.



Conclusion:
This case highlights the importance of systematic evaluation of the ureteric insertion, fetal perineum, anorectum and spine to unmask complex diagnoses in the presence of common anomalies such as renal pelvicalyceal dilatation.