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Sonopathological correlation of cloacal malformation in male fetuses

Shuchitra S, Vidhya D, Keerthana K, Monica Reddy M, Ponmozhi G, Manikandan K, The Fetal Clinic, Puducherry, India
The Fetal Clinic, Puducherry, India

Objective

The aim is to understand the presentation and ultrasonographic features of cloacal malformation in male fetuses.

Methods

This is a case series of five male fetuses with ultrasound diagnosis of cloacal malformation with recto-urinary fistula and pathological correlation in two fetuses.

Results

Ultrasound features: The cases included in the study had a common diagnosis of distended rectum with echogenic meconium pellets. Peri- anal muscular complex and anal canal were not visualized in all five cases. The next common finding was dysplastic and shrunken kidney, either unilateral or bilateral. In addition to the above said findings, there were additional anomalies involving other systems such as spine, CNS and cardiac. Autopsy features: Autopsy was performed in two cases. In both the cases, the external anal opening was not present. The recto-sigmoid colon appeared distended and filled with meconium pellets. The distended colonic segment and the urinary bladder ended in a common pouch resulting in recto-urinary fistula. This fistulous communication causes the meconium to get precipitated thus leading to pellet formation. Cloacal anomalies, characterized by a single perineal opening for urinary, gastrointestinal and reproductive tracts are very rare malformations with an incidence of 1 per 50,000 live births. Prenatal diagnosis of cloacal malformation is made more often in female fetuses due to unmistakable hydrocolpos. Prenatal diagnosis in male fetuses is usually mistaken for simple anorectal malformation. Cloacal malformations are usually associated with anomalies of urogenital and gastrointestinal system and may occur as a component of syndromes such as VACTERL.

Conclusion

Prenatal diagnosis of cloacal malformation should be suspected if there is combination of renal, gastrointestinal and spinal anomalies. A blind ending distended rectum filled with echogenic meconium pellets is a strong ultrasound sign of cloacal anomaly in male fetuses.

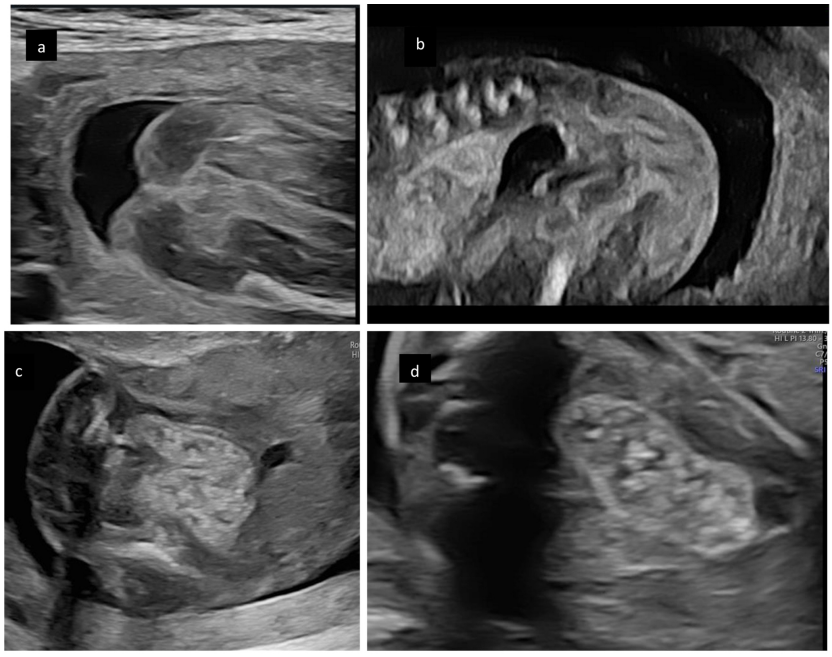

Figure 1 Ultrasonographic presentation of cloacal malformation with recto urinary fistula in male fetus. a) Axial image of the perineal region demonstrating absence of peri-anal muscular complex, b) Sagittal image showing smooth bottom and anal atresia, c) Coronal image of the abdomen demonstrating distended pelvic colon with echogenic contents, d) Distended pelvic colon filled with meconium pellets

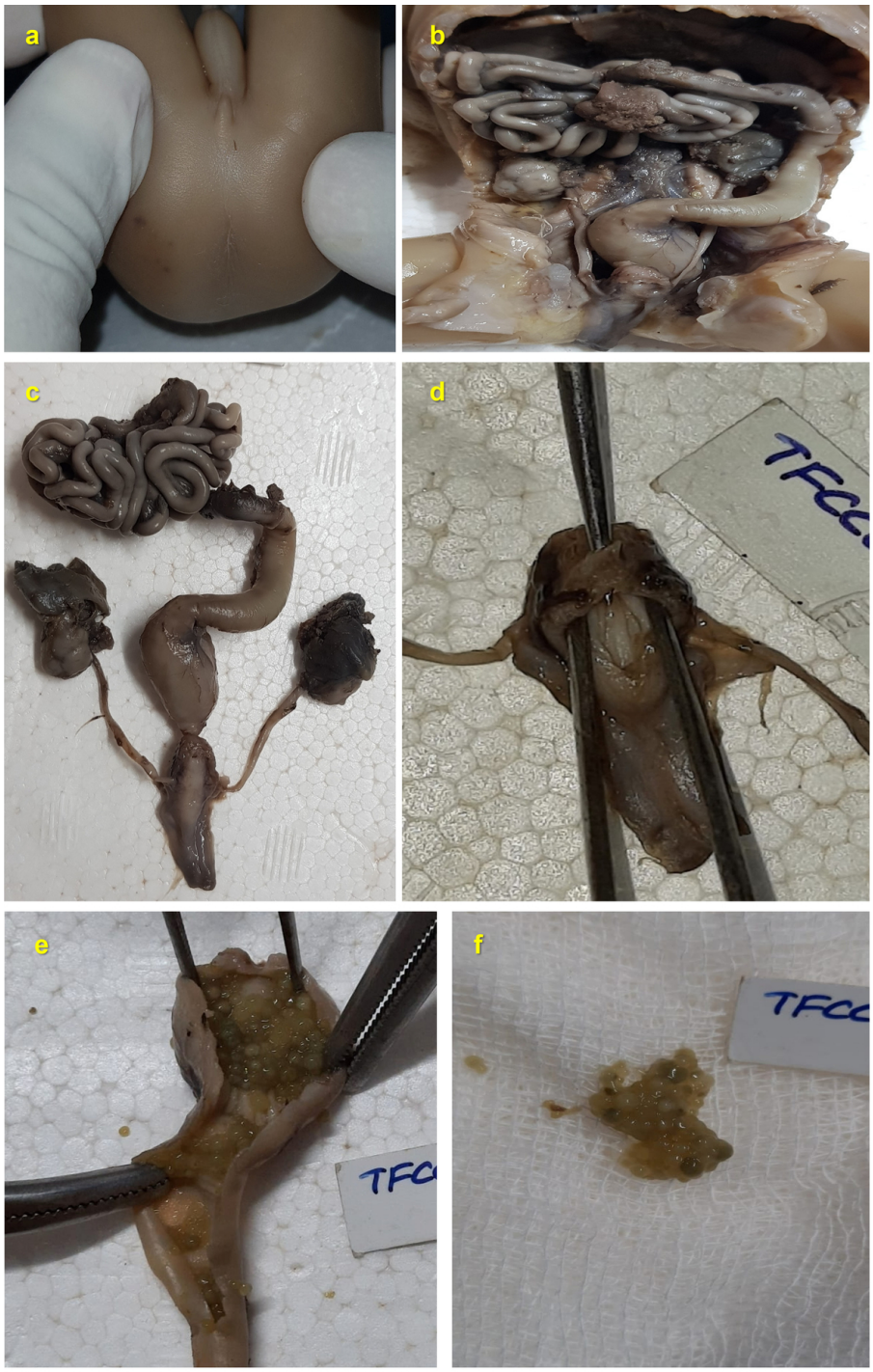

Figure 2 Autopsy correlation with ultrasound findings: a) Absent external anal opening b) Coronal section of the abdomen showing dilated recto-sigmoid colon with absent anal canal c) En block section of small bowel and KUB showing dilated blind ending rectum in close proximity with the dome of bladder d) Anterior wall of the bladder is cut open to demonstrate the fistulous opening in the dome of the bladder (Probe in the fistulous opening) e) and f) Longitudinal cut section through the distended colonic segment showing abnormal meconium pellet formation