Perineal Bowel – An Isolated Anomaly or Part of a Complex?

Dear Sir,

Perineal masses in the neonatal period are rare anomalies and may result from defective development of the cloaca. They can be associated with urogenital and anorectal malformations, and duplication anomalies of the hindgut may occasionally present as perineal masses. We present a rare case of a male neonate with a perineal mass, containing abnormal colonic tissue, intimately associated with the hypospadiac urethra and the perineal musculature. Only one similar case has been described previously in the English language literature.[1]

A 6-day-old full-term, 2.7 kg male presented with a fleshy mass protruding from the perineum noticed at birth. He was the 2nd child to a non-consanguineous couple, born by normal vaginal delivery following an uneventful antenatal period. There were no complaints of feed intolerance, excessive crying, abdomen distension, constipation or delayed passage of meconium or urinary retention. Examination revealed a 5 cm × 2 cm × 1 cm, fleshy, pinkish mass lying between the perineal urethral meatus and a normally placed anal opening, and it was protruding through a 1-cm subcutaneous opening just adjacent to the meatus [Figure 1a]. The scrotum was anteriorly transposed; however, the phallus was of adequate size. A water-soluble contrast, gastrografin, was injected into the mucus-filled lumen of the mass demonstrating a blind-ending track with no communication to the bowel or the bladder as seen on fluoroscopy. An ultrasound scan revealed the absence of the left kidney, and cystoscopy showed normal bladder anatomy except for the absence of the left ureteric orifice. With the above-mentioned evaluation, it was concluded that the mass had consistency and appearance of the bowel without any internal communication.

The mass was excised in toto, and the superficial perineal muscles were approximated. The urethral meatus required reconstruction as perineal urethrostomy, which was intubated with a 6-F Foley catheter till the 14th post-operative day [Figure 1b and c]. Histopathological examination of the mass showed epithelium, submucosa and muscle layers consistent with the architecture of a normal colonic wall; however, the ganglion cells were absent [Figure 2]. At present, the baby is doing well after 1 month of follow-up and is waiting for urethroplasty.

Perineal masses in the neonate are rare and may result from developmental anomalies or neoplastic etiology.[2] Duplication anomalies, especially of the hindgut, constitute an important differential diagnosis, and rectal duplications may present as cystic masses with perineal communication.[3] In our case, the perineal mass fulfilled Ladd's criteria of duplication cyst as it contained colonic mucosa and the muscle, although not attached, was close to the anorectum.[4] The unique feature was its external protrusion. The defective mesenchyme around the cloacal eminence leads to the development of abnormal perineum and anomalous external genitalia.[5] This might have happened in our neonate. Gupta *et al*. described the perineal rupture of a rectal duplication cyst associated with bladder exstrophy. They hypothesised that enlarging rectal duplication prevented mesodermal migration, leading to a weak perineum, which ruptured, allowing prolapse of the duplication cyst.[6] Thomas *et al*. described a hemiperineal anomaly in a neonate with hypospadias and severe penoscrotal transposition. It was thought to be due to antenatal rupture of the perineum, prolapse of the duplication cyst and healing of the ruptured area with scarring.[7] Sun *et al*. reported a 5-week-old infant having a perineal mass associated with hypospadias, like our case;[1] however, our patient was younger.

Prolapse of the rectal duplication through a malformed perineum is a rare anomaly. It might result from an early insult during the development of the cloacal eminence, and renal agenesis occurs as an association.

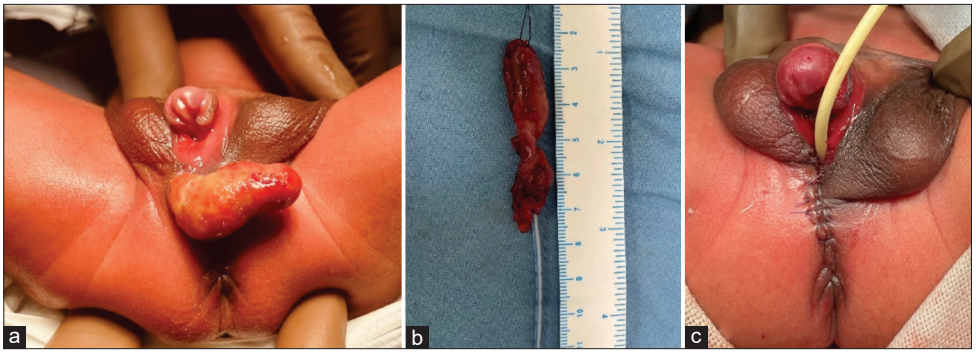


Figure 1: (a) Perineal bowel protruding between the normal anus posteriorly and hypospadiac urethra anteriorly. (b) Excised piece of the bowel with lumen. (c) Repaired perineum with urethral meatus

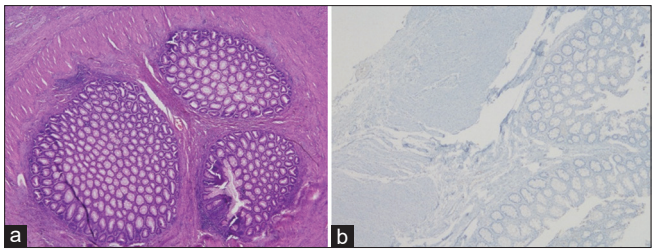


Figure 2: (a) Section from the mass demonstrating all the layers of the colon (H and E, ×20). (b) PHOX2B immunohistochemistry demonstrating the lack of ganglion cells in the submucosa and muscularis propria (×20)

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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