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ORIGINAL ARTICLE

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Perineal-mound defects

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Abstract Perineal-mound (PMD) and genital-fold defects cause anorectal malformations (ARM) in both sexes. They are common in females and usually present as a low anomaly (except rectovestibular fistula). They are rare in males and present as an intermediate anomaly. A common embryological explanation for these defects with varied presentation in males and females is discussed. These anomalies should be grouped separately in the classification of ARM. We present five male patients with PMD, three of whom had imperforate anus with rectobulbar fistula and perineal hypospadias and two who had imperforate anus with rectoperineal fistula.

Key words Anorectal malformation · Perineal-mound defect · Perineal hypospadias

Introduction

Perineal-mound defects (PMD) include embryological variations in both the perineal mound and genital folds. Despite the common embryological defect, the presentation is of differing severity in both sexes. These defects are common in females, and present as a low anomaly (except rectovestibular fistula). Anomalies in females include perineal canal, perineal groove, anovestibular fistula, anovulvar fistula, and anteriorly-placed anus. These defects are rare in males, presenting as an intermediate anomaly that clinically may simulate a low anomaly. Anomalies in males include rectobulbar fistula (RBF) with perineal hypospadias and rectoperineal fistula (RPF). These anomalies are so rare in males that they are not given a proper place in the classification of anorectal malformations (ARM). Management is according to the defect. Five males presented to us with imperforate anus with RBF and perineal hypospadias and with imperforate anus with RPF.

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Case reports

Cases 1–3

Three male infants presented with an absent anal opening, perineal hypospadias, bifid scrotum, and normally-placed testicles. A single opening was present in the perineum with passage of both feces and urine. Dye studies performed through the perineal opening were suggestive of an intermediate anomaly with a RBF. A preliminary colostomy was done in all cases followed by posterior sagittal anorectoplasty (PSARP). On exploration, the RBF was only 1.5 to 2.0 cm from the skin margin. The colostomy was closed 6 weeks after the definitive repair. In one case a urethroplasty was done, the other two boys are waiting for hypospadias repair.

Case 4

A 2-day-old male weighing 3.2 kg was admitted with an absent anal opening and a small opening at the penoscrotal junction, through which meconium was visible (Fig. 1). He had a normal scrotum with normally-descended testicles. An invertogram was suggestive of an intermediate anomaly. Cannulation of the fistulous tract at the penoscrotal junction was done using a fine feeding tube and a dye study was suggestive of a RPF. The patient was treated as an intermediate anomaly.

Case 5

A 2-day-old male was admitted with an absent anal opening and no other obvious congenital anomaly. An invertogram was suggestive of an intermediate anomaly. A left transverse divided colostomy was performed. During investigation for definitive repair, a distal loop cologram was done, which revealed a RPF that was missed during the initial local examination of the perineum (Fig. 2). A PSARP was performed, followed by colostomy closure.

Discussion

At the 16-mm stage in the embryo, when subdivision of the internal cloaca is complete, the urogenital and anal

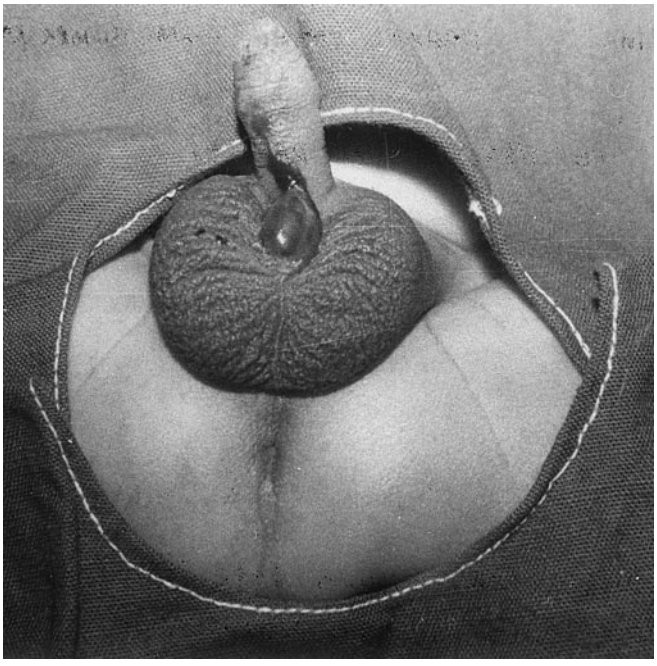

Fig. 1 Photograph showing meconium flyspeck at penoscrotal junction in a patient with rectoperineal fistula

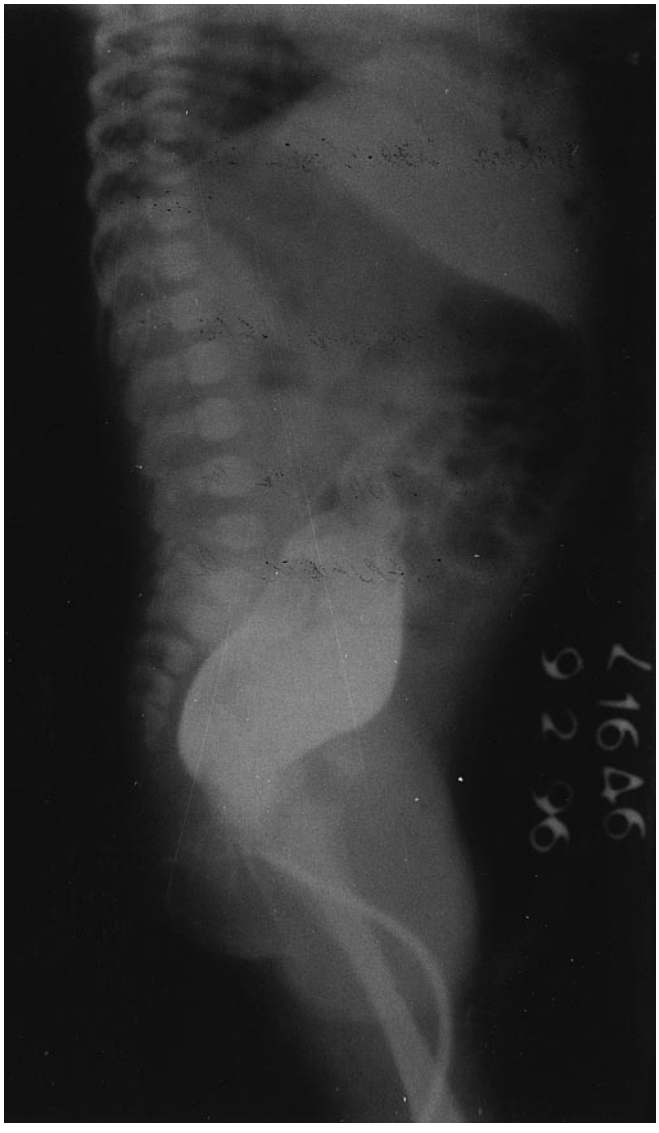

Fig. 2 Dye study showing long rectocutaneous fistula with dilated rectal pouch in a case of rectoperineal fistula

pits normally lie closely approximated. When the external cloaca is partitioned, the anal pit migrates posteriorly secondary to interposition of the perineal mound and genital folds [4, 5]. The perineal mound is the extension of the urorectal septum of the internal cloaca into the external cloaca, and forms the deep part of the perineum. The genital folds bordering the external cloaca grow medially over the mound, forming the superficial part of the perineum [6]. Excessive fusion of the genital folds posteriorly forms a covered anus. Defective fusion of the inner genital folds anteriorly leads to hypospadias.

Absence of development of the perineal mound, with defective fusion of both the inner and outer genital folds anteriorly and excessive fusion posteriorly, causes a RBF with perineal hypospadias in ARM. A RPF is caused by a rudimentary perineal mound with defective fusion of the outer genital folds anteriorly and excessive fusion posteriorly.

In females with a rudimentary perineal mound, the genital folds assume a major role in formation of the perineum. If, however, the genital folds fail to migrate medially, the perineum remains cleft as a perineal groove, the floor being the perineal mound and the sides the genital folds. If the cleft is bridged over by the genital folds, it becomes a perineal canal. If the medial migration of the folds affects a greater extent of the external cloaca, the anus may be covered and its lumen projected anteriorly onto the perineum or vestibule to form an anocutaneous or anovulvar type of female covered anus [6].

In 1982, the Japan Study Group [2] on ARM presented a case similar to ours where a fistulous opening was present at the tip of the penis with the rectum ending above the puborectalis sling. A dye study showed a fistulous tract parallel to the urethra. The study group recommended that this case should be considered an intermediate type in spite of the external orifice of the fistula at the tip of the penis.

In 1994, Shanbhogue et al. [3] presented three cases of RPF similar to ours. They stated that the malformation in males may be of the intermediate or high variety even when there is a perineal fistula. A suspected low ARM in males should have a detailed local examination of the perineum for the development of buttocks and location of the fistula. An X-ray film of the pelvis should be reviewed for sacral abnormalities. If a perineal fistula can be cannulated, the dye study is diagnostic. In low ARM in males during anoplasty, if there is any difficulty in locating the rectum the procedure should be abandoned

and the patient should be further investigated and treated appropriately.

In 1994, Currarino [1] presented two males with imperforate anus associated with a long rectocutaneous fistula running deep into the scrotum and communicating in its mid-portion with the bulbar urethra. He believes that in cases in which the perineal mound is present but is rudimentary, the rectum is rolled even more anteriorly between the mound and the fused inner genital folds to form a long, slender rectocutaneous fistula running deep into the scrotum and terminating on the undersurface of the penis. Imperforate anus with RBF and perineal hypospadias is still very rare, and no such case was found in the available English literature.

The reason for presenting this experience is to stress that PMDs are embryologically analogous in both sexes with varied presentation. In males the condition, being rare, should be treated with caution. Various classifications of ARM have been put forward, but no emphasis has been placed on PMD. These patients should be classified separately in the miscellaneous group of the Wingspread classification of ARM for better understanding of the anomaly.

References

1. Currarino G (1994) Imperforate anus associated with a recto-bulbar-cutaneous fistula. J Pediatr Surg 29: 102–105
2. Japan Study Group of Anorectal Malformations (1982) A group study for the classification of anorectal anomalies in Japan with comments to the international classification. J Pediatr Surg 17: 302–308
3. Shanbhogue LKR, Langemeijer RATM et al (1994) Recto-perineal fistula in newborn boys. J Pediatr Surg 29: 536–537
4. Stephens FD, Smith ED (1971) Anorectal malformations in children. Year Book Medical Publishers, Chicago, pp 128
5. Stephens FD, Smith ED (1988) Anorectal malformations in children, update 1988, Liss, New York, pp 186
6. Stephens FD, Smith ED, Hutson JM (1996) Congenital anomalies of the urinary and genital tracts. ISIS Medical Media, Oxford, pp 39, 45