Unusual association of diseases/symptoms

Association of anorectal malformation with duplicated colon, sacral meningocele and scrotal anomalies

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Correspondence to Dr Sunita Singh, drsunitasingh28@gmail.com**Summary**

A 3-day-old male baby presented with anorectal agenesis, sacral meningocele (MMC), bifurcated scrotal raphae, abdominal distension and septicaemia. X-ray in the prone position cross table view showed high anorectal malformation. On exploration for colostomy, an 8 cm terminal (sigmoid and descending colon) colonic tubular duplication was found. Both the colonic segments were communicating proximally to each other and distally with urinary bladder, along with complete anorectal agenesis. The excision and repair of the colovesical fistula were done. The colonic segment communicating proximally to the normal colon was exteriorised by window colostomy. The high-resolution ultrasonography of the scrotum revealed two well-developed testis. The bifurcated scrotal raphae and sacral MMC were left to be treated in the future. Unfortunately, the baby died in the immediate postoperative period due to ongoing sepsis.

BACKGROUND

The neural tube defects may be associated with thoracic and upper gastrointestinal tract anomalies. We present a case where terminal tubular colonic duplication was associated with sacral meningocele (MMC). It also had additional features of bifurcated scrotal raphae at the mid-scrotum to demarcate the third sac in the scrotum. The duplication of lower genitourinary tract may be associated with the duplication of lower gastrointestinal tract; however, triseptated scrotal sac had never been reported in association with colonic duplication. The other unique feature of the baby was that both the duplicated and normal colons were communicating with the bladder, along with complete anorectal agenesis. To the best of our knowledge, this is the first case having complete anorectal agenesis, tubular colonic duplication, sacral MMC and triseptated scrotum.

CASE PRESENTATION

A 3-day-old full term male baby, weighing 3.2 kg, was referred to us with absent anal opening; single, midline lower sacral cystic transilluminant swelling; and bifurcated median scrotal raphae (figure 1). On examination, the pulse rate was 170/min and respiratory rate was 40/min. There was intercostal muscle retraction. The abdomen was distended and perineum was flat. A single midline, painless, non-tender, soft cystic, transilluminant sacral swelling of normal temperature was present. The swelling neither showed cross fluctuations nor any cry impulse. No other obvious congenital anomaly was noticed in the baby.

INVESTIGATIONS

Blood investigations were normal. X-ray in the prone position cross table view showed high ARM. The plain x-ray of the abdomen in erect posture did not reveal any specific abnormality other than absence of gas in the pelvis. The high resolution ultrasonography (HRUSG) of scrotum

revealed two well-developed testis, out of which the left testis can be pulled into the inferiorly midline placed scrotal sac. The findings of the sacral swelling on HRUSG were consistent with MMC (MMC). The ECG, abdominal ultrasonography, MRI of head and spine were done for VACTERL and currarino's triad, and were found to be normal.

TREATMENT

The baby was resuscitated with intravenous fluids. The nasogastric intubation was done for decompression of the stomach. Injections of ceftriaxone (50 mg/kg intravenous 12 hourly), amikacin (7.5 mg/kg intravenous 12 hourly) and metronidazole (1.5 mg/kg intravenous 8 hourly) were administered. Preoperatively, at the first stage, a colostomy for high ARM was planned. On exploration, an 8 cm terminal tubular colonic duplication was found. Both the colonic segments were communicating proximally to each other and distally with urinary bladder, along with complete anorectal agenesis (figure 2). The excision and repair of the colovesical fistula was done. The colonic part, which was in continuity with the normal transverse colon, was exteriorised by window colostomy.

OUTCOME AND FOLLOW-UP

Unfortunately, because of the ongoing sepsis, the baby died on the second postoperative day.

DISCUSSION

In 1937, Ladd coined the term duplication of the alimentary tract.¹ A duplication cyst should have at least one outer muscular layer lining with varying types of gastrointestinal mucosa and intimate anatomic association with the some portion of gastrointestinal tract.¹ The gastric mucosa can be found in over 50% of duplications including the colon and rectum.² The duplications can be cystic or tubular.^{2 3} The most common site for duplication is the ileum.^{1 2} It

Figure 1 Photograph of the perineum showing absent anal opening, sacral meningocele and bifurcated scrotal raphe containing left testis.

Figure 2 Duplicated terminal colon; note both were communicating distally with bladder.

may also be found in the oesophagus, duodenum or stomach. The colonic duplications are very rarely reported and represent about 5–6% of intestinal duplications.^{3–5} Only 75 tubular duplications of the colon have been described

in the literature, out of which most cases occurred mainly in adults.^{6 7}

Tubular colonic duplications usually depict specific criteria: communication with the normal bowel, rare mucosal

inclusions of gastric mucosa islets and frequent association with other malformations (vertebral and genitourinary anomalies).²⁻⁵ The proposed embryonic theory to explain this association is the split-notochord theory by Bentley. According to this, developmental anomalies of the neuroenteric canal (connecting the yolk sac and the amnion) are responsible for associated spinal defects.¹ During development of the split notochord, an endodermal-ectodermal adhesion between the cord leads to the persistence of an endomesenchymal tract between the yolk sac and the amnion. This endomesenchymal tract formed is responsible for the anomalies of the entire gastrointestinal system.⁸

Associated anomalies which might be indicative of internal duplications are two anal dimples (two imperforated anus), one functioning ani with additional anal dimple (imperforated anus) and cutaneous fistulae as previously reported in literature. The findings that strongly suggest a long segment colonic duplication include two functioning perineal ani or the presence of a rectovaginal, rectovulvar or rectoperineal fistula, plus a midline or off-midline perineal anus in a female child. In a male child, the presence of one functioning perineal anus and a rectourinary fistula with or without an additional anal dimple should be diagnostic. The tubular duplication is often associated with double vagina and uterus, double panis, duplicated bladder and duplicated urethra,⁹ but in our case it was associated with triseptated scrotum, which has not been mentioned in the literature.

Various radiological signs that may also suggest a diagnosis of colonic duplication are the duplication of the vertebral column, hemivertebrae or separation of the symphysis pubis.¹⁰ In our case, it was associated with sacral defect with MMC. In the literature, cases of anal canal duplication with internal sacral MMC have been described, but no case of colonic duplication with sacral defects has ever been mentioned.¹⁰

Most colonic duplication cysts remain asymptomatic and may not be diagnosed until adulthood, however, when symptomatic, abdominal pain is the most common presentation.¹¹ Other rare presentations are obstruction, volvulus and bleeding.^{2-5, 11} These duplicated colonic cysts may be isolated or have an external fistula to the skin, urinary tract or normal colon. Malignant degeneration of the duplication cyst is a rare occurrence.¹²

The best treatments for long tubular duplications are resection of duplicated colon due to shared blood supply of the native and duplicated bowel.¹⁻⁶ Furthermore, if the duplicated part is communicating with genitourinary tract, it should also be separated as due to diversion of urine through fistula in duplicated colon there is risk of hyperchloremic metabolic acidosis with uremic sepsis.¹⁻¹³ Keeping in mind the above factors, we had carried out excision and repair of fistula. Other authors had exteriorised both segments of duplicated colon as adjacent osteomy, but we have done single window colostomy in the part of duplicated colon, which was more in alignment with the normal transverse colon. The advantage of this procedure was that the adjacent duplicated colonic segment was left undisturbed and can be utilized in the definitive procedure so that maximum native colon can be used to gain

the colonic length.¹⁴ Other techniques used to preserve the colonic length are dividing. Other techniques used to preserve the colonic lengths are dividing the septum to convert it into common colonic channel, long side-to-side anastomosis of adjacent normal colon and duplicated bowel, mucosal stripping etc.^{1-8, 14} In cases where duplicated colon is fused with the main colon and a common muscular wall exist with cautious identification of the relationship of the vascular supplies to the two bowels, the duplication can be completely excised by leaving the common muscular layer to the main colon.¹⁴ For cystic duplication or very short tubular duplications, resection of duplicated colon and terminal end pull through for anorectoplasty can be done.¹⁴

Learning points

- Terminal colonic duplications like thoracic and upper abdominal duplications can also be associated with spinal defect. So babies with terminal colonic duplications should be examined for the occult spinal anomalies.
- If anorectal agenesis is associated with duplicated colon, every effort should be made to save the native colon.
- Ligation of colovesical fistula is an essential procedure in the first stage itself to prevent lethal complications.

Competing interests None.

Patient consent Obtained.

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