Cystic rectal duplication in infants: A case report and review of literature

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ABSTRACT

Rectal duplication is described in a 1-month-old male infant who presented with constipation for the last 5 days. A presence of a cystic mass in relation to posterior wall of rectum revealed on clinical and radiological examination provided a clue to diagnosis. The clinical presentations and the management protocol of this rare entity are discussed. Authors review their experience with this rare entity and the pertinent literature.

KEY WORDS: Infants, intestinal obstruction, rectal duplication

INTRODUCTION

As described by Ladd and Gross, alimentary tract duplications are rare congenital anomalies characterized by a well-developed muscle coat, epithelial lining and attachment to a portion of gastrointestinal tract.[1] Among gastrointestinal duplications, rectal duplications form a small subgroup, with less than 100 cases having been reported in literature till date.[1,3,5-8] But surprisingly, a detailed review of cystic rectal duplication in infants has rarely been reported in Indian literature. Thus, recognition of its varied clinical presentations and the subsequent management protocol is mandatory so as to avoid undue morbidity and mortality associated with delayed diagnosis and treatment. Thus, rarity justifies reporting of the present case with the aim to make physicians aware of the different clinical presentations of this rare entity, thus achieving a satisfactory recovery with an early surgical intervention.

CASE REPORT

A 1-month-old male infant presented with increasing constipation for the last 5 days. The other associated gastrointestinal and urinary symptoms were absent. On examination, the baby was alert, active, feeding vigorously and had no other external congenital anomalies. The general physical and chest examination revealed no abnormality. The abdominal examination revealed a soft, nontender abdomen with normal bowel sounds. Digital rectal examination suggested a nontender, firm cystic mass in relation to posterior rectal wall with mobile overlying rectal mucosa. The neurological and genitourinary examination was normal.

Laboratory investigations revealed Hb. 10 gm%, TLC 9000/mm$^3$ and normal renal and liver function tests. The plain roentgenogram of abdomen and spine suggested no abnormality. Radiological evaluation by contrast enhanced CT scan suggested a well-defined cystic mass posterior to rectum in the presacral space. This precipitated surgery, and surgical excision was planned through posterior sagittal route. Surgical exploration revealed a large cystic mass with a well-defined muscle coat in relation to posterior wall of lower one-third of rectum. After decompression by aspiration of clear fluid, complete excision of mass became feasible. The mass had a narrow communication with rectum, which was repaired subsequently. Histopathology of resected specimen showed presence of columnar epithelial lining with a well-developed muscle coat, thereby confirming diagnosis of rectal duplication. The postoperative period remained uneventful. The child has been asymptomatic for the last 8 months and is having normal anorectal functions.

DISCUSSION

Rectal duplication is an extremely uncommon congenital anomaly in infants.[1,3,5,7] A well-defined muscle coat,
epithelial lining and an attachment to rectum makes this duplication a true gastrointestinal duplication.[1,2] Rectal duplications constitute less than 2% of all the gastrointestinal duplications, thus demanding recognition of their clinical presentation so as to avoid complications and hence undue morbidity.\[^{[1,3,7]}\] The embryogenesis of rectal duplications is unclear, but still the theory of embryonic diverticulum formation can be considered to explain it.\[^{[1,4,5]}\] Similarly, the pinching off of diverticulum from the posterior wall of developing rectum explains the embryogenesis of rectal duplication in the present case. But still the theory of partial twining has been proposed by a few authors to explain its embryogenesis.

The clinical presentation of rectal duplications is usually variable and depends upon its size, fistulation, infection, presence of ectopic gastric mucosa and malignant degeneration.\[^{[1]}\] Clinically, rectal duplications are commonly present in presacral space posterior to the rectum, protruding either into rectum or into perineum as spherical or elliptical cystic lesions filled with clear fluid. The presence of perianal swelling, perineal fistula, presacral mass, intestinal prolapse as ‘polyp,’ bleeding per rectum and lower gastrointestinal obstructing symptoms due to mass effect are the common clinical presentations.\[^{[1,4,8]}\] Similarly, the mass effect resulting from the presence of a large duplication explains the clinical presentation in the present case.

The diagnosis of rectal duplication is usually clinched on a thorough clinical evaluation and a vigilant digital rectal examination. But still a number of clinical conditions like anterior sacral meningocele, teratoma, dermoid cyst, chordoma, etc., provide a diagnostic challenge and hence need a preoperative clinical differentiation.\[^{[1]}\] Moreover, the presence of associated genitourinary, skeletal, cardiovascular anomalies demands a thorough evaluation in patients with rectal duplications.\[^{[2]}\] Thus, a radiological evaluation, which may include ultrasonography, CT scan, MRI and contrast roentgenogram, is required depending upon the clinical presentation and the available resources.\[^{[2,3,5]}\] Similarly as experienced in the present case, CT scan appears to be the diagnostic modality of choice both for confirming the diagnosis and for preoperative anatomical assessment.

The treatment of rectal duplication is primarily surgical.\[^{[1,4,6]}\] The treatment aims at complete excision with preservation of anorectal function.\[^{[1,5]}\] An early surgical intervention is recommended so as to avoid complications like bleeding, infection and malignant degeneration.\[^{[1,4]}\] The various surgical approaches recommended for rectal duplications include 1) transanal, 2) transsccygeal (Kraske) and 3) posterior sagittal.\[^{[1,5,6]}\] But as experienced in the present case, the posterior sagittal approach provides the best surgical access, thus facilitating complete excision of rectal duplications with preservation of anorectal function.

Thus we conclude that although occurrence of rectal duplication is rare, a thorough clinical evaluation should be done in infants presenting with constipation with the aim to diagnose its presence at the earliest. We also recommend an early complete surgical excision through posterior sagittal route in such cases so as to avoid resulting complications and hence undue morbidity.

**REFERENCES**