Segmental Ileal Dilatation Presenting with Neonatal Intestinal Obstruction

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Submitted: 20-Aug-2019.
Revised: 12-Oct-2019.
Accepted: 04-Jan-2020.
Published: 01-Sep-2020.

ABSTRACT

Segmental ileal dilatation is an uncommon cause of neonatal intestinal obstruction. This report highlights a rare combination of abnormal distribution of muscles in the muscularis propria and partial loss of interstitial cells of Cajal as causative factors for segmental intestinal dilatation.

KEYWORDS: Interstitial cells of Cajal, muscularis propria, neonatal intestinal obstruction, segmental ileal dilatation

INTRODUCTION

Segmental intestinal dilatation (SID) is a rare entity. Children can have a spectrum of clinical presentations, with nearly 50% presenting in neonatal age.1 Diagnosis is seldom anticipated preoperatively, and surgery is the definitive treatment modality. Herein, we describe a rare case of intestinal obstruction in a newborn to highlight the etiopathogenesis contributing to its occurrence.

CASE REPORT

A preterm (33-week gestational age) female baby, weighing 1720 g, was born through normal vaginal delivery. She passed meconium and urine on day 1 of life. The child developed episodes of bilious vomiting starting from day 3 of life. It was associated with a history of gradually increasing abdominal distension. On examination, the abdomen was grossly distended with palpable bowel loops, and bowel sounds were exaggerated. While the serum electrolytes and sepsis screen were negative, erect X-ray of the abdomen revealed multiple air-fluid levels.

Emergency laparotomy performed on day 4 of life, revealed segmental ileal dilatation of the proximal ileum, measuring 8 cm × 4 cm in dimensions. The rest of the bowel was healthy, and distal atresia was ruled out. Resection of the dilated segment with end-to-end anastomosis was performed [Figure 1].

The child made an uneventful recovery and was discharged on postoperative day 7. At present, she is under regular follow-up and achieving milestones according to her age. Histopathological examination revealed anatomic malformation of the muscularis propria with reversal of inner circular and outer longitudinal muscular layer [Figure 2a-f]. There was associated desmosis or loss of fibrous muscular sling [Figure 2d], and partial loss of interstitial cells of Cajal (ICC) [Figure 2f, g and h].

DISCUSSION

Segmental intestinal dilatation, first coined by Rossi and Giacomoni, presents as a neonatal intestinal obstruction in nearly half of the cases.2 Remaining cases usually present during infancy and childhood with abdominal pain, failure to thrive, and anemia. There is a slight preference for the male gender, and ileum is the most frequent site involved.3 The exact incidence of SID is not reported yet in literature. However, there are around 150–200 published cases of segmental dilatation of the gut in the literature. Reports on microanatomical abnormality causing SID are extremely rare, and only 15 cases are found in the English literature. Among these reports, those describing the reversal of layers of muscularis propria are even rarer.1,3-5

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How to cite this article: Anand S, Dhua AK, Sankar J, Das P, Goel P, Bajpai M, et al. Segmental ileal dilatation presenting with neonatal intestinal obstruction. J Indian Assoc Pediatr Surg 2020;25:306-9.
In 1959, a diagnostic criterion was established for segmental dilatation by Swenson and Rathauer. It includes limited bowel dilatation up to 3–4 times the rest of bowel, an abrupt transition between the normal and ectatic segment, no distal obstruction (intrinsc or extrinsic), clinical features fitting into an intestinal obstruction, normal neuronal plexus, and complete recovery after resection of the dilated segment.[6] Diagnostic criteria for the reversal of layers of muscularis propria are complex and should be left to expert pathologists, as they should be sure about the orientation of intestinal segments and be aware of how the sections were taken, circumferentially or longitudinally to the gut axis. As shown in Figure 2a and c, on sectioning parallel to the gut axis, the layers appeared just reversed in the dilated intestinal segment, versus the normal segment represented in Figure 2b. ICC is extremely labile and can be lost secondarily in any other anatomical or functional disorders of the neuromuscular gut structure. Loss of continuity of CD117-positive spindle cells (ICC) surrounding the intermuscular nerve plexus, as well as the loss of these positive interspersed CD117-positive spindle cells in inner circular or outer longitudinal muscle layers, would define mesenchymopathy or loss of ICC. Desmosis is the loss of fibrous sling backbone which supports the muscularis propria. It can be a primary disorder or can be totally or partially lost in other neuromuscular disorders affecting intestine, seen in postabdominal surgery and secondary to serositis. These latter disorders in conjunction with the primary neuromuscular defects can contribute to gut aperistalsis or dilatation.[7]

Among the clinical differential diagnoses of SID, duplication cysts can closely mimic SID. Duplication cysts are contiguous with normal bowel and have a common muscular coat and blood supply. The lumen may (tubular duplications) or may not (saccular) be connected with the lumen of the normal bowel. There is a stark difference in the embryology of the SID versus duplication cyst. Duplications are related to the period of transition of the embryonic intestine from a solid to a

Figure 1: Intraoperative findings showing the segment of ileum disproportionately dilated in comparison to rest of the bowel. Resection (along the dashed white line) and anastomosis were performed.

Figure 2: Photomicrograph shows a segment of the oriented small intestine with complete reversal of the inner circular muscle (blue arrow) and outer longitudinal muscle layers (black arrow) (a, H and E, ×40). Sections from other areas of the segmentectomy specimen showed normal orientation of the interstitial cells of Cajal muscle (blue arrow) and outer longitudinal muscle (black arrow) layers in the index patient (h, ×100). (c) Also shows the reversal of the interstitial cells of Cajal (blue arrow) and outer longitudinal muscle layers (black arrow). Ganglion cells are normally present (c, ×200). Sirius Red stain shows complete disruption of the fibrous muscle sling in the muscularis propria (arrows) (d, ×100). Smooth muscle actin and desmin stain show normal staining pattern on the intestinal segment with reversal of the interstitial cells of Cajal and outer longitudinal muscle layers (e and f, ×100). (g and h) show marked paucity of the CD117-positive spindled interstitial cells of Cajal near the myenteric plexus. Only an occasional interstitial cells of Cajal are seen (arrows) (h and f, ×200)
hollow tube structure, which results from the coalescence of the vacuoles. There is a presence of septum in duplications, while SID shows none of the above features. Another close differential of SID is Meckel’s diverticulum. SID is a focally dilated segment along the long axis of the bowel; however, Meckel’s diverticulum is a focal diverticulum along the anti-mesenteric or mesenteric (rare) location. Some authors have also mentioned it as a giant Meckel’s diverticulum due to similarities, such as the presence of heterotopic gastric or pancreatic mucosa in both. Furthermore, finding muscularis mucosae, both inner circular and outer longitudinal layers of muscularis propria in diverticulum, is not usual. Either the pseudodiverticula shows only the presence of muscularis mucosae or a true diverticula like the Meckel’s diverticula shows outer longitudinal muscle layer only.

The exact etiopathogenesis of SID is yet to be elucidated. Various theories have been postulated for its occurrence. It includes extrinsic compression of the fetal bowel by omphalomesenteric bands, intrauterine vascular accidents, congenital damage of the myenteric plexus, vacuolization of smooth muscle suggesting myopathy, and the presence of heterotopic mucosa causing disorganization of the neuromuscular peristaltic complex. Other rare reasons for its occurrence include anatomic abnormalities in muscularis propria and deficiency/loss of ICC. Quite interestingly, we found both of them in the index case, in the form of reversal of inner circular and outer longitudinal muscular layers and partial loss of ICC. In human embryo, the annular differentiation of layers of muscularis propria from homogeneous mesenchymal layer starts between 7 and 9 weeks of gestation. This differentiation starts rostrally and proceeds caudally. During the same time, the neuroglial precursors advance up to the muscle layers and form ganglia of myenteric plexus. In contrast to that in mice, in human, both muscle layers are formed simultaneously. During this development, the developing intestine progresses to the lower part of the abdomen, which latter again regresses upward. This rostro-caudal migration of developing gut produces various enteric landmarks (e.g., sphincters and cecum) and is dictated by various transcription factors and genes. Not only during the development only vertical migration takes place but also the rotation of the gut along Z-axis takes place. Among various genes, Sonic hedgehog produced by the endoderm and bone morphogenetic proteins produced by adjacent mesenchyme are crucial to result in the radial differentiation of enteric mesenchyme and distribution of myenteric ganglion cells. Studies on the experimental model have shown that mutations affecting Sonic hedgehog signaling disturb the radial organization of the gut wall and distribution of ganglion cells. Although very intriguing, our knowledge is limited. There can be a candidate gene mutation resulting in the reversal of layers of muscularis propria in the index patient, or, the malrotation during complex development process may have resulted in the segmental reversal of the layers of muscularis propria and segmental intestinal dilatation in the index patient. Identification of such cases, followed by further workup, can only establish a confident hypothesis.

As already stated, till 2013, only three reports were highlighting the partial loss or absence of ICC in cases of segmental intestinal dilatation. Sakaguchi et al. reported a case of segmental gut dilatation and highlighted the lack of normal peristalsis is due to the loss of ICC as its possible cause. They also emphasized that a multitude of histopathological findings suggest different subtypes of segmental dilatation. Architectural anomalies of muscularis propria including supernumerary muscle coats and irregular orientation are known, but a reversal of muscular arrangement (circular muscle layer was outer to longitudinal layer) has not been reported earlier. Furthermore, identification of this abnormality warrants proper grossing and orientation of the intestinal sections, as improperly oriented sections can erroneously show a reversal of layers of muscularis propria. Typical arrangement of the overlying mucosal crypts, myenteric and submucosal neuronal plexus supported the identification of segmental anatomic malformation of the muscularis propria in the index case. Although the presence of the anatomical defect of the muscularis propria is possibly the primary pathology, in this case, loss of ICC and partial desmosis can be secondary to the marked dilatation of the bowel segment. ICC and the fibrous sling present supporting the muscularis propria are very labile structures and can be affected in other primary pathologies. It is also worth noticing that the child made an uneventful recovery after the surgery, denoting that the distribution of ICC and arrangement of the muscles would have been normal in the remaining bowel.

In most of the cases, segmental dilatation is detected incidentally during surgery. However, preoperative plain radiograph and contrast studies can also provide subtle signs for its diagnostic suspicion. A grossly dilated bowel loop with or without air-fluid level on X-ray abdomen and stasis of contrast with marked segmental dilatation on follow-through study are distinguishing features. Definitive treatment is surgical resection with end-to-end anastomosis of the remaining bowel.
CONCLUSION
Segmental intestinal dilatation is a rare cause of neonatal intestinal obstruction. The loss of ICC and abnormal distribution of layers in muscularis propria can contribute to its occurrence. Surgical resection and end-to-end anastomosis are curative.

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.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

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