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Case Report

Delayed presentation of congenital sigmoid colon stenosis: a rare entity

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Abstract

Congenital sigmoid colon stenosis is a rare entity that can mimic Hirschsprung disease. Presentation of congenital colon stenosis is usually within first few weeks of life. Our case presented with features of distal bowel obstruction at 2 years of age with the history of chronic constipation and progressive abdominal distention from first week of life and bilious vomiting for the last 1 week. Clinical diagnosis of Hirschsprung disease was made. Contrast enhanced CT abdomen showed bowel obstruction with transition point at the level of proximal sigmoid colon. There was a short segment stenosis at the level of proximal sigmoid colon. Contrast enema showed stenosis at proximal sigmoid colon. The bowel distal to stenosis was normal in calibre. Similar findings were seen during surgery. Mesocolon was present in stenosed segment of the bowel. The resected stenotic segment showed adequate ganglion cells in histopathology.

Keywords: barium enema; contrast enhanced CT; ganglion cells; Hirschsprung disease; stenosis.

Introduction

Congenital intestinal stenosis is a condition in which there is presence of a short, rigid portion of bowel with persistent reduced distensibility connecting a grossly dilated proximal bowel loop and a normal calibre distal loop. The stenosis being present since birth. It differs from colonic atresia in that there is presence of minimal patent lumen and mesentery.[1] Congenital colonic stenosis is a very rare entity with only few cases reported till date. The reported incidence is 1/40 000 birth.[2] Congenital sigmoid colon stenosis mimics Hirschsprung disease. In Hirschsprung disease, there is complete aganglionosis of bowel segment distal to stenosis and is usually seen in rectum and anorectal junction.[3]

Clinical presentation

A 2-year old male child presented to the emergency department of a tertiary referral hospital with the history of chronic constipation and progressive abdominal distention from first week of life and bilious, projectile vomiting for 7 days. On examination, the abdomen was grossly distended with visible superficial veins and was tympanic on percussion. Weight of the child was 10.1 kg, had signs of dehydration. The child had mild hyponatremia ($Na^+ = 131$ mEq/L and hypokalemia ($K^+ = 3.4$ mEq/L). The case was clinically diagnosed as Hirschsprung disease and planned for surgery.

Imaging

X-ray abdomen (erect) showed dilated small and large bowel loops with few air fluid levels. The bowel loops in pelvis were relatively small in calibre. A metallic density object was seen within a bowel loop in left lower abdomen which was likely ingested foreign body (Figure 1).

Contrast enhanced CT abdomen showed grossly dilated bowel loops with stricture at the level of proximal sigmoid colon and normal calibre rest of sigmoid colon and rectum (Figure 2).

Contrast enema showed a short segment persistent reduced distensibility at proximal sigmoid colon and failure of reflux of contrast proximal to the segment. Rest of sigmoid colon, rectum, and anal canal were normal in calibre (Figure 3).

Differential diagnosis

With the clinical history, the possibility of Hirschsprung disease was considered. After imaging, possibility of congenital sigmoid colon stenosis was also considered. Other differential diagnosis can be acquired stenosis of sigmoid colon.

Case management and treatment

Adequate fluid and electrolyte resuscitation was done in emergency. Preoperative investigations were done. Next day, the child was taken for surgery. Laparotomy was done which showed similar finding (Figure 4). The stenosis was seen at the level of proximal sigmoid colon. Mesocolon was present in stenosed segment. The stenosed segment was resected and double barrel colostomy was made. The resected segment was sent for histopathological examination which showed non-specific inflammation and adequate ganglion cells. Presence of ganglion cells ruled out the possibility of Hirschsprung disease and diagnosis of congenital sigmoid colon stenosis was made. Secondary end to end colo-colic anastomosis was done after 12 weeks. The postoperative period was uneventful. The child was doing well at the first follow up after colostomy closure (after 1 month). The child was

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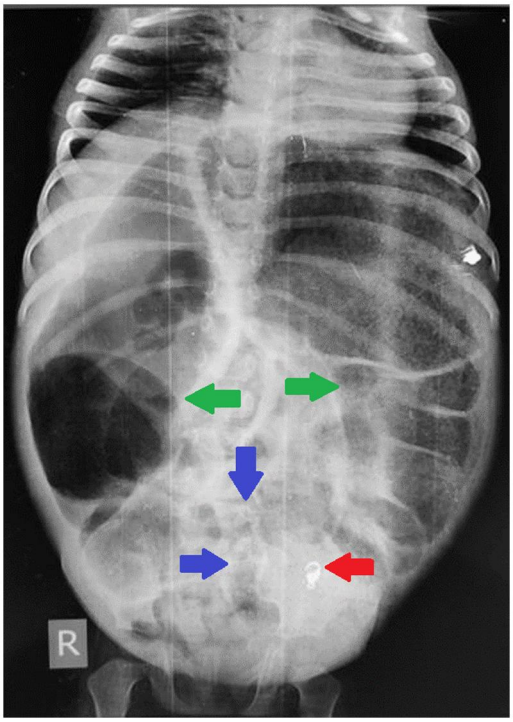

Figure 1. X-Ray abdomen (Erect) of the child shows grossly dilated bowel loops (green arrows). Bowel loops in pelvis are normal in calibre (blue arrows). A metallic density foreign body is also seen in left lower abdomen (red arrow).

defecating once daily and the consistency of stool was semi-solid. There was no abdominal distention.

Discussion

Congenital or acquired bowel stenoses/atresias are common cause of intestinal obstruction in paediatric population. They occur commonly at the level of jejunum or ileum. Although the exact incidence is unknown, colonic atresias/stenoses account for 1.8%-15% of all bowel atresias.[4] Hirschsprung disease is a common cause of bowel obstruction in newborn and children. Congenital colonic stenosis is, however, a rare cause of intestinal obstruction with only few cases reported till date.[4] There are several theories proposed as cause of congenital bowel stenosis. Some of them include segmental *in vitro* ischaemic insults due to embolus/intrauterine vascular insufficiency; use of vasoconstrictor drug during pregnancy; intrauterine cytomegalovirus (CMV) or Varicella infection; and overexpression of transforming growth factor (TGF)-beta in a segment of colon.[5] Most commonly involved colonic segment in congenital colonic stenosis is ascending and transverse colon.[2] Patients present with features of low bowel obstruction with failure to thrive, dehydration, and electrolyte imbalance. The presentation may be early in neonatal period or late as in our case. The presentation mimics Hirschsprung disease and maybe misdiagnosed. Imaging findings in X-Ray are dilated bowel loops with air fluid level proximal to stenosis and absence of air distal to stenosis. Stenosed segment is seen in CT with grossly dilated bowel loops proximal to stenosis and normal calibre bowel loops

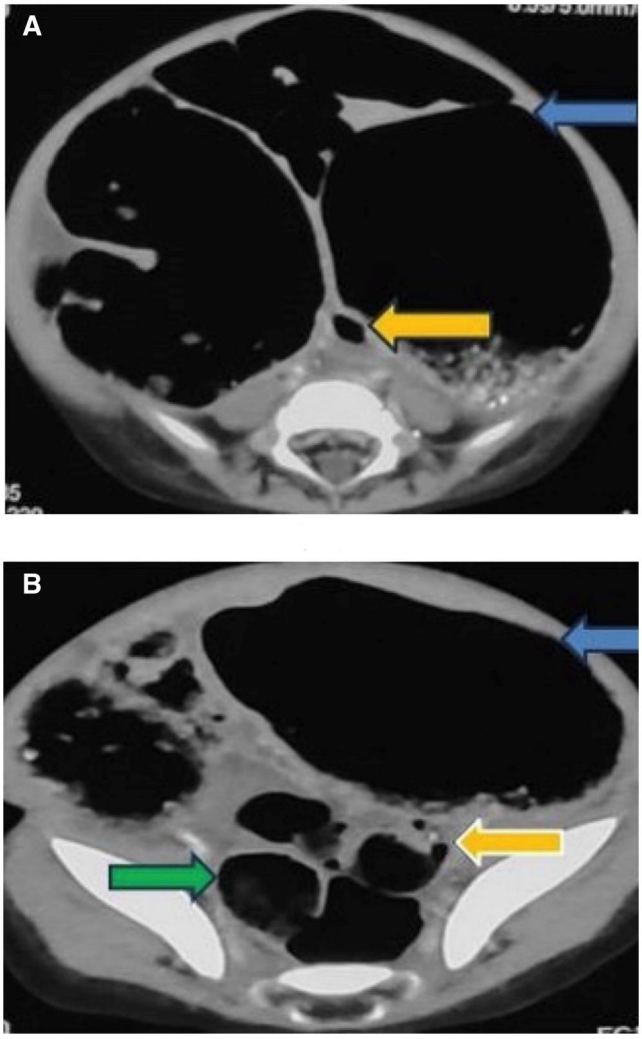

Figure 2. Contrast enhanced CT Images of abdomen. In (A, B), stenosis is seen in proximal sigmoid colon (yellow arrow). Bowel proximal to the stenosis (Blue arrow) is dilated and filled with gas and faecal matter. Bowel distal to the stenosis (green arrow) is normal in calibre.

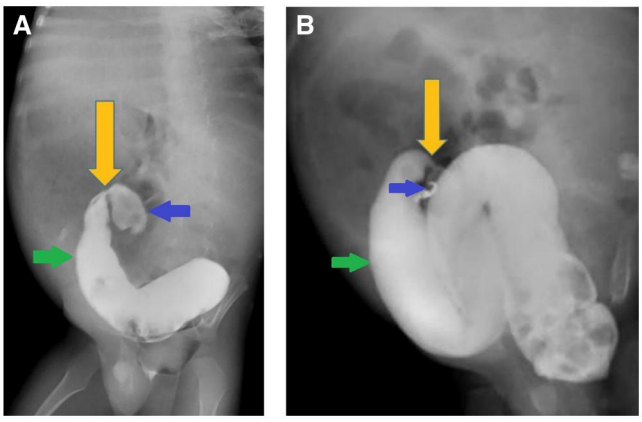

Figure 3. Image (A) is AP view of contrast enema, (B) is oblique view of contrast enema. Stenosis is seen in proximal sigmoid colon (yellow arrow). Rest of sigmoid colon, rectum, and anal canal are normal in calibre (marked with green arrow). There is reflux of contrast proximal to stenosis (marked with blue arrow).

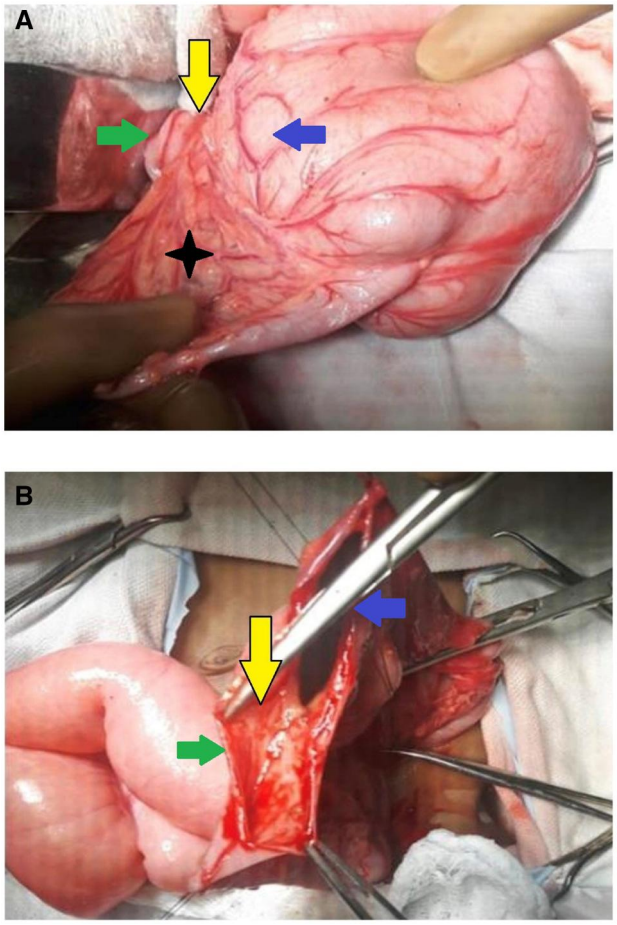

Figure 4. Intraoperative pictures of congenital sigmoid colon stenosis. In (A) stenosis is seen at transition point (yellow arrow) in proximal sigmoid colon. Bowel proximal to the stenosis is dilated (blue arrow) and distal to stenosis is normal in calibre (green arrow). Mesocolon is present in the stenosed segment (marked with black star). In (B), minimal patent lumen (yellow arrow) is seen in the stenosed part. The bowel proximal to stenosis is decompressed (blue arrow) and bowel loop distal to the stenosis is normal in calibre (green arrow).

distal to stenosis. Contrast enema shows persistent reduced distensibility at stenosed segment and failure of reflux of contrast proximal to the stenosis. Normal calibre bowel loops are seen distal to the stenosis. In histology, presence of adequate ganglion cells in the stenosed segment rules out the possibility of Hirschsprung disease.[3] One may argue our case might be a case of acquired stenosis due to late presentation. But, presence of symptoms from first week of life suggests congenital rather than acquired cause. Delayed presentation of congenital stenosis has also been reported by Gupta et al,[5] Saha et al,[6] and Natarajan et al.[7] Further, no obvious history of perinatal/neonatal infection or necrotizing enterocolitis is present, which are the common cause of acquired stenosis. Delayed presentation in our case might be because of location (ie, distal colon) of the stenosis. Surgery is the mainstay of treatment for congenital sigmoid colon stenosis. The surgical implication of differentiating between congenital sigmoid colon stenosis and Hirschsprung disease is that the distal bowel, which has normal ganglion cells, can be preserved in congenital sigmoid colon stenosis, and only a short segment resection would be sufficient. In Hirschsprung disease, the entire distal bowel with aganglionosis should be resected, and an anastomosis with the anal canal should be performed. Preoperative differentiation of these two cases helps in proper surgical planning.

Learning points

- Congenital colonic stenosis may have late presentation.
- Clinically, congenital colonic stenosis mimics Hirschsprung disease and maybe easily misdiagnosed.
- There is presence of mesocolon and minimal patent lumen in colonic stenosis and these features differentiate colonic stenosis from colonic atresia.
- Presence of ganglion cell in stenosed segment is the hallmark differentiating feature in histology between colon stenosis and Hirschsprung disease.
- Preoperative differentiation of congenital sigmoid colon stenosis from Hirschsprung disease is essential for proper surgical planning.

Funding

None declared.

Conflicts of interest

None declared.

Consent

The parents have signed consent form for publication.

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