CASE REPORT

Segmental dilatation of small bowel presenting with severe anaemia: a diagnostic puzzle

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SUMMARY

A 7-year-old boy presented to Paediatric outpatient with worsening lethargy and tiredness. On examination he had extreme pallor. Blood investigations confirmed severe iron deficiency anaemia. He was started on iron supplements and received blood transfusion. However, the response to iron treatment was suboptimal, he therefore underwent extensive workup for the cause of iron deficiency anaemia. The barium meal showed dilated segments of ileum with two distal stenoses. The surgical resection of the involved segment was performed with end to end anastomosis. Histology of the resected segment was inconclusive of inflammatory bowel disease, malignancy or vascular malformation. The child has remained well since surgery with no further blood transfusion or iron therapy.

BACKGROUND

Segmental dilatation of intestine is a rare structural abnormality with unknown aetiology and varied clinical presentation. It is characterised by localised dilatation of the intestine in the absence of intrinsic or extrinsic obstruction. In our knowledge so far 125 cases have been reported in the literature.¹ Both radiological and histopathological findings are non-specific. Definitive management is surgical resection of dilated segment with end to end anastomosis. All reported cases have a favourable outcome after surgical intervention.¹ We describe a case of a 7-year-old boy with segmental dilatation of the intestine presenting with severe anaemia.

CASE PRESENTATION

A 7-year-old boy presented with a 2-month history of worsening lethargy and tiredness. Parents reported a recent decrease in his appetite and intermittent vomiting. Vomiting was non-projectile and non-bilious. They also thought that he had lost some weight. There was no significant medical history.

Examination revealed extreme pallor, tachycardia and a soft systolic murmur. Blood investigations confirmed severe iron deficiency anaemia: haemoglobin (Hb) 32 g/L (normal 115–140 g/L), mean corpuscular volume (MCV) 61 fL (normal 77–95 fL), reticulocyte count $109 \times 10^9/L$, iron 2 $\mu\text{mol/L}$ (normal 10–30 $\mu\text{mol/L}$), ferritin 1 $\mu\text{g/L}$ (normal 15–300 $\mu\text{g/L}$) and transferrin saturation 2.2% (normal 20–50). His vitamin B₁₂ and folate levels were normal and coeliac serology was negative.

He was transfused with packed red cells, and commenced on oral iron supplementation. He

required further blood transfusions and iron infusion while awaiting further investigations into the cause of his anaemia.

As further investigation into presumed occult gastrointestinal (GI) blood loss (box), he underwent a Meckel's isotope scan which did not reveal any ectopic gastric mucosa. His upper GI endoscopy was normal and colonoscopy showed a slightly erythematous terminal ileal mucosa, but no bleeding point or blood in the lumen. Histology of biopsies obtained from various sites during both endoscopic procedures showed no abnormalities.

INVESTIGATIONS

Barium meal and follow-through was performed which showed dilated segments of ileum with two distal stenoses (figure 1).

DIFFERENTIAL DIAGNOSIS

Differential diagnosis included all causes leading to iron deficiency: inadequate dietary intake, blood loss from GI tract and malabsorption.

TREATMENT

Laparotomy was undertaken which showed a dilated ileal segment and two strictures (figure 2), approximately 60 cm from the ileocaecal valve. The dilated segment with the stenoses was resected and an end-to-end anastomosis was performed. The resected segment measured 8 cm in length (figure 2). Histological examination showed non-specific mucosal ulceration, inflammation, oedema and fibrosis, with no features of inflammatory bowel disease (IBD), malignancy or vascular malformation. At further exploration no Meckel's diverticulum was found.

OUTCOME AND FOLLOW-UP

Following surgery, the child has remained asymptomatic with normal haemoglobin values and without further need for blood transfusions or iron supplementation. Four months after his operation, repeat investigations showed: haemoglobin 149 g/L, MCV 80 fL and ferritin 45 $\mu\text{g/L}$.

DISCUSSION

Segmental intestinal dilatation is a rare structural abnormality of the intestine. It was first described by Swenson and Rathauer in 1959.² It is characterised by a focally distended segment of bowel with normal afferent and efferent connection, in the absence of any obstruction or deficient

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Box Causes of occult blood loss in school-aged children (>5 years)

- ▶ Juvenile polyp
- ▶ Meckel's diverticulum
- ▶ Inflammatory bowel disease
- ▶ Small bowel diverticula
- ▶ Ileal duplication
- ▶ Infection: *Helicobacter pylori* infestation
- ▶ Haemolytic-uraemic syndrome
- ▶ Henoch-Schoenlein purpura
- ▶ Anorectal fissure
- ▶ Segmental dilatation of intestine

innervations. The transit time of intestinal contents or contrast media through the small bowel is not delayed unless associated with other congenital abnormalities such as exomphalos, malrotation or Meckel's diverticulum.³

Segmental dilatation of the intestine has been described in infants and young children. It has also been described in neonates where it usually presents with sharply demarcated intestinal dilatation and features of abdominal obstruction, when it can be difficult to differentiate it from more common causes of intestinal obstruction at this age, namely intestinal atresia, Hirschsprung's disease and meconium ileus.^{4 5} Older children may present with anaemia, hypoproteinaemia, malabsorption and GI bleeding. The presence of heterotopic gastric mucosa has been described in the dilated small bowel segment.^{6 7}

The aetiology of the condition is still not clearly understood but there are several hypotheses postulated to explain its occurrence. Irving and Lister suggest that the condition may arise from prolonged obstruction of the bowel during fetal life by an external structure which subsequently involutes, especially in cases associated with other GI anomalies.^{1 8} Other authors suggest in-utero vascular accident, neurogenic component or myopathy as possible causes.¹

Figure 1 Contrast follow-through showing dilated ileal segment with distal stenosis (arrow).

Figure 2 Resection specimen of the dilated ileal segment. Note the presence of two stenoses (at the clamps).

Radiological investigation reveals dilatation of the intestinal segment. Histological examination shows normal ganglionic cells and normal bowel structure in the majority of cases. The definitive management is surgical resection of the dilated segment with end-to-end anastomosis which is curative.

The clinical picture in our case was dominated by severe iron deficiency anaemia and non-specific GI signs without overt bleeding. As both a Meckel scan and a contrast study can be false negative, the typical work up for Meckel's diverticulum would be gastroduodenoscopy to rule out upper GI bleeding, followed by explorative laparoscopy. However, as the child did not present with the typical symptoms of visible fresh blood in the stool, the diagnostic pathway described was chosen. Intestinal dilatation with stenosis was then seen on contrast imaging. This finding stopped further diagnostics and led to the surgical treatment. The diagnosis was confirmed on histopathological examination of the resected segment which showed non-specific mucosal ulceration, inflammation, oedema and fibrosis but no evidence of IBD, malignancy or vascular malformation. The mucosal damage and ulceration is presumed to be caused by stasis in dilated segment.⁹

Learning points

- ▶ Iron deficiency anaemia is a symptom, the cause of which should be explored.
- ▶ Thorough history can rule out common causes of anaemia, while other requires detailed investigations.
- ▶ Unexplained iron deficiency anaemia in children require thorough assessment of the entire gastrointestinal (GI) tract as GI tract is the most likely source of occult chronic blood loss.

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