Discordant monozygotic total colonic Hirschsprung's disease presenting with neonatal isolated ileal perforation

Ramnik V Patel,^{1,2} Suzanne Lawther,³ William Alexander McCallion³

¹Department of Paediatric Urology, University College London Hospitals NHS Foundation Trust, London, UK

²Department of Paediatric Urology, Great Ormond Street Children Hospital NHS Trust, London, UK

³Department of Paediatric Surgery, Royal Belfast Hospital for Sick Children, Belfast, UK

Correspondence to

Ramnik V Patel,
ramnik@doctors.org.uk

DESCRIPTION

A full-term monozygotic twin, one presented with failure to pass meconium at 48 h of birth, refusal to feeds and abdominal distention. She was unwell with tachycardia and temperature 38.5°C. Abdomen was distended and tender with absent bowel sounds. She had haemoglobin 75 g/L, white cell count $20 \times 10^9/L$, neutrophils $10 \times 10^9/L$, platelets $593 \times 10^9/L$, C reactive protein 21 mg/L with normal renal, liver and bone profiles. Abdominal and decubitus radiographs showed pneumoperitoneum (figure 1A, B). She underwent exploratory laparotomy, resection of perforation and primary anastomosis uneventfully and was discharged home after 5 days. She presented with poor feeding, vomiting, no weight gain, irritability, constipation and abdominal distention at 2 weeks of age. Abdominal radiograph was normal with gas in the rectum (figure 1C). Urine cultured *Enterobacter*

cloacae. She was treated with antibiotics, blood transfusion and recovered but had constipation and abdominal distention. Rectal suction biopsy confirmed Hirschsprung's disease; her twin sister was normal. She underwent re-exploration with mapping seromuscular biopsies and frozen section biopsy which showed total colonic Hirschsprung's disease and levelling ileostomy was performed uneventfully. She then underwent modified Duhamel pull through procedure at 1 year of age and is thriving well at 2 months of follow-up. Total colonic Hirschsprung's disease presenting as an isolated terminal ileal perforation in the neonatal period as primary presentation is very rare.^{1 2} In a term baby with isolated ileal perforation, primary repair is the usual procedure especially when the monozygotic twin was normal. Discordant Hirschsprung's disease in monozygotic twins is very rare.³

Figure 1 (A and B) Abdominal and lateral decubitus radiographs showing normal stomach and upper small bowel obstructive gas pattern and gasless pelvis and peripheral colonic area and massive pneumoperitoneum suggestive of terminal small bowel perforation in the background of obstructive pattern. (C) Abdominal radiograph at re-admission showing normal bowel gas with gas in the rectum.

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Learning points

- ▶ Hirschsprung's disease should present in both identical twins but can present in one of the monozygotic twins as a rare variant and can be missed if the other twin is asymptomatic and well; discordance may lead to confusion and delay in diagnosis.
- ▶ In isolated perforation of ileum especially with a transitional zone funnelling appearance of bowel beyond the perforation, index of suspicion for total colonic Hirschsprung's disease should be high.
- ▶ If available, frozen section biopsy and levelling ileostomy or mapping seromuscular biopsies and bringing perforation site as stoma may give a correct diagnosis and may help early treatment.

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