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Intestinal perforation in Hirschsprung‘s disease

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Title image: Heinrich Hirschsprung, source Wikimedia Commons

This male infant was born to a 27-year-old G3/P1 at 40 5/7 weeks of gestation with a birth weight of 3850 g. The perinatal period was unremarkable. At the age of 8 weeks, the infant was readmitted because of repetitive vomiting, episodes of crying, pallor, a history of constipation and abdominal distension.

Initial abdominal x-ray, ultrasonography and routine laboratory studies revealed unspecific changes and were thought to be compatible with gastroenteritis. On hospital day 5, vomiting increased and follow-up abdominal X-rays showed a pneumoperitoneum (Fig. 1, 2).

On emergency laparatomy, there was ascites, an appendiceal perforation and a cone-shaped transition zone with a widened ileum, 7 cm proximal to the ileocecal valve. Biopsies of rectum, transverse colon and terminal ileum revealed complete lack of ganglion cells. This was documented on conventional as well as immunological stains (Fig. 3-6). The postoperative course after appendectomy and ileostomy was unremarkable. A two-staged definitive surgical treatment was performed some weeks later.

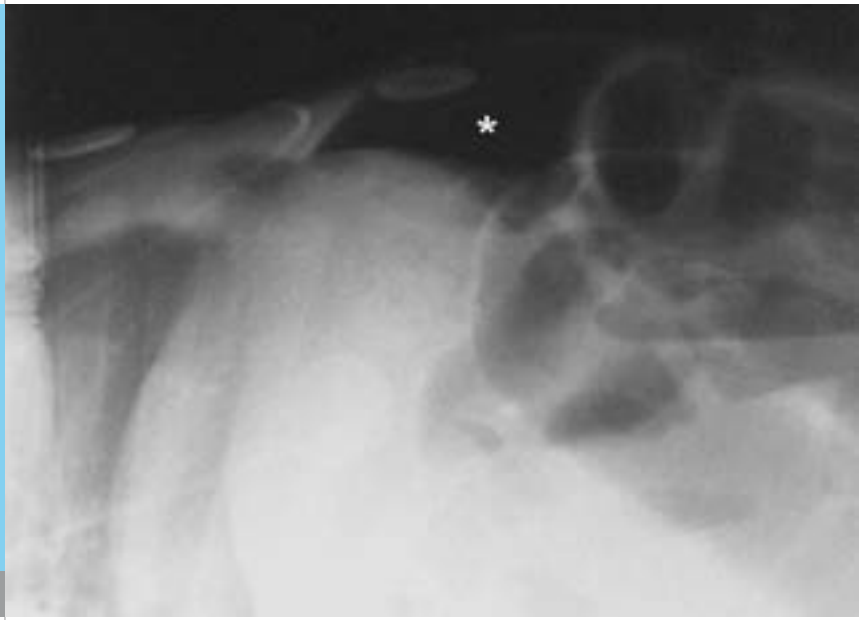

Fig. 1

Abdominal X-ray using a horizontal beam technique: pneumoperitoneum with free air between the liver and anterior abdominal wall (asterisk).

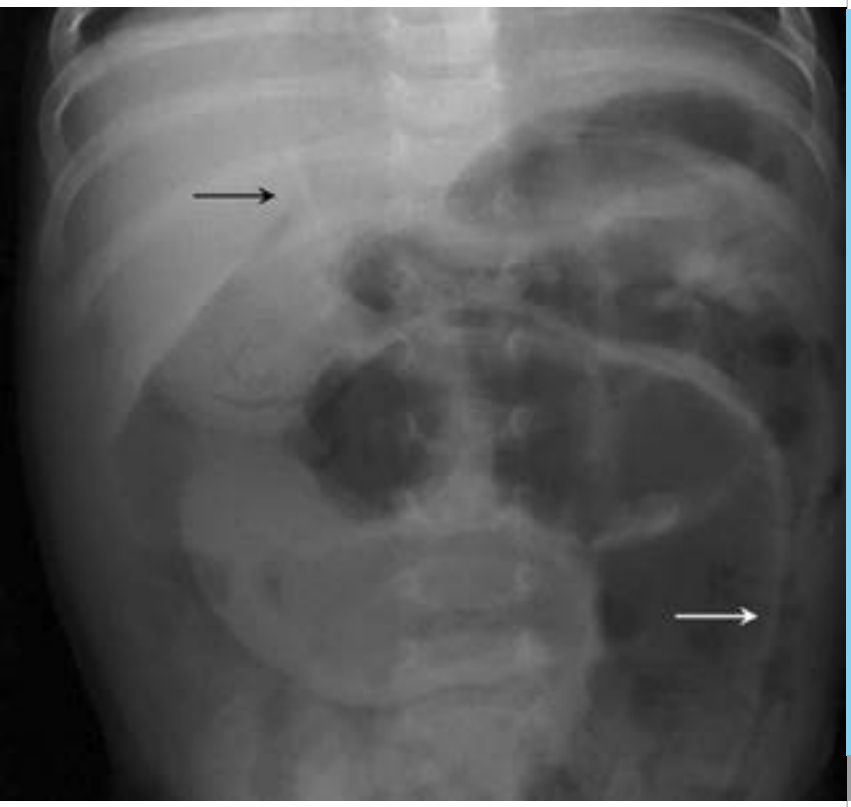

Fig. 2

Abdominal X-ray (supine view): pneumoperitoneum with a large oval lucency overlying the entire abdomen (football sign), visibility of both sides of the bowel wall (Rigler sign, white arrow) and falciform ligament outlined by air on both sides (black arrow).

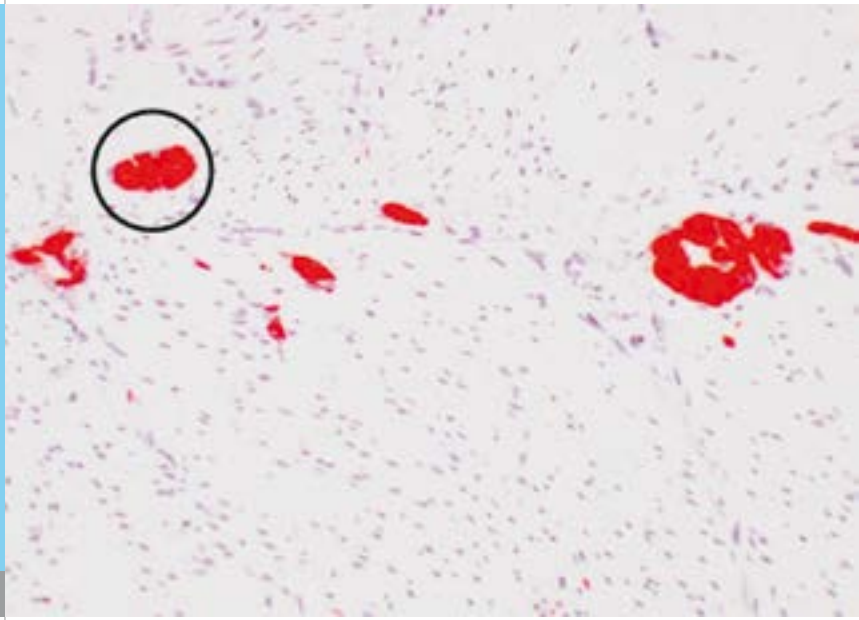

Fig. 3

Immunostain for neurofilament protein: presence of normal ganglion cells (circle) in a normal section.

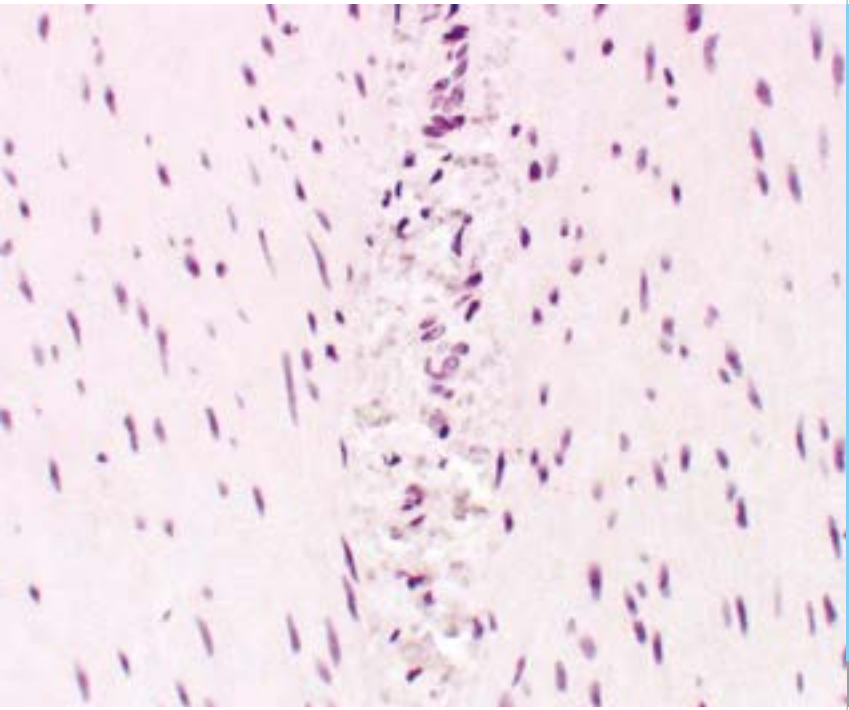

Fig. 4

Immunostain for neurofilament protein: absence of ganglion cells in the myenteric plexus (transverse colon).

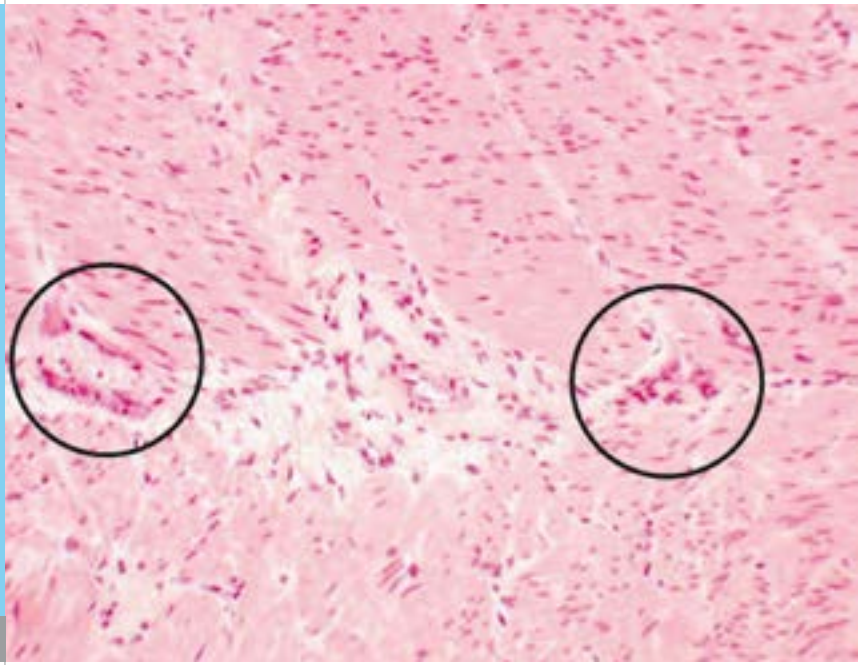

Fig. 5

H&E stain: presence of ganglion cells (circles) in the myenteric plexus (normal ileum).

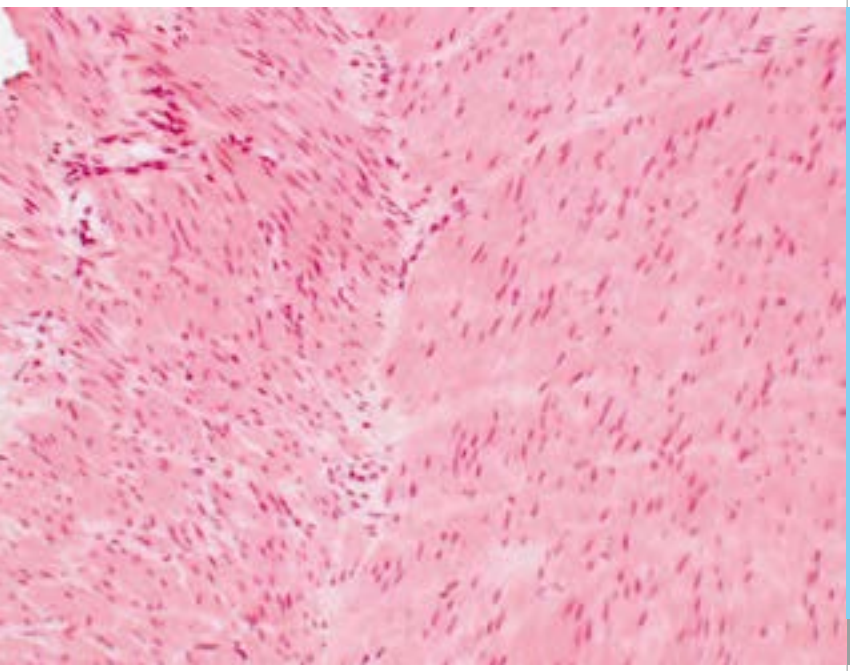

Fig. 6

H&E stain: absence of ganglion cells in the myenteric plexus (transverse colon).

DISCUSSION

Hirschsprung's disease (HD) is caused by congenital, usually focal absence of intramural ganglion cells in the rectum and distal intestine. Peristaltic failure and spasm of the affected segment result in proximal intestinal obstruction.

Disturbed neural crest migration into the rectosigmoid is responsible for the abnormal innervation in HD. Embryonic precursors of colonic ganglion cells enter the foregut and migrate caudally to innervate the small and large intestine. The length of the aganglionic segment is determined by the site of the most distal migration of neural crest cells. In 75% of cases, the aganglionic segment is limited to the rectum and distal sigmoid colon (short-segment disease), in 15% aganglionosis extends from the rectum to some point in the transverse colon. In the remaining patients, there is total colonic aganglionosis or even aganglionosis of some or all of the small intestine. In our patient, aganglionosis extended to the terminal ileum.

In HD, both the myenteric (Auerbach) and submucous (Meissner) plexuses are aganglionic. Histological evaluation of rectal suction biopsy has become the standard diagnostic procedure. In HD the nerve fibers of the plexus are present, but ganglion cells are absent.

The incidence of HD is reported to be about 1:5000 live births. Males are affected more commonly (75%). Most cases are sporadic, but 5% are familial. Associa-

tions with Down syndrome, colonic ileal atresia, congenital heart disease, genitourinary abnormalities or neurofibromatosis type I are seen.

Clinically, one third of patients with HD present with colonic obstruction during the first month of life. HD is responsible for 15% of cases of neonatal intestinal obstruction. In patients with delayed passage of meconium (more than 48 hours after birth) as well as patients with chronic constipation later in the first years of life, the possibility of HD should be considered.

Hirschsprung-associated enterocolitis is an uncommon and dreaded complication of untreated HD, presenting with paradoxical diarrhea, fever and a fulminant sepsis-like picture with bacterial translocation due to the combined effects of increased intraluminal pressure and large amounts of stagnant stool.

REFERENCE

Keeling J (2001). Fetal and Neonatal Pathology. Springer (London, Berlin, Heidelberg)

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