



Allied disorders of Hirschsprung's disease

J. Qiu¹ · G. Yang¹ · A. Lin²

Received: 16 April 2019 / Accepted: 6 May 2019 / Published online: 16 May 2019
© Springer Nature Switzerland AG 2019

Dear Sir,

Colonic obstruction is a common clinical manifestation which may be caused by a wide range of disorders. The most common causes of obstruction in coloproctology practice are colorectal tumors, benign strictures and volvulus. However, clinicians should be aware of rare causes of colonic obstruction.

We present a case of repeated colonic constipation caused by degenerated ganglion cells and deficiency of muscularis mucosae. Briefly, a 24-year-old woman came to the colorectal surgery clinic, complaining of abdominal pain and no bowel movements for 4 days. She had a history of two episodes of colonic obstruction, both of which resolved spontaneously. Anorectal examination was negative. Plain abdominal radiography showed sigmoid dilation and sporadic meteorism (Fig. 1). A computed tomography scan of the abdomen revealed no abnormality except a dilated descending and sigmoid colon (Fig. 2). After conservative treatment including fasting, parenteral nutrition and intravenous antibiotics, the patient's symptoms resolved. However, the young woman was planning a second pregnancy and she worried that such obstruction would probably occur again, which could not be ruled out. So a subtotal colectomy was performed after discussion with the patient and her husband. Postoperative pathological examination demonstrated degenerated ganglion cells and deficiency of muscularis mucosae (Fig. 3) with seemingly normal amount of ganglion cells and interstitial cells of Cajal. The patient had an uneventful recovery and was discharged on postoperative day 7.

Hirschsprung's disease (HD) occurs in approximately 1 in 5000 live births with an male to female ratio of 3:1 to 4:1

[1]. The cause of HD is commonly attributed to defective craniocaudal migration of neuroblasts originating from the neural crest during the first 12 weeks of gestation, resulting in functional intestinal obstruction [2]. HD is seldomly seen in adults as most patients are diagnosed early in life and are treated surgically. However, some patients with mild symptoms may go undiagnosed into adulthood. A manometry test and biopsy can help to establish the diagnosis of adult HD. In this case, the patient did not have any symptoms of obstruction until she gave birth to her first child and a normal rectoanal inhibitory reflex did not support a diagnosis of adult HD.

The term “allied disorders of Hirschsprung's disease” refers to a group of diseases that is characterized by symptoms and signs similar to those of HD despite the presence of

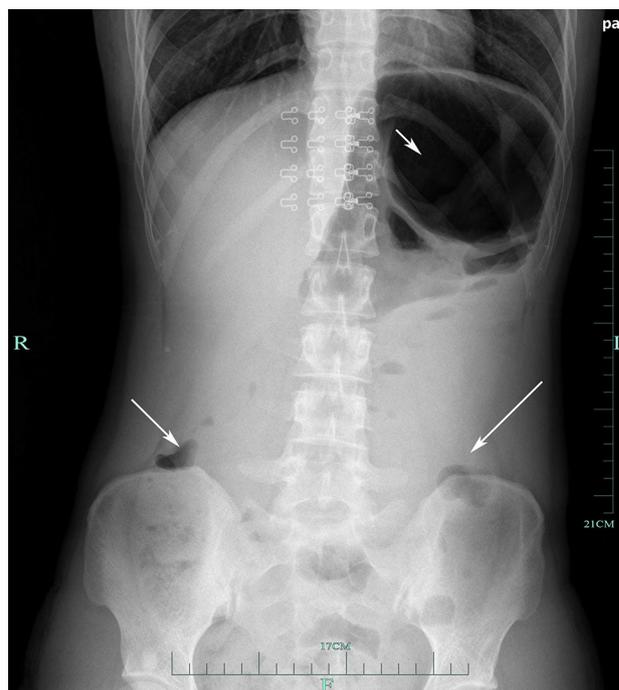


Fig. 1 Plain abdominal radiography showed sigmoid dilation (short arrow) and sporadic meteorism (long arrow)

✉ A. Lin
15168427344@163.com

¹ Department of Colorectal Surgery, The Third Hospital of Hangzhou, Hangzhou, China

² Department of Pathology, The Third Hospital of Hangzhou, West Lake Ave 38#, Hangzhou, China



Fig. 2 An abdominal computed tomography scan revealed dilated descending and sigmoid colon

ganglionic cells in the rectum. Allied disorders of HD can be divided into seven categories: (1) immaturity of ganglia; (2) isolated hypoganglionosis; (3) intestinal neuronal dysplasia (IND); (4) megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS); (5) segmental dilatation of intestine; (6) internal anal sphincter achalasia (IASA); and (7) chronic

idiopathic intestinal pseudo-obstruction [3]. However, the case we described here cannot be classified into any of the above seven subtypes.

Constipation owing to atrophy of the tendinous connective tissue of muscularis propria may result in an aperistaltic syndrome, which is a rare cause of constipation among children [4], and has similarities with our case. However, we believe degeneration of ganglion cells was more important in explaining the patient's clinical presentation.

In a patient with pseudo-obstruction or intractable constipation adult-onset HD or allied disorders of HD should be suspected [5]. A suction biopsy will help to differentiate the two diagnoses. In our case, a normal rectoanal inhibitory reflex and postoperative pathological analysis showing a normal amount of ganglion cells did not support either diagnosis. In clinical practice, we should think of the possibility of ganglion cell degeneration and deficiency of colon muscle when there is no proof of either hypoganglionosis or adult-onset HD. To the best of our knowledge this is the first report of such a rare condition. Although there are no guidelines about treatment, timely surgical intervention seems a reasonable choice. Due to the repeated episodes of colonic obstruction, we resected the involved colonic segment in this case. One year after surgery the patient has, on average, 2–5 bowel movements per day and has not experienced any bowel obstruction. Longer follow up is required to see if ganglion cell degeneration and muscularis mucosae deficiency in the remaining colon will cause recurrent symptoms.

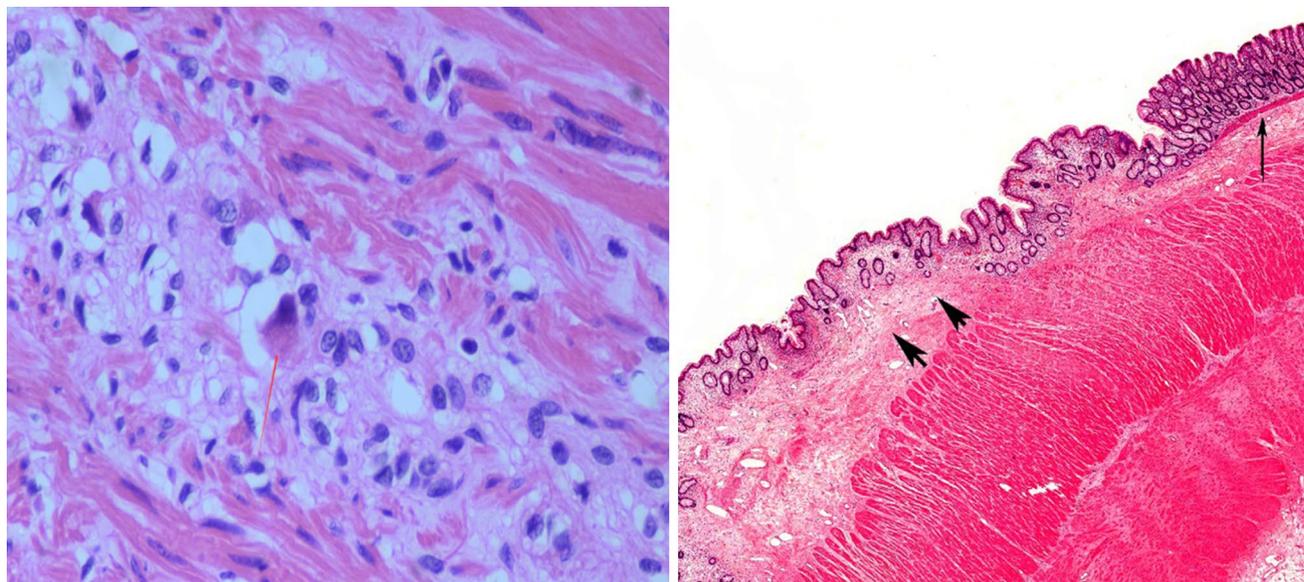


Fig. 3 Pathological examination demonstrated degenerated ganglion cells (left, red dotted line) and deficiency of muscularis mucosae (right, short arrow, while the long arrow indicates normal muscularis mucosae)

Author contributions JQ provided the study concept. GY collected the data, and AL did the study design and data interpretation and analysis.

Funding This study was funded by the medical technology foundation of Hangzhou (Grant no. 201764522).

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Ethical approval Institutional Review Board (IRB) approval has been obtained from the Ethical Committee of the Third Hospital of Hangzhou in compliance with the Principals Helsinki Declaration in 1964.

Informed consent All study participants, or their legal guardian, provided informed written consent prior to study enrollment including the possibility of future publication according to the Chinese bioethics laws.

References

1. Muto M, Matsufuji H, Taguchi T et al (2018) Japanese clinical practice guidelines for allied disorders of Hirschsprung's disease. *Pediatr Int* 60:400–410
2. Do MY, Myung SJ, Park HJ et al (2011) Novel classification and pathogenetic analysis of hypoganglionosis and adult-onset Hirschsprung's disease. *Dig Dis Sci* 56:1818–1827
3. Feichter S, Meier-Ruge WA, Bruder E (2009) The histopathology of gastrointestinal motility disorders in children. *Semin Pediatr Surg* 18(4):206–211
4. Chen F, Winston JH, Jain SK, Frankel WL (2006) Hirschsprung's disease in a young adult: report of a case and review of the literature. *Ann Diagn Pathol* 10:347–351
5. Bakari AA, Gali BM, Lbrahim AG, Nggada HA, Ali N, Dogo D, Abubakar AM (2011) Congenital aganglionic megacolon in Nigerian adults: two case reports and review of the literature. *Niger J Clin Pract* 14:249–252

Publisher's Note Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.