Double check: diagnosis and management of adult Hirschsprung disease

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SUMMARY

Hirschsprung disease (HD) is a gut motility disorder usually diagnosed acutely in infancy, although variants of HD may present later in life with indolent symptoms. This report highlights the rarity of diagnosing HD and hypoganglionosis in adulthood and the nuances that need consideration for their surgical management.

We present a report of a 49-year-old man presenting with chronic constipation. A full thickness rectal biopsy confirmed aganglionosis, and HD in adulthood was diagnosed. He underwent a defunctioning left-sided colostomy to ensure histological confirmation of ganglia in his left colon, and adequate colonic function via the colostomy.This served also as an assessment of the proximal conduit for any future anastomosis. He later underwent ultra-low anterior resection, coloanal anastomosis and loop ileostomy with subsequent reversal. His final histology revealed hypoganglionosis of the resected segment, with normal innervation to the site of the colostomy. He made full recovery with normal bowel movements.

BACKGROUND

Hirschsprung disease (HD) is a motility disorder of the gut usually diagnosed in infancy and caused by failed migration of the neural crest cells during fetal development. It is characterised by a non-functional, aganglionic segment of the colon, most commonly presenting with large bowel obstruction in infancy.[1] It occurs in approximately 1 in 5000 live births, affecting four times as many male as female patients.[2] Less than 6% of cases present after the age of 5,[3] making adult presentation of HD an often-overlooked differential. In the setting of adult presentation, hypoganglionosis must be considered, where there may be a relative lack of innervation of the colon, rather than a complete absence of ganglia.[4] In these cases, the condition follows the pattern of HD in that the hypoganglionosis is maximal distally, from the anorectal margin, and the nerves become progressively more functional proximally in the colon and there are no skip lesions.

One of the difficulties with HD in adulthood is that the affected segment will be stenosed, with chronic dilation of the colon proximal to the aganglionic or hypoganglionic segment.[5] This poses particular challenges to management, as the functional effects of long-term dilation and obstruction on a normally innervated colonic segment are poorly understood.

We present a case of an adult-diagnosed HD. We discuss the diagnostic and management difficulties this presentation poses and the rationale behind our management strategy which differs from those traditionally described for HD.

CASE PRESENTATION

A 49-year-old man was referred for specialist opinion with lifelong chronic constipation and abdominal pain. His problems began in childhood, with constipation from infancy which responded to laxative therapy. His problems persisted, and at age 8 he underwent colonoscopy and barium enema, which failed to reveal a diagnosis. When he was 13, he underwent sigmoid colectomy for an acute bowel obstruction. Histology on this bowel segment provided no clues to any underlying diagnosis. This alleviated his symptoms for a few years, but he became dependent on high-dose laxatives for the following three decades, with progressively worsening constipation.

INVESTIGATIONS

On presentation for specialist opinion at age 49, endoscopy and CT showed significant megarectum distally and a distended left colon, with a gradual transition zone in the transverse colon (figure 1). A full thickness rectal biopsy revealed an absence of ganglion cells in the submucosal and myenteric plexus and HD was confirmed.

After nearly half a century of chronic distension, there was concern that his colonic function in the innervated segment may be compromised and using it primarily as a conduit to a coloanal anastomosis would have unacceptable risk; however, he was very averse to a permanent stoma.

TREATMENT

Ultimately, he underwent a three-stage reconstruction, with full restoration of bowel continuity. First, he underwent a defunctioning left-sided colostomy, with a synchronous full thickness biopsy of the colon at that level. This was performed a few weeks after his clinic visit, as he emergently presented with impending large bowel obstruction. Histopathological analysis revealed normally innervated colon at the site of the colostomy in his descending colon, which reassured the surgical team that the hypoganglionosis was isolated to the segment of the colon distal to the colostomy, since we know that this condition does not have skip lesions. After a number of months, it was also clear from the patient clinically that his constipation and pain were gone—that is, he had normal colonic function to the level of the colostomy. This ‘double

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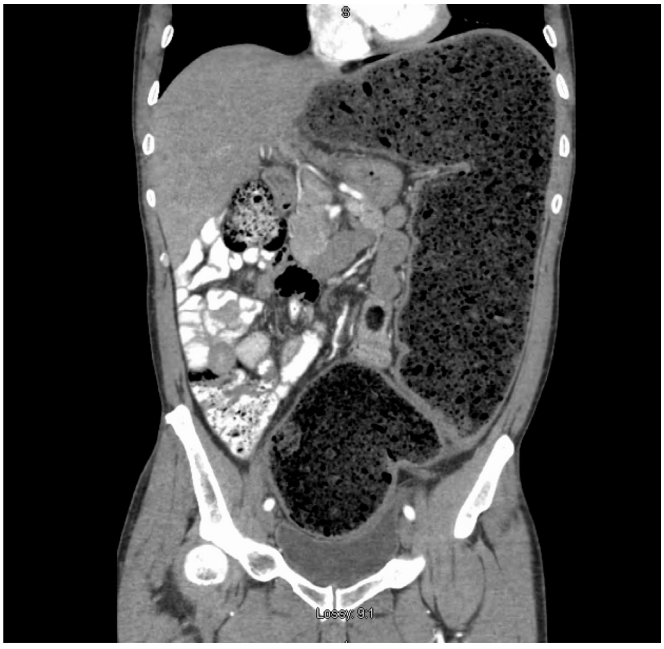

Figure 1 Coronal view of the megacolon at presentation to specialist referral service at age 49.

check' combination of histological and clinical evidence of normal colon function allowed optimism about restoring bowel continuity.

He then underwent open ultra-low anterior resection, coloanal anastomosis and loop ileostomy formation. This was performed with a standard approach to the total mesorectal excision plane, preserving the nerves and dissecting to the pelvic floor. A stapled coloanal anastomosis was performed and a drain left in situ, which was removed on the second postoperative day. Interestingly, final histological analysis on the resected colorectum confirmed hypoganglionosis as well as HD. Findings at the distal margin were consistent with aganglionosis (HD), but innervation at the proximal margin was entirely normal. The colon demonstrated progressive hypoganglionosis moving from the proximal margin distally, with fibrosis in place of the submucosal plexus, and smooth muscle and myenteric plexus hypertrophy.

OUTCOME AND FOLLOW-UP

After an anastomosis check clinically and endoscopically, he underwent subsequent reversal of the ileostomy and made an excellent recovery. He has excellent continence and restoration of normal bowel function, which has been maintained over 5 years since his coloanal reconstruction.

DISCUSSION

HD is due to failed migration of the neuroblasts in the large bowel, which are precursors to the myenteric and submucosal plexuses. The absence of these intrinsic intestinal nerves leads to unopposed extrinsic innervation and resultant increased smooth muscle tone with functional obstruction of the affected segment. There have been reports of adult presentation of HD, but many have been found to have hypoganglionosis rather than complete absence of both nerve plexuses. It is likely to represent a spectrum of HD. Our case demonstrates the atypical presentation which arises with diffuse-type hypoganglionosis and demonstrates the complexities of a diagnosis which may be missed. Its prevalence in adulthood is unknown and indeed it may explain many cases of chronic constipation worldwide.

Definitive management of HD includes resection of the aganglionic segment and anastomosis to properly innervated tissue with multiple described techniques[6]; the vast majority of reported cases involve a pull-through procedure[7] and most are completed in infancy. The first successful definitive operation for HD was described by Swenson in the 1950s and consisted of a pull-through procedure with an oblique coloanal anastomosis.[8] Shortly after this the modified Duhamel procedure was described, with a side-to-side anastomosis, avoiding the extensive pelvic dissection anterior to the rectum seen in Swenson's.[9 10] Soave's technique shortly followed, involving prolapse of the excess colon through the anus and spontaneous anastomosis in the postoperative period.[11] These approaches were popularised at a time when minimally invasive surgery and newer anastomotic techniques were unavailable. Our procedure is most similar to a conventional coloanal anastomosis as used for a low rectal cancer. There is limited literature regarding the above methods in the adult population, and in fact most similarly described cases in adulthood result in permanent colostomy. However, with newer devices and techniques, a stapled coloanal anastomosis is our approach of choice in a well-selected patient for whom continence is a priority. This may not always be the case, and depending on local expertise or facilities, or in resource-limited regions, permanent colostomy may be an appropriate alternate course of management.

The three-staged approach adopted with our patient also differs from those described in the literature. The lack of obviously demonstrable transition point and the unknown sequelae of his chronic distention meant that forming a primary conduit had unacceptable associated risk. The formation of the colostomy allowed adequate time to test the bowel function in the weeks postoperatively, as well as a histological 'double check' to assess ganglion distribution at that site. This highlights the importance of understanding hypoganglionosis and HD as a spectrum of disorders and their consideration in definitive management.

The varied management approaches highlight the complexities in treating these patients and reiterate that an armamentarium of options may be required to tailor patient treatment. Our patient with hypoganglionosis diagnosed late in adulthood had an excellent recovery and functional outcome using a three-staged technique.

Learning points

- Hypoganglionosis is likely an under-recognised condition and should be considered in adult patients with a history of slow-transit constipation since childhood.
- This variant of Hirschsprung disease leads to chronic hypertrophy of colonic smooth muscle with fibrosis and dysfunction.
- Performing a left-sided colostomy safely allows a 'double check' to histopathologically and clinically assess the colostomy site as a point for future anastomosis.

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REFERENCES

1 Schlund D, Jochum SB, Favuzza J, *et al*. A national analysis of operative treatment of adult patients with Hirschsprung's disease. *Int J Colorectal Dis* 2020;35:169–72.
2 Chen F, Winston JH, Jain SK, *et al*. Hirschsprung's disease in a young adult: report of a case and review of the literature. *Ann Diagn Pathol* 2006;10:347–51.
3 Qiu J-F, Shi Y-J, Hu L, *et al*. Adult Hirschsprung's disease: report of four cases. *Int J Clin Exp Pathol* 2013;6:1624–30.
4 Qiu J, Yang G, Lin A. Allied disorders of Hirschsprung's disease. *Tech Coloproctol* 2019;23:509–11.
5 Kim HJ, Kim AY, Lee CW, *et al*. Hirschsprung disease and hypoganglionosis in adults: radiologic findings and differentiation. *Radiology* 2008;247:428–34.
6 Wester T, Granström AL. Hirschsprung disease-Bowel function beyond childhood. *Semin Pediatr Surg* 2017;26:322–7.
7 Doodnath R, Puri P. A systematic review and meta-analysis of Hirschsprung's disease presenting after childhood. *Pediatr Surg Int* 2010;26:1107–10.
8 Swenson O. A new surgical treatment for Hirschsprung's disease. *Surgery* 1950;28:371–83.
9 Duhamel B. A new operation for the treatment of Hirschsprung's disease. *Arch Dis Child* 1960;35:38–9.
10 Natsikas NB, Sbarounis CN. Adult Hirschsprung's disease. An experience with the Duhamel-Martin procedure with special reference to obstructed patients. *Dis Colon Rectum* 1987;30:204–6.
11 Soave F. Hirschsprung's disease: a new surgical technique. *Arch Dis Child* 1964;39:116–24.