Case report of a skip segment Hirschprung's disease: A real phenomenon

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ABSTRACT

INTRODUCTION AND IMPORTANCE: Hirschprung’s disease is a congenital anomaly that results from an incomplete cranio-caudal migration and maturation of intestinal ganglion progenitor cells leading to distal intestinal aganglionosis. Skip segment Hirschprung’s disease is an extremely rare phenomenon. We report a case involving only the small bowel with confirmed colonic ganglionosis.

CASE PRESENTATION: A case report of a 14-month-old with a skipped segment involving the distal 50 cm of the small bowel associated with colonic ganglionosis is presented. A current review of the literature is discussed.

CLINICAL DISCUSSION: Our patient had persistent obstructive symptoms despite undergoing a technically good, ganglionic pull-through operation at an outside institution. A laparoscopic-assisted pull-through might have documented a small bowel wall diameter discrepancy.

CONCLUSION: Although rare, skip segment Hirschprung’s disease is a real phenomenon that paediatric surgeons should be aware of and could involve small and large bowels.

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1. Introduction

Hirschsprung’s disease is a congenital anomaly that manifests as functional intestinal obstruction due to the failure of cranio-caudal migration and maturation of intestinal ganglion progenitor cells, leading to distal intestinal aganglionosis [1]. Depending on the level of arrested caudal migration, a patient may manifest with a short segment, long segment, total colonic, or total intestinal aganglionosis. In all cases, the congenital absence of ganglion cells starts in the distal rectum and extends proximally. A rare entity has been described in the literature of patients with Hirschsprung’s disease presenting with a skipped segment [2]. This goes against the embryological theory of arrested cranio-caudal migration of neural crest-derived cells. O’Donnell et al. described in their systematic review 24 cases of segmental Hirschsprung’s disease, all of which involved the colon [2]. We describe a rare case of segmental Hirschsprung’s disease involving the distal 50 cm of small bowel with confirmed colonic ganglionosis.

2. Presentation of case

A 14-month-old boy with no relevant family or drug history underwent a trans-anal only Soave pull-through at an outside institution for a reported short segment Hirschsprung’s disease when he was two weeks of age. The patient was doing well until he was 4-months old when he developed an episode of enterocolitis. Subsequently, the patient had eight enterocolitis episodes, all requiring hospital admissions despite undergoing daily rectal irrigation and receiving four separate internal anal sphincter Botox injections. He took 45 mg of Senna daily; however, he had persistent obstructive symptoms and abdominal distension. He was referred to our centre for a second opinion regarding his ongoing abdominal distention and recurrent enterocolitis. A scout film during the contrast enema revealed several dilated small bowel loops with evidence of faecalization concerning for partial small bowel obstruction (Fig. 1A). However, despite evidence of small bowel dilatation on the scout imaging, the contrast refluxed into non-dilated terminal ileum suggesting an obstruction caused by an internal hernia more proximally (Fig. 2B). The next day, he was taken to the operating room by the senior author for a presumed internal hernia since his original operation was a trans-anal only approach. An examination under anesthesia of the anus revealed an intact dentate line with the anastomosis visible, but no evi-
Fig. 1. (A) Scout film with several dilated small bowel loops with evidence of faecalization concerning for partial small bowel obstruction. (B) Contrast enema with contrast filling normal caliber “question mark-like” colon and terminal ileum.

Fig. 2. Exploratory laparotomy revealing aganglionic distal 50 cm of the small bowel (*) decompressed with dilated proximal small bowel (+). The caecum (x) and colon (#) were all ganglionic.

dence of a stricture. We proceeded with a diagnostic laparoscopy and did not find evidence of an internal hernia to account for the diffuse proximal small bowel dilatation. We converted to an exploratory laparotomy through a lower midline incision. The terminal 50 cm of small bowel was decompressed (Fig. 2*) and identified a transition zone proximally to a more dilated small bowel without any clear aetiology (Fig. 2*). Full-thickness biopsies were initially taken from the terminal ileum (Fig. 2*) and caecum (Fig. 2*). Ganglion cells were present in the caecum, but the terminal ileum was aganglionic. This prompted us to take full-thickness biopsies at 10, 15, 20, 25, 45, and 50 cm from the ileocaecal valve (Fig. 2*). All were aganglionic except for the dilated small bowel measured at 50 cm from the ileocaecal valve (Fig. 3). Biopsies of the hepatic flexure, descending colon, and rectum confirming ganglionic bowel (Fig. 3). The patient was diverted with a divided ileostomy and mucous fistula. The patient was then reversed six months later with the aganglionic bowel resected and an ileocolic anastomosis performed. He was followed in the surgical outpatients and is now thriving with no further episodes of enterocolitis.

3. Discussion

We present a segmental Hirschsprung's disease case involving only the small bowel, with confirmed colonic ganglionosis. To the best of our knowledge, there have been 40 reported cases of skip segment Hirschsprung's disease reported in the literature, the vast majority of which involve the colon (Table 1) [2–6]. While the disease is believed to result from arrested craniocaudal migration of neural crest-derived cells, these cases go against the classic embryological explanation. In O'Donnell et al. systematic review of 24 cases of segmental Hirschsprung's disease, all of the cases involved only the colon, with 22 out of 24 cases being total colonic aganglionosis [2]. It has been theorized the skip lesion results from extramural migration of neuroblasts across the mesenteric border and into the colon, thereby ending up ahead of the wavefront
Fig. 3. Haematoxylin and Eosin staining of the full-thickness biopsies of different parts of the intestine.
resulting in the skip segment [2]. With 41 reported cases in the literature, including our case, skip segment Hirschsprung’s disease must be considered a real phenomenon (Table 1). We suspect there are probably several unreported cases in the world with segmental Hirschsprung’s disease that paediatric surgeons have dismissed since it goes against the dogma of arrested cranio-caudal migration. Our case is unique because the aganglionic segment involves only the distal small bowel with confirmed colonic ganglionosis. Since the original pull-through operation was done via a trans-anal only approach, this might have led the surgeon to miss two transition points, which could have been identified laparoscopically. We recommend against a trans-anal only approach for any pull-through procedure to document the transition zone and offer the patient the correct surgery the first time. This case illustrates how careful mapping of bowel via multiple biopsies can yield the maximal amount of bowel possible for a pull-through and avoid the necessity of multiple operations.

It is clear from the 41 cases of the skip segment variant of Hirschsprung’s disease that there is a strong male predilection with an almost 5:1 male-to-female ratio and strong preponderance in total colonic aganglionosis. However, a question mark remains (Fig. 1B) over the embryological explanation of skip segment Hirschsprung’s disease. Until further studies are done, it will remain an enigma.

4. Conclusion

Trans-anal only pull-through is ill advised. Although rare, skip segment Hirschsprung’s disease is a real phenomenon that paediatric surgeons should be aware of and could involve small and large bowels.

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Declaration of Competing Interest

The authors report no declarations of interest.

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Ethical approval

Ethical approval is exempt at our institution.

Table 1

| Reference  | Year | Sex | Number of patients | Location of skip segment |
|------------|------|-----|--------------------|--------------------------|
| Keeper     | 1954 | M   | 1                  | Rectosigmoid with skip in sigmoid |
| sprinz     | 1961 | F   | 1                  | Rectosigmoid with skip in sigmoid |
| Mac Iver   | 1972 | M   | 1                  | TCA except skip in transverse colon |
| Martin     | 1979 | M   | 1                  | TCA except skip in transverse colon |
| De Chadarevian | 1982 | M   | 1                  | TCA except skip in transverse colon |
| Yunis      | 1983 | M   | 5                  | TCA except skip in transverse colon |
|            |      | M   |                    | TCA except skip in transverse colon |
| Taguchi    | 1983 | M   | 1                  | TCA except skip in ascending colon |
| Seldenrijk | 1986 | M   | 2                  | TCA with multiple skips |
| Anderson   | 1986 | M   | 1                  | TCA except skip in ascending colon |
| Kapur      | 1995 | F   | 3                  | TCA except skip in ascending colon |
| Yang       | 2005 | M   | 3                  | TCA except skip in ascending colon |
| Ziad       | 2006 | M   | 2                  | TCA except skip in transverse colon |
| Oshio      | 2008 | M   | 1                  | TCA except skip in ascending colon |
| Puri       | 2010 | M   | 1                  | TCA except skip in transverse colon |
| castle     | 2011 | M   | 1                  | Aganglionosis terminal 8 cm of TI and TCA except skip in transverse colon and caecum |
| Doi        | 2011 | F   | 1                  | TCA except skip in transverse colon |
| Burjonrappa| 2012 | M   | 1                  | TCA except skip in caecum |
| Skelly     | 2012 | M   | 1                  | TIA except stomach, 10 and 45 cm distal to DJF |
| moore      | 2013 | F   | 2                  | Skip in right and ascending colon |
| Erten      | 2014 | M   | 1                  | Skip in ascending colon and appendix |
| RAGUNATH   | 2014 | M   | 1                  | TCA except skip in ascending colon and 2 cm aganglionic ileum 6 cm from TI |
| Gross      | 2015 | M   | 1                  | TCA with skip in ascending colon and hepatic flexure |
| Ruiz       | 2016 | M   | 1                  | Skip in transverse and descending colon |
| COE        | 2016 | M   | 2                  | TCA except skip in caecum and ascending colon |
| Alfawaz    | 2017 | M   | 1                  | Distal rectal skip segment |
| Shenoy     | 2019 | M   | 1                  | Skip in proximal sigmoid |
| Yu         | 2019 | M   | 2                  | Distal rectal skip segment |
| El-Gohary  | 2020 | M   | 1                  | Skip in sigmoid |

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Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Authors’ contribution

Study conception and design: Richard J Wood.
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Registration of research studies

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Guarantor

Richard J Wood.

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References

[1] J.C. Langer, Hirschsprung disease, Curr. Opin. Pediatr. 25 (3) (2013) 368–374, http://dx.doi.org/10.1097/MOP.0b013e32836c2a0.
[2] A.M. O’Donnell, P. Puri, Skip segment Hirschsprung’s disease: a systematic review, Pediatr. Surg. Int. 26 (11) (2010) 1063–1069, http://dx.doi.org/10.1007/s00383-010-2692-4.
[3] E.E. Ertan, Y.H. Cavusoglu, N. Arda, et al., A rare case of multiple skip segment Hirschsprung’s disease in the ileum and colon, Pediatr. Surg. Int. 30 (3) (2014) 349–351, http://dx.doi.org/10.1007/s00383-013-3428-x.
[4] S. Burjonrappa, L. Rankin, ‘Hop the skip’ with extended segment intestinal biopsy in Hirschsprung’s disease, Int. J. Surg. Case Rep. 3 (5) (2012) 186–189, http://dx.doi.org/10.1016/j.jsrcr.2012.02.004.
[5] S. Castle, A. Suliman, K. Shayan, K. Kling, S. Bickler, B. Losasso, Total colonic aganglionosis with skip lesions: report of a rare case and management, J. Pediatr. Surg. 47 (3) (2012) 581–584, http://dx.doi.org/10.1016/j.jpedsurg.2011.11.067.
[6] B.L. Shelby, E. Ervine, M. Bisharat, C. Gannon, A.C. Dick, Small bowel skip segment Hirschsprung’s disease presenting with perforated Meckel’s diverticulum, Pediatr. Surg. Int. 28 (6) (2012) 645–648, http://dx.doi.org/10.1007/s00383-011-3050-x.
[7] G.P. Keefe, J.F. Mokrohisky, Congenital megacolon (Hirschsprung’s disease), Radiology 63 (2) (1954) 157–175, http://dx.doi.org/10.1148/63.2.157.
[8] H. Sprinz, A. Cohen, L.D. Heaton, Hirschsprung’s disease with skip area, Ann. Surg. 153 (1) (1961) 143–148, http://dx.doi.org/10.1097/00000499-197203252-233.
[9] A.G. MacVicar, R. Whitehead, Zonal colonic aganglionosis, a variant of Hirschsprung’s disease, Arch. Dis. Child. 47 (252) (1972) 233–237, http://dx.doi.org/10.1016/s0022-3468(79)80245-6.
[10] L.W. Martin, J.J. Buchino, C. Le Coutre, E.T. Ballard, W.W. Nebbett, Hirschsprung’s disease with skip area (segmental aganglionosis). J. Pediatr. Surg. 14 (6) (1979) 666–668, http://dx.doi.org/10.1016/s0022-3468(82)80213-3.
[11] J.P. de Chadarevian, M. Slim, S. Akel, Double zonal aganglionosis in long segment Hirschsprung’s disease with a “skip area”: a report of seven colon, J. Pediatr. Surg. 17 (2) (1982) 195–197, http://dx.doi.org/10.1016/0022-3468(78)90028-8.
[12] E. Yunis, W.K. Sieber, D.R. Akers, Does zonal aganglionosis really exist? Report of a rare variety of Hirschsprung’s disease and review of the literature, Pediatr. Pathol. 1 (1) (1983) 33–49, http://dx.doi.org/10.1055/s-2000-1059995.
[13] T. Taguchi, K. Tanaka, K. Ikeda, A. Hata, Double zonal aganglionosis with a skipped oligoaganglionosis, Z Kinderchir. 38 (5) (1983) 312–315, http://dx.doi.org/10.1007/bf00710909.
[14] C.A. Seldenrijk, H.J. van der Harten, P. Kluck, D. Tibboel, K. Moorman-Voestermans, C.J. Meijer, Zonal aganglionosis. An enzyme and immunohistochemical study of two cases, Virchows Arch. A Pathol. Anat. Histopathol. 410 (1) (1986) 75–81, http://dx.doi.org/10.1007/bf0022-3468/s00006-9.
[15] K.D. Anderson, R. Chandra, Segmental aganglionosis of the appendix, J. Pediatr. Surg. 21 (10) (1986) 852–854, http://dx.doi.org/10.1016/s0022-3468(82)80245-6.
[16] R.P. Kapur, D.J. deSa, M. Luquette, R. Jaffe, Hypothesis: pathogenesis of skip areas in long-segment Hirschsprung’s disease, Pediatr. Pathol. Lab. Med. 15 (1) (1995) 23–37, http://dx.doi.org/10.1080/15513815.2014.971685.
[17] H.Y. Yang, Q.L. Liu, J.X. Wang, H.F. Xu, Clinical study of multiple zonal aganglionosis in long segment Hirschsprung’s disease, Zhonghua Yi Xue Za Zhi (35) (2005) 2777–2774, http://dx.doi.org/10.1007/s11655-009-9760-1.
[18] F. Ziad, K.C. Katchy, S. Al Ramadan, S. Alexander, S. Kumar, Clinicopathological features in 102 cases of Hirschsprung disease, Ann. Saudi Med. 26 (3) (2006) 200–204, http://dx.doi.org/10.1016/j.asmed.2006.10.005.
[19] T.O. Oshio, Imperforate anus, malrotation, and Hirschsprung’s disease with double zonal aganglionosis: an extremely rare combination, J. Pediatr. Surg. 43 (1) (2008) 222–226, http://dx.doi.org/10.1016/j.jpedsurg.2007.09.005.
[20] A. Coe, J.R. Avansino, R.P. Kapur, Distal rectal skip-segment hirschsprung disease and the potential for false-negative diagnosis, Pediatr. Dev. Pathol. 19 (2) (2016) 123–131, http://dx.doi.org/10.23550/15-08-1686-0A.1.
[21] T. Doi, A.M. O’Donnell, M. McDermott, P. Puri, Skip segment Hirschsprung’s disease: a case report and review of literature, Fetal Pediatr. Pathol. 38 (5) (2019) 437–443, http://dx.doi.org/10.1080/15513815.2019.1680806.
[22] A.R.C. Allawaz, K. Rao, J. Sola, Neville H. Skip segment hirschsprung disease: avoiding the potential pitfall of a failed pull-through procedure, EC Paediatrics Si. 1 (2017) 25–28.
[23] S.H.C. Ruiz, R. Fernandes, J. Osorio, R. Nunez, Skip segment Hirschsprung’s disease in a patient with Shah-Waardenburg Syndrome, J. Pediatr. Surg. Case Rep. 15 (2016) 44–47, http://dx.doi.org/10.1016/j.jsrcc.2016.08.012.
[24] E.G.G. Gross, J. McCarrick, J. Zarzembowski, M. Arca, Skip segment Hirschsprung disease and Waardenburg syndrome, J. Pediatr. Surg. Case Rep. 3 (4) (2015) 143–145, http://dx.doi.org/10.1016/j.jsrcc.2015.02.005.
[25] B.V. Raghunathan, G. Shankar, M.N. Babu, et al., Skip segment Hirschsprung’s disease: a case report and novel management technique, Pediatr. Surg. Int. 30 (1) (2014) 119–122, http://dx.doi.org/10.1007/s00383-013-3367-8.
[26] S.W. Moore, D. Suller, P.A. Schubert, Segmental aganglionosis (zonal aganglionosis or “skip” lesions) in Hirschsprung disease: a report of 2 unusual cases, Pediatr. Surg. Int. 29 (5) (2013) 495–500, http://dx.doi.org/10.1007/s00383-013-3286-5.
[27] R.A. Agha, T. Franchi, C. Sohrabi, G. Mathew, for the SCARE Group. The SCARE 2020 guideline: updating consensus surgical Case Report (SCARE) guidelines, Int. J. Surg. 84 (2020) 226–230.