CASE REPORT – OPEN ACCESS

International Journal of Surgery Case Reports 80 (2021) 105630

Contents lists available at ScienceDirect

International Journal of Surgery Case Reports

journal homepage: www.casereports.com

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

Yousef El-Gohary[a], Clare Skerritt[b], Vinay Prasad[c], Ihab Halaweish[d], Richard J. Wood[d,*]

[a] Department of Paediatric Surgery, Leeds Children's Hospital, Leeds, UK
[b] Department of Paediatric Surgery, Bristol Royal Hospital for Children, Bristol, UK
[c] Department of Pathology and Laboratory Medicine, Nationwide Children's Hospital, Columbus, OH, USA
[d] Department of Pediatric Colorectal and Pelvic Reconstructive Surgery, Nationwide Children's Hospital, 700 Children's Drive, Columbus, OH, USA

ARTICLE INFO

Article history:
Received 16 December 2020
Received in revised form 4 February 2021
Accepted 4 February 2021
Available online 9 February 2021

Keywords:
Hirschsprung's disease
Skip segment
Aganglionosis
Case report

ABSTRACT

INTRODUCTION AND IMPORTANCE: Hirschsprung's disease is a congenital anomaly that results from an incomplete craniocaudal migration and maturation of intestinal ganglion progenitor cells leading to distal intestinal aganglionosis. Skip segment Hirschsprung'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 Hirschsprung's disease is a real phenomenon that paediatric surgeons should be aware of and could involve small and large bowels.

1. Introduction

Hirschsprung's disease is a congenital anomaly that manifests as functional intestinal obstruction due to the failure of craniocaudal 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 craniocaudal 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 distention. 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-

* Corresponding author at: Department of Pediatric Colorectal and Pelvic Reconstructive Surgery, Nationwide Children's Hospital, 611 E. Livingston Ave., Columbus, OH, 43205, USA.
E-mail address: richard.wood@nationwidechildrens.org (R.J. Wood).

https://doi.org/10.1016/j.ijscr.2021.02.016

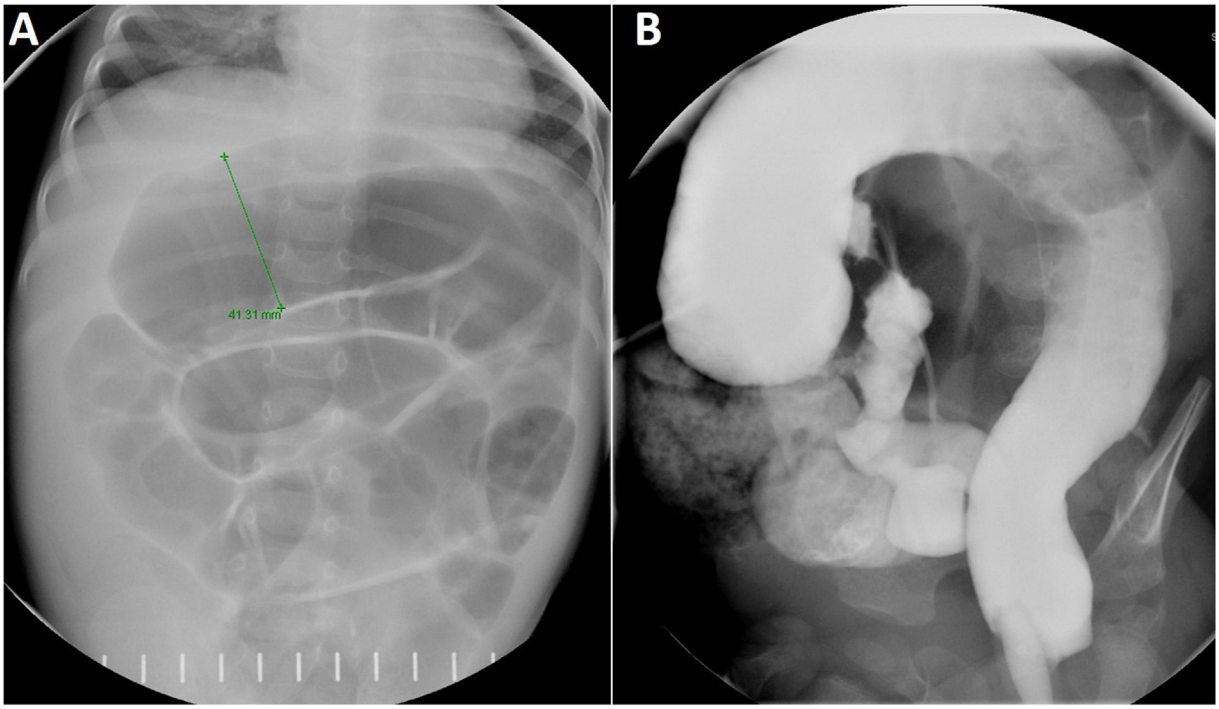

Fig. 1. (**A**) Scout film with several dilated small bowel loops with evidence of faecalization concerning for partial small bowel obstruction. (**B**) Contrast enema with contrast filling normal caliber "question mark-like" colon and terminal ileum.

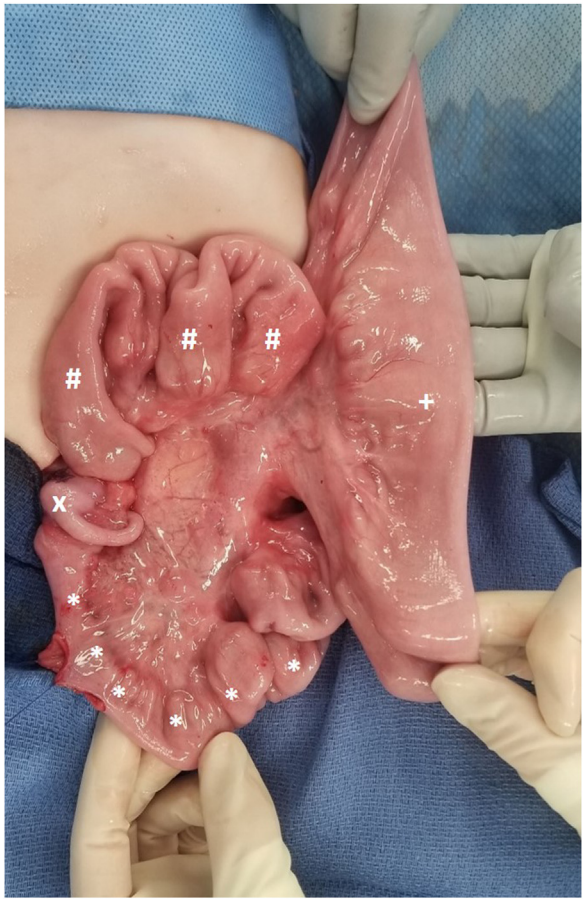

Fig. 2. Exploratory laparotomy revealing aganglionic distal 50 cm of the small bowel (*) decompressed with dilated proximal small bowel (+). The caecum (x) and colon (#) were all ganglionic.

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 dilation. 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. 2x). 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 ileocecal 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

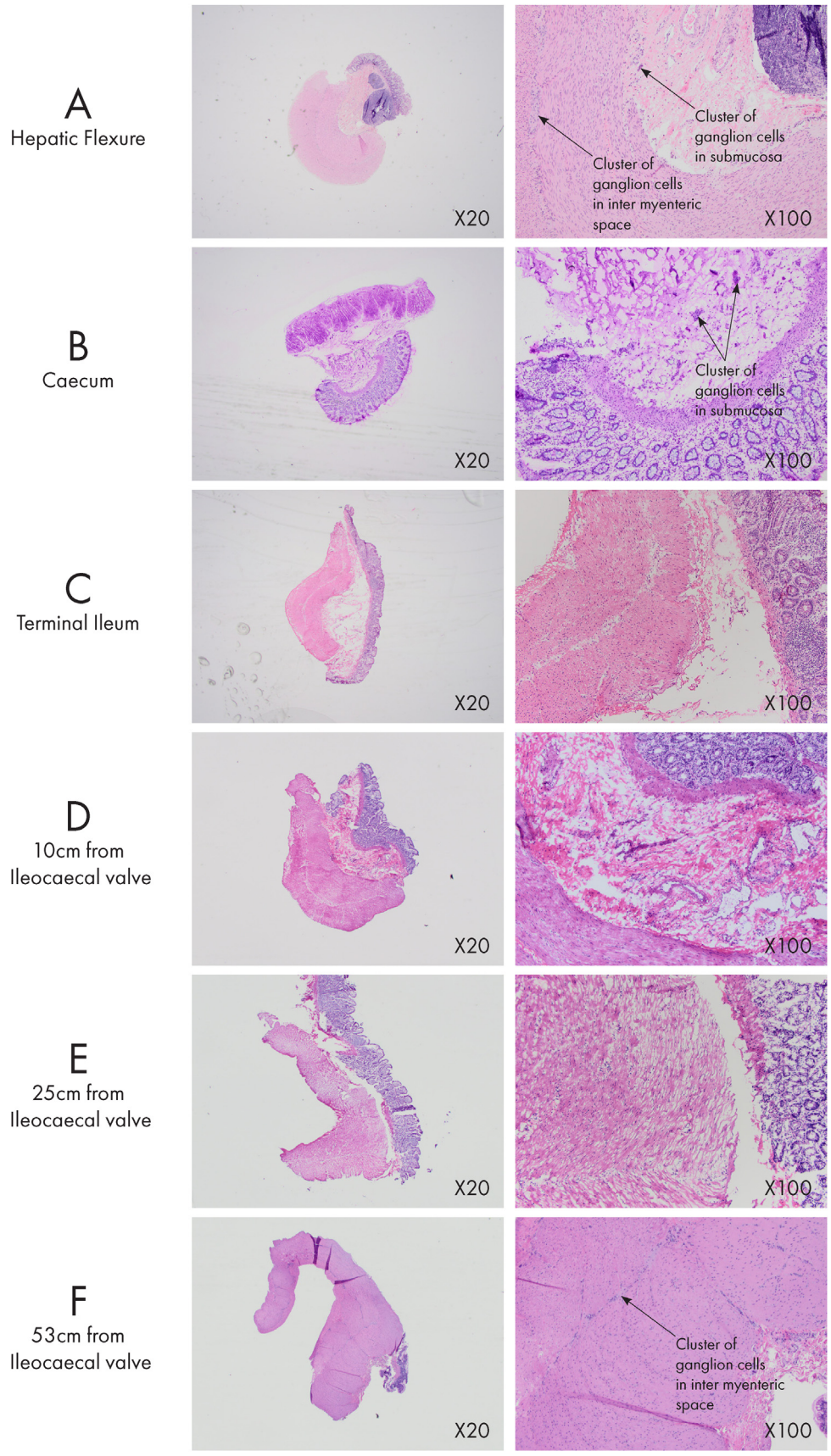

Fig. 3. Haematoxylin and Eosin staining of the full-thickness biopsies of different parts of the intestine.

Table 1
Summary of all cases of skip segment Hirschsprung's disease reported in the literature from 1954 to 2020 [7–28]. TCA: Total colonic aganglionosis; TIA: Total intestinal aganglionosis; TI: terminal ileum; DJF: Duodenal jejunal flexure.

Reference	Year	Sex	Number of patients	Location of skip segment
Keefer	1954	M	1	Rectosigmoid with skip in sigmoid
sprinz	1961	F	1	TCA except skip in transverse colon
Mac Iver	1972	M	1	Rectosigmoid with skip in sigmoid
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
		M		TCA except skip in transverse colon
		M		TCA except skip in transverse colon
		M		TCA except skip in ascending colon
Taguchi	1983	M	1	TCA except skip in ascending colon
Seldenrijk	1986	M	2	TCA with multiple skips
		F		TCA with multiple skips
Anderson	1986	M	1	TCA except skip in ascending colon
Kapur	1995	F	3	TCA except skip in ascending colon
		F		TCA except skip in caecum
		F		TCA except skip in ascending colon
Yang	2005	M	3	TCA with multiple skips
		M		TCA with multiple skips
		M		TCA with multiple skips
Ziad	2006	M	2	TCA except skip in transverse colon
		F		TCA except skip in caecum
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	M	2	Skip in right and ascending colon
		F		Skip in ascending colon and appendix
Erten	2014	M	1	TCA except skip in ascending colon and 2 cm aganglionic ileum 6 cm from TI
RAGUNATH	2014	M	1	TCA with skip in ascending colon and hepatic flexure
Gross	2015	M	1	Skip in transverse and descending colon
Ruiz	2016	M	1	TCA except skip in caecum and ascending colon
COE	2016	M	2	Distal rectal skip segment
Alfawaz	2017	M	1	Skip in proximal sigmoid
Shenoy	2019	M	1	Distal rectal skip segment
Yu	2019	M	2	Skip in sigmoid
		M		Skip in transverse colon
El-Gohary	2020	M	1	Aganglionosis terminal 50 cm of TI

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 paeditric surgeons have dismissed since it goes against the dogma of arrested craniocaudal 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 advisable. 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.

This manuscript was written in line with the SCARE 2020 Guideline [29].

Declaration of Competing Interest

The authors report no declarations of interest.

Funding

The authors declare they have received no funding for the preparation of this document.

Ethical approval

Ethical approval is exempt at our institution.

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.
Data acquisition: Yousef El-Gohary, Vinay Shah.
Analysis and data interpretation: Yousef El-Gohary, Clare Skerritt, Vinay Shah, Ihab Halaweish, Richard J Wood.
Drafting of the manuscript: Yousef El-Gohary, Clare Skerritt.
Critical revision: Clare Skerritt, Vinay Shah, Ihab Halaweish, Richard J Wood.

Registration of research studies

Not Applicable.

Guarantor

Richard J Wood.

Provenance and peer review

Not commissioned, externally peer-reviewed.

References

[1] J.C. Langer, Hirschsprung disease, Curr. Opin. Pediatr. 25 (3) (2013) 368–374, http://dx.doi.org/10.1097/MOP.0b013e328360c2a0.
[2] A.M. O'Donnell, P. Puri, Skip segment Hirschsprung's disease: a systematic review, Pediatr. Surg. Int. 26 (11) (2010) 1065–1069, http://dx.doi.org/10.1007/s00383-010-2692-4.
[3] E.E. Erten, 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-z.
[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.ijscr.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. Skelly, E. Ervine, M. Bisharat, C. Gannon, A.C. Dick, Small bowel skip segment Hirschprung'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. Keefer, 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/10.1136/adc.47.252.233.
[9] A.G. MacIver, 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. LeCoultre, E.T. Ballard, W.W. Neblett, Hirschsprung's disease with skip area (segmental aganglionosis), J. Pediatr. Surg. 14 (6) (1979) 686–687, 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" in transverse colon, J. Pediatr. Surg. 17 (2) (1982) 195–197, http://dx.doi.org/10.3109/15513818309048283.
[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-2008-1059995.
[13] T. Taguchi, K. Tanaka, K. Ikeda, A. Hata, Double zonal aganglionosis with a skipped oligoganglionic ascending colon, 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/10.1016/s0022-3468(86)80006-9.
[15] K.D. Anderson, R. Chandra, Segmental aganglionosis of the appendix, J. Pediatr. Surg. 21 (10) (1986) 852–854, http://dx.doi.org/10.3109/15513819509026937.
[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/10.5144/0256-4947.2006.200.
[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. 85 (39) (2005) 2772–2774, 10.3760/j:issn:0376-2491.2005.39.011.
[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/10.5144/0256-4947.2006.200.
[19] T. 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/10.2350/15-08-1686-OA.1.
[21] T. Doi, A.M. O'Donnell, M. McDermott, P. Puri, Skip segment Hirschsprung's disease: a rare phenomenon, Pediatr. Surg. Int. 27 (7) (2011) 787–789, http://dx.doi.org/10.1007/s00383-011-2903-7.
[22] Q. Yu, M. Du, W. Zhang, et al., Mesenteric neural crest cells are the embryological basis of skip segment hirschsprung's disease, Cell. Mol. Gastroenterol. Hepatol. (2020), http://dx.doi.org/10.1016/j.jcmgh.2020.12.010.
[23] A. Shenoy, Y. De Los Santos, K.N. Johnson, R. Petroze, Distal rectal skip segment Hirschsprung disease: case report and review of literature, Fetal Pediatr. Pathol. 38 (5) (2019) 437–443, http://dx.doi.org/10.1080/15513815.2019.1608606.
[24] A.R.C. Alfawaz, K. Rao, J. Sola, Neville H. Skip segment hirschsprungs disease: avoiding the potential pitfall of a failed pull-through procedure, EC Paediatrics SI. 1 (2017) 25–28.
[25] S.H.C. Ruiz, R. Fernandes, J. Ossorio, 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.epsc.2016.08.012.
[26] E.G.G. Gross, J. McCarrier, J. Jarzembowski, 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.epsc.2015.02.005.
[27] B.V. Raghunath, 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.
[28] S.W. Moore, D. Sidler, P.A. Schubert, Segmental aganglionosis (zonal aganglionosis or "skip" lesions) in Hirschsprungs disease: a report of 2 unusual cases, Pediatr. Surg. Int. 29 (5) (2013) 495–500, http://dx.doi.org/10.1007/s00383-013-3286-8.
[29] 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.