

## Case report

# Skip-segment Hirschsprung's disease: Report of a rare case and literature review

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## Abstract

We present an unusual case of skip-segment Hirschsprung's disease, initially diagnosed with total colonic aganglionosis. A term newborn male presented with distal intestinal obstruction and underwent exploratory laparotomy at 3 days old after an ultrasound-guided contrast enema. Intra-operative frozen sections of full-thickness biopsies from each part of the colon and terminal ileum revealed no ganglion cells. Therefore, we performed a double-ended ileostomy. Although all final pathology reports confirmed the absence of ganglion cells, which were similar to those in the frozen sections, we were concerned about the problematic bowel because preoperative imaging (at 30 months of age) showed a dilated transverse colon. Therefore, we repeated the contrast enema, and the procedure showed a transition zone at the splenic flexure. An intra-operative frozen section of the normal caliber ascending colon was positive for ganglia in both layers, and then a pull-through operation with a protective ileostomy was performed. The final pathology report showed no ganglion cells in the rectum and transverse colon, whereas normal ganglion cells were observed in the sigmoid area. To date, approximately 30 cases of skip-segment Hirschsprung's disease have been reported; therefore, this phenomenon is rare but important. All patients who do not present with a typical picture of Hirschsprung's disease should be carefully evaluated, as the spectrum of this disease can alter the resected bowel length.

**Keywords:** Diagnosis, Hirschsprung's disease, contrast enema, segmental aganglionosis, skip segment Hirschsprung's disease.

Hirschsprung's disease is a congenital intestinal obstruction that results from the absence of the ganglion of the colonic submucosal and myenteric plexus. The most common sites of involvement are the rectum and distal sigmoid colon.<sup>(1)</sup> However, the disease sometimes extends proximally to other parts of the colon, and occasionally it also involves the small intestine. Typical Hirschsprung's disease is seen in 1 in 5,000 live births.<sup>(1)</sup> While the clinical manifestation of the disease usually presents with delayed passage of meconium, progressive abdominal distension, bilious vomiting, and abdominal pain in the neonatal period, some patients can present with chronic constipation later in life. However, there have been reports of a

rare form of Hirschsprung's disease called skip-segment Hirschsprung's disease (SSHD). This is defined as the occurrence of a normal ganglionated segment surrounded by proximally and distally aganglionic bowel segments.<sup>(2, 3)</sup> The definitive diagnosis of this phenomenon is challenging and requires a high index of suspicion. Herein, we report a case of SSHD, which was previously diagnosed with total colonic aganglionosis (TCA).

## Case report

A term male newborn presented with feeding intolerance and hematemesis at 22 h of life. He developed progressive abdominal distension with delayed passage of meconium. There was no family history of neonatal intestinal obstruction or Hirschsprung's disease. A plain abdominal radiograph revealed diffuse dilatation of the intestine (**Figure 1A**), and rectal irrigation was attempted, but it failed to remove the meconium. Further investigation

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into the cause of intestinal obstruction was requested, and ultrasonography of the colon with colonic irrigation was performed under ultrasound guidance. The procedure revealed a large amount of meconium in the sigmoid colon up to the dilated ileum (**Figure 1B**). During an abdominal ultrasound, several attempts at normal saline irrigation were performed, but only a minimal amount of meconium was passed. After the evaluation, an exploratory laparotomy was performed with serial biopsies, and the intra-operative frozen sections were examined. The operative finding showed an abrupt disproportion of caliber around the splenic flexure (between the transverse and descending colons (**Figure 1C**); therefore, the first two specimens were full-thickness sections of the transverse and descending colons. Both were reported to be devoid of ganglion cells. Later, biopsies of the ascending colon and multiple segments of the ileum were obtained. The intra-operative frozen section reported that the 14 cm proximal to the ileocecal valve had normal ganglion cells. Therefore, a double-end ileostomy without a skin bridge was performed at this site. Permanent section pathology reported no ganglion cells in the submucosal and myenteric plexus specimens from the descending colon, transverse colon, cecum, and terminal ileum 14 cm proximal to the ileocecal valve. The patient was discharged on postoperative day 20 with a double-end ileostomy. Definitive surgery was discussed with the parents during the follow-up period at the outpatient clinic.

At 30 months of age, in preparation for definitive surgery, a chest radiograph revealed abnormal dilatation of the suspected transverse colon. Based on this finding, a contrast enema was requested because the imaging findings were discrepant with previous pathological reports. During the investigation, the patient became distressed; therefore, the radiologist had to change the imaging modality to distal loopography. The study revealed a dilated transverse colon with a transition zone at the splenic flexure (**Figure 2**). The preoperative diagnosis was thus changed from TCA to long-segment Hirschsprung's disease. An intra-operative pathology consultation was requested to reevaluate ganglion cells in the transverse

and ascending colons. Both sites were ganglionated in the submucosal or myenteric plexuses; however, the transverse part was resected because of its chronic dilation and poor peristalsis. The definitive operation was an abdominal subtotal proctocolectomy with an endorectal pull-through and a protective ileostomy. Post-operative recovery was uneventful, and the patient was discharged on the 7<sup>th</sup> day after the operation.

The pathological report included the presence of ganglion cells in the sigmoid and transverse colons, as well as in biopsies of the ascending colon, but ganglion cells were absent in the rectum and descending colon (**Figure 3**). After the permanent section result was revealed, a pathologic review of the first operation's specimens was requested. The review revealed that no ganglion cells were observed in the frozen sections of the descending and transverse colons, the terminal ileum, or the undetermined cecum. In contrast, some permanent cuts of the ileum, cecum, and transverse colon had ganglion cells, whereas no ganglion cells were seen in the descending colon. Therefore, the final diagnosis for this patient was SSHD.

Eight weeks after the pull-through procedure, ileostomy closure was scheduled. An end-to-end anastomosis was completed via a peristomal incision. While there was moderate intra-abdominal adhesion, we decided to perform anastomosis without lysis of the interbowel loop adhesion because of the incision limitation. The patient developed postoperative partial intestinal obstruction following ileostomy closure. As his clinical condition had not improved 72 h after conservative management, an exploratory laparotomy with adhesion lysis and repair of the small bowel with a side-hole ileostomy was performed. After this operation, the post-operative condition was uneventful, and fecal content was excreted through the stoma and anus.

The patient underwent a side-hole ileostomy closure 12 weeks after the last operation. He was discharged from the hospital on post-operative day 8. During the follow-up period (the latest is 4 months after the last operation), he is doing well and has been stooling regularly once daily.

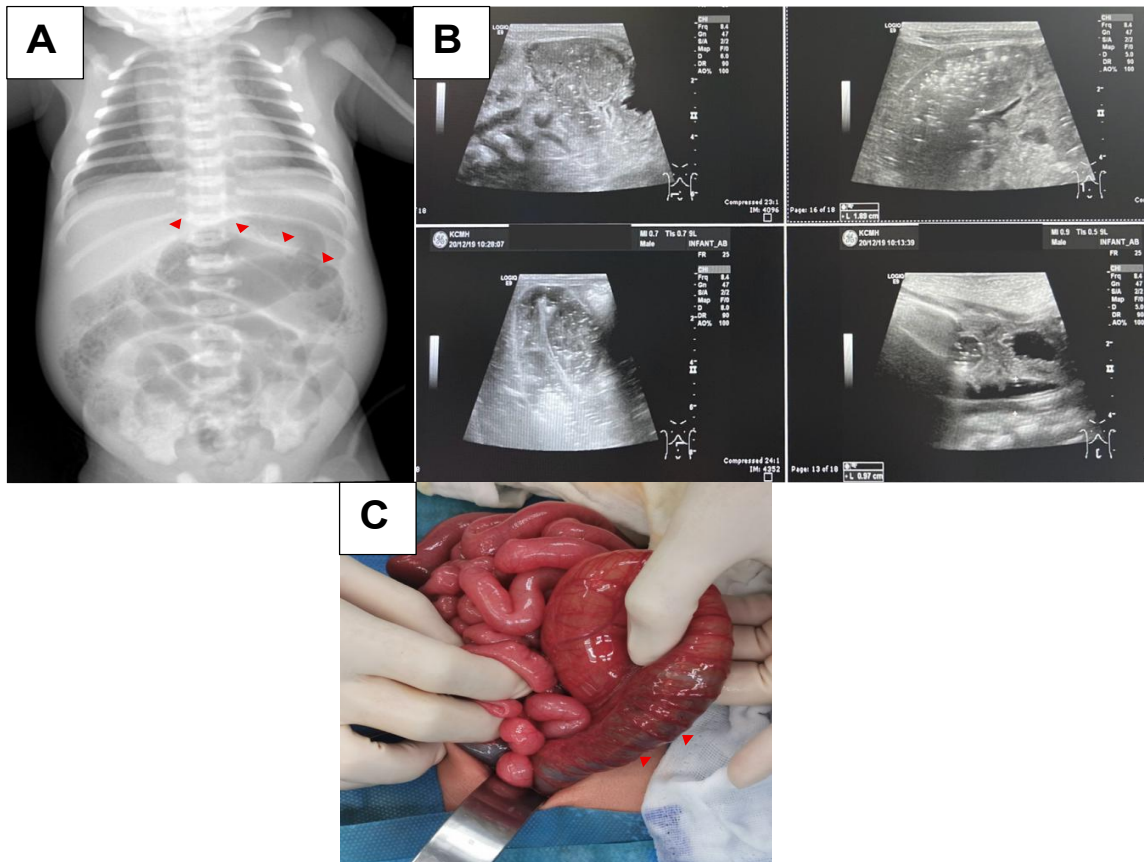


**Figure 1.** (A) A plain abdominal radiography showed diffuse dilatation of the intestine, (B) ultrasonography of the colon with colonic irrigation under ultrasound guidance shows a large amount of meconium in right, transverse, and left colon, (C) descending colon with a large amount of meconium.

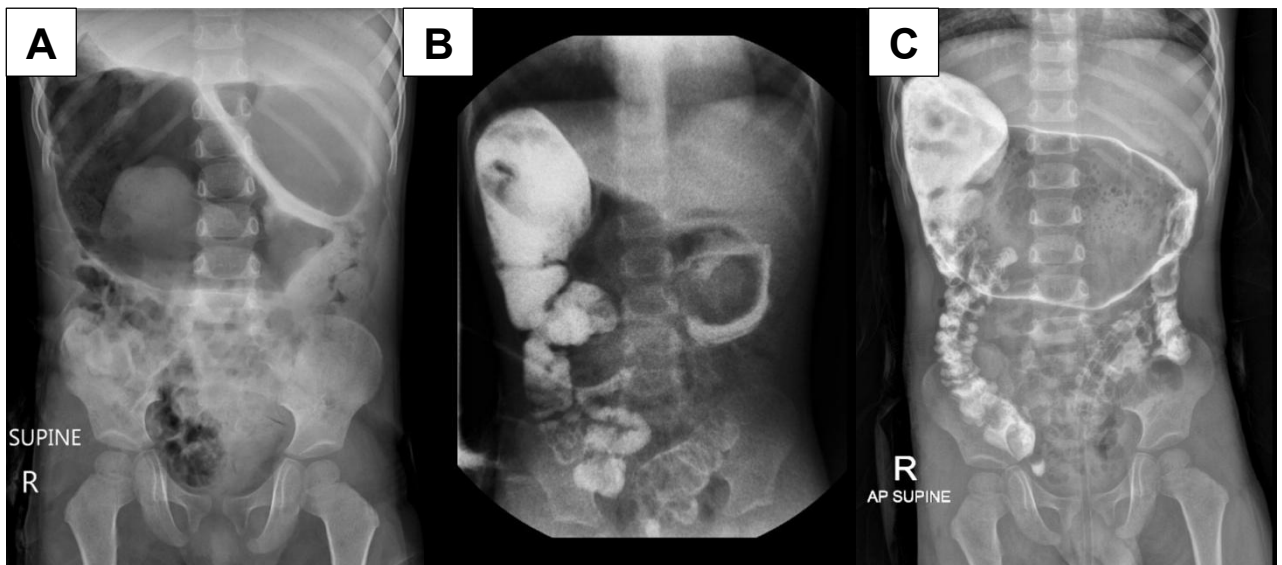


**Figure 2.** (A) plain abdominal radiograph prior to admission for definitive surgery, (B) contrast study via distal ileostomy, and (C) 24-hour imaging after contrast study revealed transition at splenic flexure.

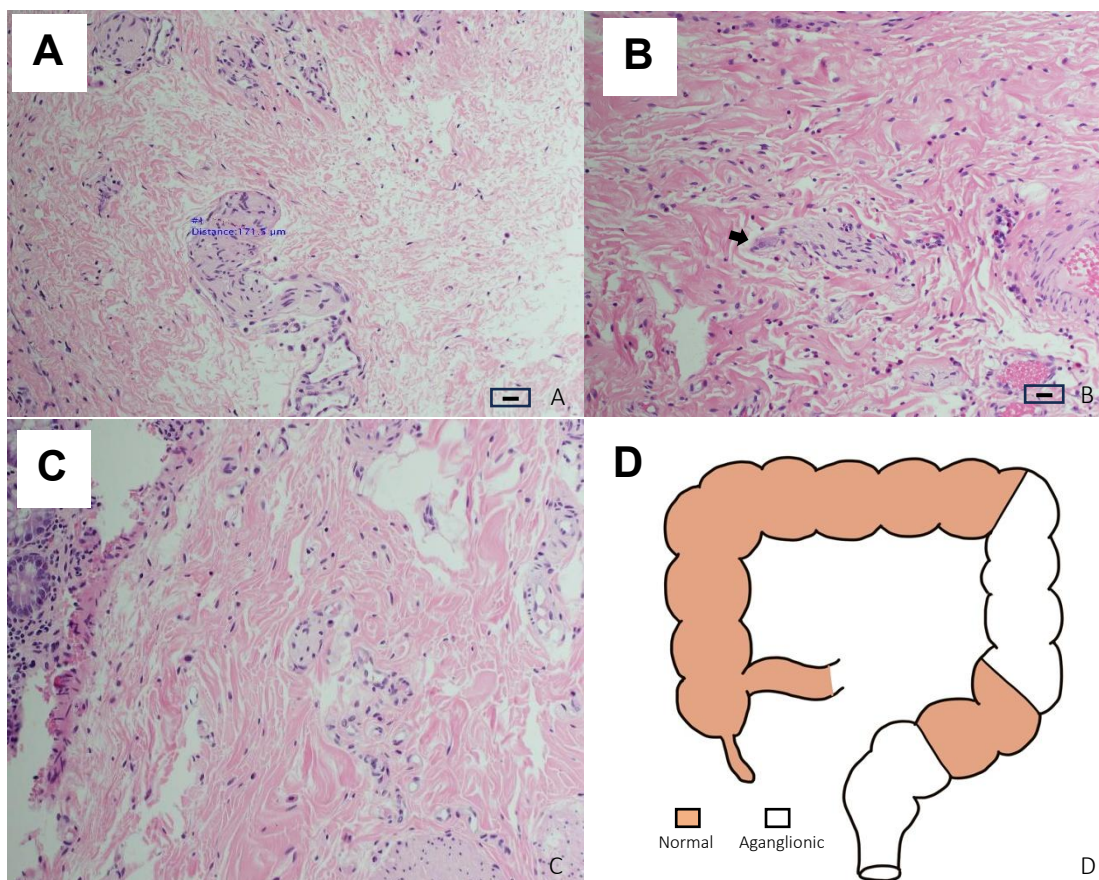


**Figure 3.** Specimen from the pull-through procedure (Magnification = 200X and scale bar = 20  $\mu$ m for all images). (A) Upper rectum – no ganglion cell with hypertrophic nerve trunks 171.5  $\mu$ m (hematoxylin and eosin stain), (B) sigmoid colon – normal ganglion cell (black arrow) (hematoxylin and eosin stain), (C) descending colon - no ganglion cells (hematoxylin and eosin stain), and (D) diagrammatic representation of the current case.

## Discussion

SSHD is a rare form of Hirschsprung's disease in which a normal ganglionic bowel occurs within the distal intestinal aganglionic intestine. To the best of our knowledge, there have been 47 patients in the English literature, including our report (**Table 1**). Coe A, *et al.* classified patients with SSHD into three groups: Group 1, a variant of TCA; Group 2, a skip segment located distal to the splenic flexure; and Group 3, other.<sup>(24)</sup> While our patient is likely to be in Group 2 as the skip lesion is in the sigmoid colon, the majority of patients are classified in Group 1 (32/47). Based on this observation, patients with a provisional diagnosis of TCA should be evaluated for skip lesions, as their management may be altered. According to Ahmad H, *et al.* they reported a case of TCA with a skip in the right colon that was managed by a pull-through of the right colon, which was anastomosed to the normal ileum after resection of the distal aganglionic colon and ileocecal segment. This surgical approach might help preserve bowel length and colonic function.

Due to the rarity of these variations, patients can be easily misdiagnosed, resulting in either overresection or inadequate resection of the intestine. Pediatric surgeons should keep this spectrum in mind in patients with unimproved symptoms after surgery<sup>(3)</sup> and in patients with disparate clinical and histopathology findings.<sup>(24)</sup> As seen in our case, the patient was initially diagnosed with TCA owing to the pathologic report of the first procedure's specimens. The patient later had discordant clinical findings, including a dilated transverse colon, which prompted us to reevaluate and investigate for a more accurate diagnosis. Zhou JL, *et al.* suggested that a 24-h delayed abdominal radiograph following a barium enema can serve as a good reminder of this rare phenomenon.<sup>(26)</sup> Our case did not undergo a contrast enema at the first presentation at the recommendation of the pediatric radiologist, as our patient was provisionally diagnosed with distal intestinal obstruction, probably meconium plug obstruction; therefore, colonic saline irrigation under ultrasound guidance would provide information and therapeutic benefit without radiation exposure.

**Table 1.** Summary of all cases of skip segment Hirschsprung's disease reported in the literature from 1954 to 2023 <sup>(4-31)</sup>

| References                           | Year | Sex | (N) patients | Location of skip segment                                                                     |
|--------------------------------------|------|-----|--------------|----------------------------------------------------------------------------------------------|
| Keefer GP.                           | 1954 | M   | 1            | Rectosigmoid with skip in sigmoid                                                            |
| Sprinz H.                            | 1961 | F   | 1            | TCA except skip in transverse colon                                                          |
| MacIver AG.                          | 1972 | M   | 1            | Rectosigmoid with skip in sigmoid                                                            |
| Martin LW.                           | 1979 | M   | 1            | TCA except skip in transverse colon                                                          |
| de Chadarevian J-P                   | 1982 | M   | 1            | TCA except skip in transverse colon                                                          |
| Yunis E.                             | 1983 | M   | 4            | TCA except skip in transverse colon                                                          |
|                                      |      | M   |              | TCA except skip in transverse colon                                                          |
|                                      |      | M   |              | TCA except skip in ascending colon                                                           |
|                                      |      | M   |              | TCA except skip in transverse colon                                                          |
| Taguchi T.                           | 1983 | M   | 1            | TCA except skip in ascending colon                                                           |
| Seldenrijk CA.                       | 1986 | M   | 2            | TCA with multiple skips                                                                      |
|                                      |      | F   |              | TCA with multiple skips                                                                      |
| Anderson KD.                         | 1986 | M   | 1            | TCA except skip in ascending colon                                                           |
| Kapur RP.                            | 1995 | F   | 3            | TCA except skip in ascending colon                                                           |
|                                      |      | F   |              | TCA except skip in caecum                                                                    |
|                                      |      | F   |              | TCA except skip in ascending colon and descending colon                                      |
| Yang H.<br>(Full Article in Chinese) | 2005 | M   | 3            | TCA with multiple skips                                                                      |
|                                      |      | M   |              | TCA with multiple skips                                                                      |
|                                      |      | M   |              | TCA with multiple skips                                                                      |
| Ziad F.                              | 2006 | M   | 2            | TCA except skip in transverse colon                                                          |
|                                      |      | F   |              | TCA except skip in caecum                                                                    |
| Oshio T.                             | 2008 | M   | 1            | TCA except skip in ascending colon                                                           |
| Puri P.                              | 2010 | M   | 1            | TCA except skip in transverse colon                                                          |
| Doi T.                               | 2011 | F   | 1            | TCA except skip in transverse colon                                                          |
| Castle S.                            | 2012 | M   | 1            | TCA except skip in transverse and ascending colon                                            |
| Burjonrappa S.                       | 2012 | M   | 1            | TCA except skip in caecum                                                                    |
| Skelly BL.                           | 2012 | M   | 1            | Total intestinal aganglionosis except stomach, 10 and 45 cm distal to duodenojejunal flexure |
| Moore SW.                            | 2013 | M   | 2            | Skip in descending colon                                                                     |
|                                      |      | M   |              | TCA with skip in right transverse, ascending colon and appendix                              |
| Erten EE.                            | 2014 | M   | 1            | TCA except skip in ascending colon and 2 cm aganglionic ileum 6 cm from terminal ileum       |
| Raghunath                            | 2014 | M   | 1            | TCA with skip in ascending colon and hepatic flexure                                         |
| Gross ER.                            | 2015 | M   | 1            | Skip in transverse and descending colon                                                      |
| Ruiz SL.                             | 2016 | M   | 1            | TCA except skip in caecum and ascending colon                                                |
| Coe A.                               | 2016 | M   | 2            | Distal rectal skip segment                                                                   |
|                                      |      | M   |              | Distal rectal skip segment                                                                   |
| Shenoy A.                            | 2019 | M   | 1            | Distal rectal skip segment                                                                   |
| Zhou JL.                             | 2019 | M   | 1            | TCA with skip in transverse colon                                                            |
| Yu Q.                                | 2021 | M   | 2            | Rectosigmoid with skip in sigmoid                                                            |
| Almumtin A.                          | 2021 | M   | 1            | TCA with skip in ascending, transverse, descending colon                                     |
| Ahmad H.                             | 2021 | M   | 1            | TCA except skin ascending colon, hepatic flexure, descending colon                           |
|                                      |      | M   |              | Skip in transverse colon                                                                     |

**Table 1.** (Cont.) Summary of all cases of skip segment Hirschsprung’s disease reported in the literature from 1954 to 2023 <sup>(4-31)</sup>

| References   | Year | Sex | (N) patients | Location of skip segment                                                                        |
|--------------|------|-----|--------------|-------------------------------------------------------------------------------------------------|
| Chang X.     | 2021 | M   | 3            | Aganglioneosis terminal ileum and rectosigmoid, Skip in Ascending, Transverse, Descending colon |
|              |      | M   |              | Skip in sigmoid                                                                                 |
|              |      | M   |              | Skip in upper rectum and sigmoid                                                                |
| Yang         | 2021 | F   | 2            | TCA except skip in ascending colon                                                              |
|              |      | F   |              | TCA except skip in ascending colon                                                              |
| Current case | 2023 | M   | 1            | Skip in sigmoid                                                                                 |

TCA, total colonic aganglioneosis.

Although this method of investigation could provide some clues, we had to admit that it was more challenging to map the anatomical disproportion than with the contrast enema. The patient underwent a contrast study for the second evaluation before definitive surgery, and an obvious transition zone at the splenic flexure was observed on 24-h imaging after the procedure. Therefore, 24-h imaging after a contrast enema can provide valuable information in typical Hirschsprung’s disease and its variants. Furthermore, laparoscopic evaluation in patients with nonclassic clinical features might offer care providers a clue to suspect skip-segment pathology. According to the study by Chang X, *et al.*, the authors detected isolated colonic narrowing in patients with clinical suspicion for TCA or long-segment Hirschsprung’s disease by carefully reviewing dynamic barium enema imaging and laparoscopically rechecking the suspected areas.<sup>(30)</sup> As the authors experienced one misdiagnosed patient, the latter two patients with SSHD could be managed and diagnosed correctly in the first operation.

The pathogenesis of SSHD remains unclear; however, several studies in animal models have been conducted. In typical Hirschsprung’s disease, the widely accepted etiology is an incomplete migration of vagal neural crest cells into the distal hindgut, which results in the absence of ganglion cells in the rectum.<sup>(32, 33)</sup> This rare spectrum refers to a normally ganglionated bowel surrounded proximally and distally by an aganglionic bowel, which does not correlate with the mentioned theory. Several hypotheses have been proposed to explain the origin of ganglion cells present in the distal bowel in SSHD. Recent experiments in mice by Yu Q, *et al.* revealed that mesenteric neural crest cells (mNCCs), a subset of vagal neural crest cells (vNCCs), could explain SSHD.<sup>(27)</sup> This model demonstrates that the majority of vNCCs migrate along the gut, whereas other vNCCs (i.e., mNCCs) migrate through the mesentery. In considering SSHD,

failure of migration along the gut occurs; however, mNCCs continue to migrate and give rise to ganglia in the distal segment. The second proposed hypothesis by Nishiyama *et al.* suggests that most enteric ganglia are derived from the transmesenteric migration of vNCCs, promoted by RET/GDNF signals.<sup>(34)</sup> Therefore, genetic forms of Hirschsprung’s disease that reduce RET/GDNF signaling may result in the inadequate expansion of NCC migration and result in SSHD. Another proposed theory for the pathogenesis of SSHD is that sacral NCCs colonize the distal gut independent of vNCC migration.<sup>(35)</sup> At present, there is no single hypothesis that can explain all SSHD cases, and each patient might probably have a different etiology.

In view of the pathology, the absence of ganglion cells in full-thickness rectal biopsies is the gold standard for diagnosis.<sup>(36)</sup> Other features for diagnosis include hypertrophic nerves, increased acetylcholinesterase staining, and absence of calretinin staining.<sup>(37)</sup> Patients with ambiguous clinical and/or pathology findings should be promptly reevaluated, as physicians might miss the presence of skip lesions. In this report, the first laparotomy revealed a transition zone at the splenic flexure. However, the frozen section of the dilated transverse colon reported an absence of ganglion cells in the Meissner and Auerbach plexuses. In addition, no ganglion cells were observed in any frozen or permanent sections of colonic tissue; therefore, the diagnosis was TCA. However, the imaging study before definitive treatment again revealed a markedly dilated transverse colon and a transition point at the splenic flexure. We decided to rebiopsy the transverse and ascending colon, and both revealed normal ganglion cells. This can encourage surgeons to perform a rebiopsy when the clinical findings do not correlate with the pathological reports. Another method to increase accuracy in frozen sections is the use of acetylcholinesterase staining,<sup>(24)</sup> but this evaluation is not available in every hospital, including ours.

## Conclusion

SSHD is a rare variant of Hirschsprung's disease that can be easily misdiagnosed, leading to reevaluation and reoperation. As most patients with SSHD have clinical features of long-segment Hirschsprung's disease and TCA, physicians should carefully investigate and keep this variation in mind. In addition, patients with ambiguous clinical findings of all investigations performed should be promptly reviewed and reinvestigated to make an accurate diagnosis. Multiple biopsies to map the ganglionic bowel should be performed for all patients because this helps preserve the length of colonic tissue in the definitive surgery. Furthermore, the most effective management is a pull-through procedure with resection of the aganglionic bowel.

## Author contributions

NS contributed substantially to the study concept and design, data acquisition, literature review, data analysis and interpretation, and manuscript drafting. PV contributed substantially to data acquisition. NW contributed substantially to data and histopathology image acquisition. NS and PV edited the manuscript critically for important intellectual content. All authors approved the final version submitted for publication and take responsibility for statements made in the published article.

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## Conflict of interest statement

All authors have completed and submitted the International Committee of Medical Journal Editors Uniform Disclosure Form for Potential Conflicts of Interest. All authors declare that they have no conflicts of interest.

## Data sharing statement

Data generated or analyzed for the present report are included in this published article. Further details are available from the corresponding author on reasonable request after deidentification of the patient whose data are included in the report.

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