

Skip Segment Hirschsprungs Disease: Avoiding the Potential Pitfall of a Failed Pull-through Procedure

Case Report

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Received: November 03, 2017; **Published:** December 05, 2017

Abstract

Background/Purpose: SSHD is rare, with 29 cases since 1954. It is defined by ganglionated segments in between regions of aganglionosis. We present a case, discuss the literature and define management strategies for SSHD.

Methods/Results: A newborn male had delayed passage of meconium, bilious emesis, and abdominal distension. Barium enema showed an inverted ratio of the sigmoid to rectum, without clear transition zone. Suction rectal biopsy suggested Hirschsprungs disease. Laparoscopic Soave Procedure was performed. Intra-operative seromuscular biopsies revealed numerous ganglion cells at the proximal sigmoid, thus the pull through was completed 3 cm proximal to that point. An anastomotic margin biopsy questioned presence of ganglion cells, and permanent section confirmed aganglionosis. Repeat pull-through was performed and an additional 10 cm of colon was resected. The patient did well. IRB approval was obtained.

Conclusion: SSHD is rare and likely under-diagnosed. No consensus exists regarding the optimal approach to biopsies to avoid this potential pitfall. Close cooperation between surgery and pathology is required. Although other authors suggest aggressive sampling techniques, we recommend full thickness frozen section biopsies of the anastomotic margin and close monitoring of the patient post-operatively to permit early diagnosis. Patients with persistent post-operative symptoms should be re-biopsied to rule out SSHD.

Keywords: *Soave; Hirschsprungs; Skip Segment; Neonatal; Pathology*

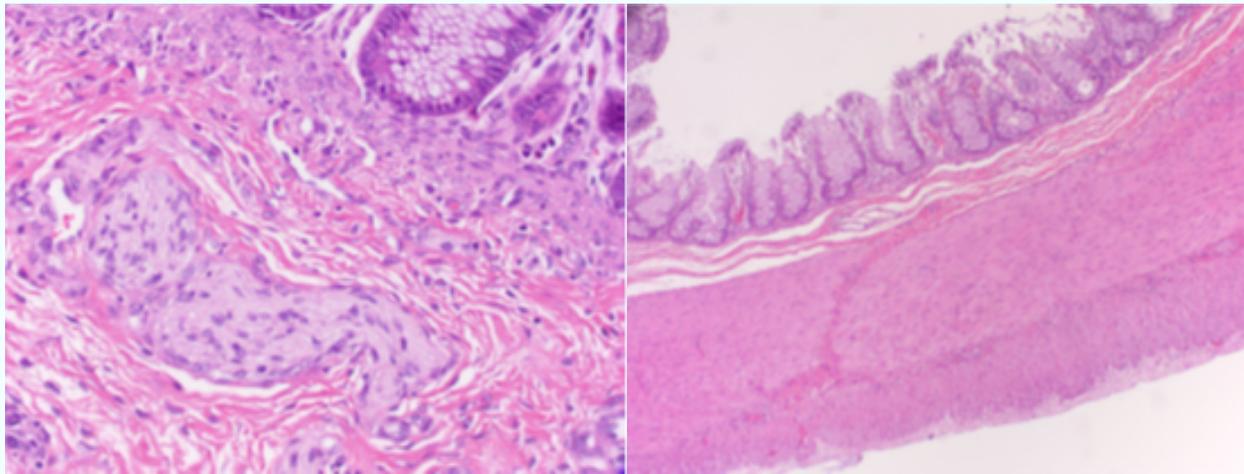
Introduction

Hirschsprungs Disease typically is defined as intestinal aganglionosis extending continuously from the distal rectum to a variable proximal extent. It most commonly involves the rectosigmoid but may rarely affect the entire gastrointestinal tract, necessitating a small bowel transplant. This abnormality is explained by the early arrest of the neuroblast migration in the cranio-caudal direction that starts in the esophagus during embryogenesis. Skip Segment Hirschsprungs Disease (SSHD) may include single or multiple segments of ganglionated bowel in between segments of aganglionosis [1]. Subsequently, SSHD poses a diagnostic and surgical dilemma and should be considered during treatment failure, recurrent enterocolitis or refractory constipation. While proximal skip lesions are still possible, these complications may be diminished sending the final margin at the time of the anastomosis to confirm presence of ganglion cells. It is a rare phenomenon, first reported in 1954, with only 29 cases reported since then. We reported a case of SSHD diagnosed by intraoperative biopsies during a laparoscopic Soave pull-through procedure and confirmed on permanent histology.

Citation: Dr. Neville H., et al. "Skip Segment Hirschsprungs Disease: Avoiding the Potential Pitfall of a Failed Pull-through Procedure". EC Paediatrics SI.01 (2017): 25-28.

Case

A 10-day-old male, born at 36 weeks via C-section, weighing 3.29 Kg, had delayed passage of meconium at 40 hours of life. He had bilious emesis, abdominal distension, and dilated loops of intestine on abdominal x-ray. Due to his prematurity, the neonatologist initially diagnosed the baby with necrotizing enterocolitis. The neonate continued to have symptoms despite medical therapy, and a contrast enema was performed which revealed an inverted rectal to sigmoid ratio. The transition zone was not identified. Suction rectal biopsy demonstrated hypertrophic nerves and absence of ganglion cells (GC) (see Figure 1). The patient was taken to the operating room for a laparoscopic-assisted Soave procedure. Seromuscular biopsies showed no GC in the rectum or distal sigmoid, but GC were present in the proximal sigmoid (see Figure 2) without hypertrophied nerve trunks. This site was chosen for the pull through and the operative team proceeded to mobilize the left colon and divide the inferior mesenteric artery to further aid in mobilization. Rectal mucosectomy and endorectal pull-through were performed. The colo-anal anastomosis was fashioned 3 cm proximal to the proximal sigmoid biopsy site that was positive for GC. The resected colon was sent for a permanent section, while the anastomotic margin biopsy was sent for frozen section. The frozen section returned without hypertrophic nerve trunks, however only questionable immature GC at the anastomosis. Due to indeterminant pathology results, the decision was made to wait for the final pathology before extending our resection. Final pathology confirmed the absence of GC at the anastomosis and the presence of SSHD. On post-operative day 2, the patient underwent a redo laparoscopic assisted Soave procedure with completion of pull through to bowel containing normal ganglion cells. A laparoscopic appendectomy was performed and frozen sections revealed GC. Seromuscular biopsies guided the colonic resection, which extended to the distal transverse colon where GC were present. This margin was used for the colo-anal anastomosis, and the final pathology confirmed the presence of GC at the anastomosis. The patient had an uneventful post-operative course, tolerated breast milk, regained regular bowel function, and was discharged home. At the two-week follow-up, the baby was stooling 5 - 7 times a day on a standard formula diet. He is now 14 months old and continues to do well with daily bowel movements with no episodes of enterocolitis.



a) distal margin- (85.34 μ m)- no ganglion cells

b) proximal margin- no ganglion cells

Figure 1: Hypertrophic nerve trunk.

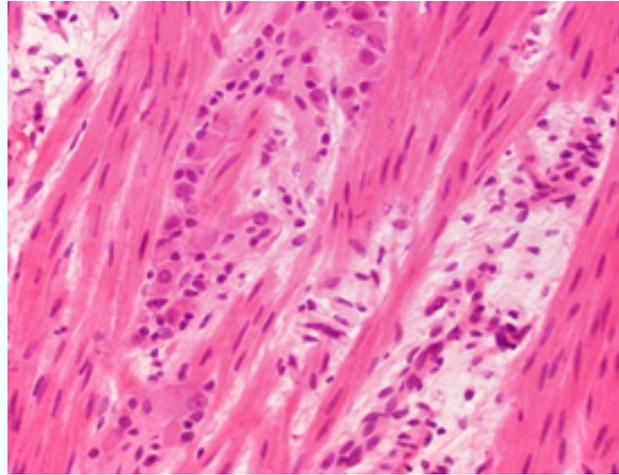


Figure 2: Proximal colon biopsy- ganglion cells present.

Discussion

SSHD is a rare variant of Hirschsprungs Disease with single or multiple ganglionated segments of varying length between segments of aganglionosis. Only 29 cases have been reported in the literature. No clear embryological explanation exists for this pathology [1]. However, a murine model theory suggests that this extra-mural neuroblast migration along the mesenteric border of the colon may have been coinciding with the anticipated intra-mural path of neuroblasts [2]. The initial clinical presentation is similar to the classic Hirschsprungs disease, but there is an inconsistent radiological and pathologic correlation concerning the transition zone [3]. Most of the reported cases of SSHD were either long segment disease or total colonic aganglionosis [1]. In our case, we had SSHD with long segment disease.

Our initial resection and anastomosis were limited to the proximal sigmoid while the remaining disease was present proximally. We elected to wait for the final pathological analysis before resecting further colon to exclude the possibility of immature ganglion cells that could be managed conservatively without resection, as these mature over time with improvement in function without the need for surgical therapy [4]. In our second operation, an appendectomy was performed, serving as a full thickness proximal colonic biopsy. It is important to note that this step is only useful to confirm the presence of GC and cannot predict total colonic aganglionosis in the absence of GC, as there have been multiple reports of appendiceal aganglionosis with normal colonic GC [5]. Other aggressive measures include routine biopsies of the entire colon and terminal ileum to avoid missing a skip segment prior to the pull-through [6,7].

We believe that this approach is not justified as an alternative to our current standard of care as it poses an extra morbidity risk from excessive sampling. As the benefit is only limited to a very small population with a rare disease process we suggest a higher index of suspicion in children with persistent constipation post-operatively who fail to improve with conservative measures, and in post-operative patients with multiple episodes of enterocolitis. These patients might benefit from further sampling with intestinal biopsies to rule out the possibility of SSHD.

Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

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