CASE SERIES

Early Symptomatic Improvement After Full-Thickness Rectal Biopsy in Child with Suspected Short-Segment Hirschsprung Disease: A Case Series

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ABSTRACT

Introduction: Full-thickness rectal biopsy (FTRB) is conventionally performed to obtain tissue for identifying aganglionic segments in Hirschsprung disease. This procedure is technically similar to the posterior rectal myectomy, which has been reported to be beneficial in patients with short-segment Hirschsprung disease. In this report, we aim to describe a case series of three babies with chronic constipation since birth who demonstrated clinical improvement following FTRB. **Case series:** Three baby with suspected HSCR presented with chronic constipation and abdominal distension since the first days of life. The patients were aged 65 days (weight 6,000 g), 35 days (weight 6,200 g), and 35 days (weight 4,100 g). Radiological evaluation included plain abdominal radiography and water-soluble contrast enema, which revealed colonic dilatation with features suggestive of a transition zone. All patients underwent anal examination under general anesthesia to exclude anatomical anorectal abnormalities prior to biopsy. A FTRB was performed using an elliptical incision, with the proximal extent of the biopsy located approximately 6 cm above the dentate line. Histopathological using hematoxylin and eosin staining confirmed aganglionosis in all cases. Postoperatively, all patients showed clinical improvement, with spontaneous daily bowel movements observed by postoperative day two to three and resolution of abdominal distension. **Conclusion:** FTRB by elliptical incision extends from the serosal to the muscular layers and may relieve non-relaxing aganglionic segments in short-segment Hirschsprung disease. Apart from its diagnostic role, FTRB may serve a therapeutic role in selected cases, warranting further evaluation in larger studies.
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INTRODUCTION

Constipation is a common condition in children and is broadly classified into functional constipation (FC) and organic constipation (OC). Functional constipation, defined as constipation without an identifiable organic cause, accounts for up to 95% of pediatric cases. The remaining 5% are attributed to organic etiologies, including Hirschsprung disease (HSCR), anorectal anomalies, and neuromuscular, metabolic, or endocrine disorders. Failure to pass meconium within the first 48 hours of life should raise clinical suspicion for HSCR. Childhood constipation is among the most frequent reasons for pediatric outpatient visits worldwide and represents an increasing public health concern. Furthermore, chronic constipation adversely affects psychological well-being, social functioning, academic performance, and overall quality of life for both children and their caregivers. A substantial proportion of children with constipation respond poorly to standard medical therapy or experience early symptom recurrence. In cases where constipation is caused by colonic neuromuscular dysfunction, such as aganglionosis, surgical intervention is often associated with favorable functional outcomes. The classic clinical triad of HSCR includes abdominal

distension, vomiting, and delayed passage of meconium beyond 24 hours after birth. Given the wide differential diagnosis of pediatric constipation, persistent symptoms despite adequate medical management should prompt further evaluation for underlying organic causes, particularly HSCR (1, 2).

Hirschsprung disease is characterized by the congenital absence of ganglion cells in segments of the colon, resulting in functional bowel obstruction. Constipation is the predominant clinical manifestation, and approximately 95% of cases are diagnosed during infancy. HSCR is among the most common congenital gastrointestinal disorders, with a global prevalence of approximately 1 in 5,000 live births and a male predominance of nearly 4:1. In Indonesia, the reported incidence is higher, estimated at 1 in 3,250 live births. Diagnostic modalities for HSCR include contrast enema, anorectal manometry, rectal suction biopsy, and full-thickness rectal biopsy (FTRB). Contrast enema is commonly used preoperatively to assess the extent of the affected bowel; however, its diagnostic accuracy is limited, and intraoperative biopsy is frequently required to accurately determine the level of resection (1, 2).

Definitive diagnosis of HSCR relies on histopathological confirmation obtained through rectal biopsy, with tissue samples taken at least 2 cm above the dentate line. The absence of ganglion cells, accompanied by hypertrophy of nerve fibers within the submucosal and myenteric plexuses, is diagnostic. Hematoxylin–eosin staining remains the standard method, with adjunctive stains such as acetylcholinesterase and calretinin used to support diagnostic accuracy. Rectal biopsy may be performed using suction, punch, or open techniques. In our institution, FTRB is routinely employed to confirm suspected HSCR. This technique yields larger and deeper tissue samples, enabling reliable evaluation of both the submucosa and myenteric plexus, thereby reducing the risk of false-negative results. While FTRB requires general anesthesia, it is considered safe and has not been associated with significant complications in our experience. The standard definitive surgical management in our department is a pull-through procedure (2, 3).

As a tertiary referral center for pediatric patients with suspected HSCR, our institution encounters a substantial number of complex cases. This case series aims to report clinical improvement in constipation following FTRB in patients with HSCR, with particular emphasis on short segment Hirschsprung disease. The FTRB technique described closely resembles the posterior rectal myectomy approach previously reported for HSCR. Herein, we present three cases of babies with chronic constipation and long-term laxative use who demonstrated symptomatic improvement following FTRB.

CASE SERIES

We report three cases of HSCR managed with FTRB between January and December 2024 at Dr. Zainoel Abidin Hospital (RSUDZA), Aceh, a tertiary pediatric surgery referral center serving approximately 5.5 million inhabitants. Clinical data were retrieved from medical records, including age, sex, presenting symptoms, radiological findings, operative details, histopathological results, and immediate postoperative outcomes. All patients underwent plain abdominal radiography and contrast (barium) enema, which demonstrated colonic dilatation with a transition zone.

Case 1

The first case was a 65-day-old female baby weighing 6,000 grams, presenting with an inability to defecate since birth that required repeated use of laxatives and enemas. Physical examination revealed abdominal distension, while other systemic examinations were unremarkable. A plain abdominal X-ray showed diffuse air distension of bowel loops. Contrast enema demonstrated a transition in bowel caliber, raising suspicion of Hirschsprung disease (Figure 1). Preoperatively, intravenous fluids were administered, and prophylactic antibiotics (ceftriaxone 200 mg every 12 hours) were given. Under general anesthesia, anal examination excluded anatomical abnormalities. A full-thickness rectal biopsy was performed using an elliptical incision at 6 cm above the suspected transition zone. Histopathological examination with hematoxylin–eosin staining confirmed the presence of aganglionic bowel. Postoperatively, the patient recovered uneventfully and was discharged on postoperative day one. On follow-up, bowel function improved markedly, with daily spontaneous defecation beginning on the second to third postoperative day, accompanied by resolution of abdominal distension. The patient's condition improved within 1 week, 1 month to 3 months of follow-up at the polyclinic.

Case 2

The second case was a 35-day-old baby weighing 6,200 grams who presented with an inability to defecate since birth, requiring intermittent rectal stimulation or always need for laxatives, rectal washout and was accompanied by abdominal distension. Physical examination revealed abdominal bloating without other systemic abnormalities. Plain abdominal radiography demonstrated bowel gas distension. Contrast enema showed radiological features consistent with short-segment Hirschsprung disease, characterized by minimal distal narrowing without a clearly demarcated transition zone (Figure 1). Preoperative management included intravenous fluid administration and prophylactic antibiotics following the same protocol as in Case 1. Under general anesthesia, anal examination excluded anatomical anorectal abnormalities. A full-thickness rectal biopsy was performed using an elliptical

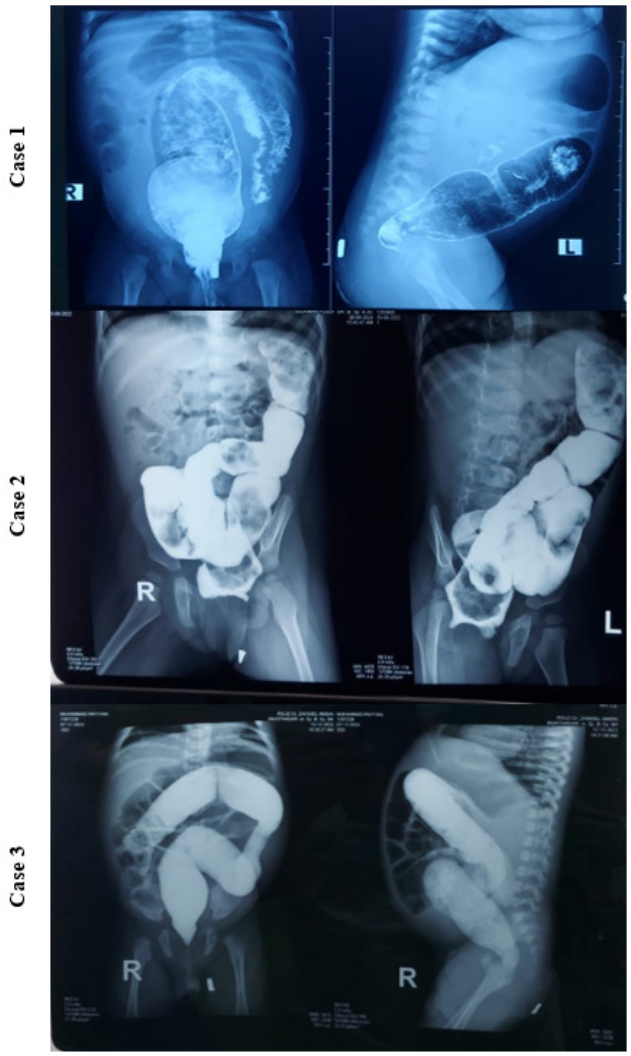


Figure 1: Water-soluble contrast enema (anteroposterior and lateral views) in three cases of Hirschsprung disease. All cases show a relatively narrowed rectum with proximal colonic dilatation, resulting in a reduced rectosigmoid caliber relationship, consistent with short segment Hirschsprung disease.

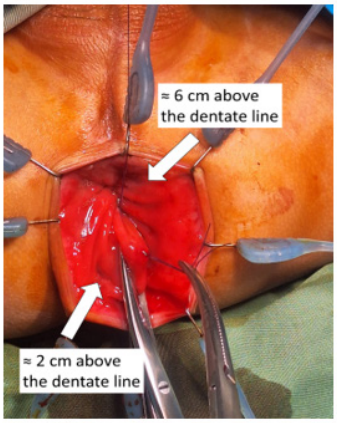
incision at approximately 6 cm above the dentate line. Histopathological examination with hematoxylin–eosin staining confirmed the presence of aganglionic bowel. The postoperative course was uneventful. The patient demonstrated marked improvement in bowel function, with spontaneous daily defecation occurring on postoperative day two to three. Abdominal distension resolved, and no immediate postoperative complications were observed. During the 1- 3-month follow-up, the patient had complete resolution of symptoms

Case 3

The third case involved a 35-day-old baby weighing 4,100 grams who presented with failure to pass stool since birth, associated with progressive abdominal distension. Clinical examination showed abdominal bloating, while other systemic findings were within normal limits. Plain abdominal radiography revealed diffuse bowel gas distension. Contrast enema demonstrated imaging findings suggestive of short-segment HSCR (Figure 1). The patient received standard preoperative preparation, including intravenous fluids and prophylactic antibiotics. An anal examination under general anesthesia revealed no anatomical abnormalities. A full-thickness rectal biopsy was performed using an elliptical incision at the posterior rectum, approximately 6 cm above the dentate line (Figure 2). Histopathological evaluation using hematoxylin–eosin staining confirmed aganglionosis. Postoperatively, the patient recovered without complications and showed significant clinical improvement. Regular spontaneous bowel movements were observed beginning on the second to third postoperative day, with complete resolution of abdominal distension. We also monitor the patient's condition for 1 week to 3 months after surgery in the polyclinic with improvement in the patient's condition.

Surgical Technique

Patient positioned prone or in dorsal lithotomy under general anesthesia. Perform anal examination to exclude anorectal abnormalities. Use contrast enema



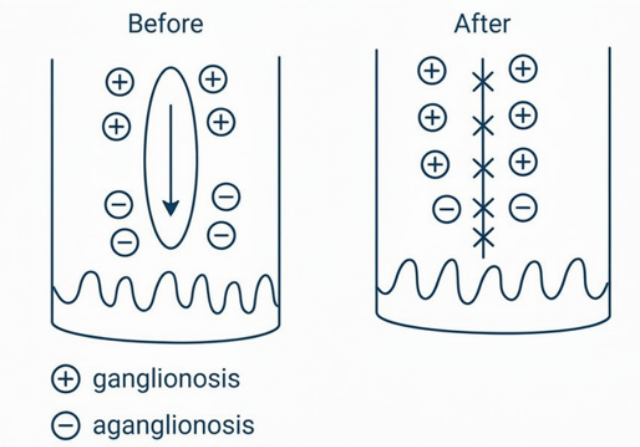


Figure 2: A Intraoperative view of full-thickness rectal biopsy. The proximal margin of the elliptical incision corresponds to approximately 6 cm above the dentate line, while the distal margin corresponds to approximately 2 cm above the dentate line, allowing excision of aganglionic rectal tissue. B. Ilustration Eliptical incision for Full thickness rectal biopsy with extended incsion

to estimate aganglionic segment length (distance from dentate line to transition zone). A posterior elliptical incision starts 2 cm above the dentate line and extends to the suspected ganglionic area according to contrast enema findings. If the aganglionic segment is 3 cm, the full-thickness rectal biopsy is extended to 5 cm. If the aganglionic segment is 6 cm, the full-thickness biopsy is extended to 8 cm. Full-thickness from mucosa to serosa, carefully excise full-thickness aganglionic tissue and submit for histopathology (H&E staining to confirm aganglionosis). Close defect with interrupted seromuscular sutures (4-0 or 5-0 absorbable Vicryl) approximating proximal to distal edges to achieve rectal relaxation while avoiding stenosis. Postoperative care monitor for bleeding or stenosis; expect spontaneous defecation by postoperative days 2-3 in short-segment Hirschsprung disease cases.

DISCUSSION

Constipation is a common problem in children, but organic causes account for only a small proportion of cases and include Hirschsprung disease (HSCR), anorectal malformations, neuromuscular disorders, and metabolic–endocrine conditions. HSCR has an estimated incidence of 1 in 5,000 live births and remains one of the most frequent causes of neonatal intestinal obstruction. In approximately 75–80% of patients, aganglionosis is confined to the rectosigmoid region, commonly referred to as short-segment HSCR. Short-segment and ultrashort-segment variants often present later, sometimes beyond infancy, with chronic refractory constipation rather than classic neonatal obstruction (2).

The diagnosis of HSCR in the present cases relied on histopathological evaluation of rectal specimens obtained via full-thickness rectal biopsy (FTRB). Rectal biopsy remains the gold standard for HSCR diagnosis, and FTRB provides larger and deeper tissue samples, allowing reliable assessment of both the submucosal (Meissner) and myenteric (Auerbach) plexuses. Compared with rectal suction biopsy (RSB), FTRB reduces the risk of inadequate submucosal sampling, which has been reported in up to 10–30% of RSB specimens. This advantage is particularly relevant in suspected short-segment or ultrashort-segment HSCR, where limited distal involvement may be missed by superficial or insufficient biopsies. The main drawback of FTRB is the need for general anesthesia and an operating-room setting, whereas RSB can be performed under sedation as a less invasive outpatient procedure. Adequate diagnostic accuracy requires specimens containing sufficient submucosa; recent series emphasize that full-thickness or deep submucosal samples, together with hematoxylin–eosin (HE) and adjunct immunohistochemical stains (e.g., calretinin, S-100), provide high diagnostic yield across all ages. Rectal biopsies are typically taken at least 2 cm above the dentate line to avoid the physiologically hypoganglionic distal rectum, but excessively proximal sampling may fail to detect disease limited to a very short distal segment (1, 2).

Histopathological confirmation of HSCR relies on demonstrating absence of ganglion cells and, in most cases, hypertrophy of nerve trunks on HE staining, supported by calretinin immunohistochemistry when indicated. In the present series, HE staining consistently revealed aganglionosis in the distal rectum. Biopsy specimens were obtained from the posterior rectal wall, approximately 2–6 cm above the dentate line, with levels guided by contrast enema findings and the suspected transition zone. Recent data suggest that the combination of radiologic transition zone on contrast enema and histology improves diagnostic confidence, although barium enema alone has a positive predictive value of about 0.9 and a lower negative predictive value. In our patients, the absence of a clearly demarcated histologic transition zone, together with distal aganglionosis limited to the rectum or rectosigmoid, supported the diagnosis of short-segment HSCR (3, 4).

Anorectal myectomy of the distal internal sphincter (Lynn procedure and its modifications) has been reported as an effective treatment option for ultrashort-segment HSCR and refractory functional outlet obstruction, with relatively low operative risk and minimal preoperative preparation. Posterior anorectal myectomy involves a longitudinal or transverse incision in the posterior anal canal and rectum, followed by selective resection of the internal sphincter and a strip of distal rectal muscle to relieve the functional obstruction while preserving the mucosa. Several series have shown good short- and medium-term outcomes, with improvement in constipation in 70–80% of children and low rates of major perioperative complications. However, important limitations of anorectal myectomy include the need for a preliminary biopsy to exclude more proximal or long-segment aganglionosis, the technical challenge of achieving an adequate myectomy length in very short segments, and the risk of only temporary symptom relief, with reported recurrence rates ranging from approximately 20% to 50% and a proportion of patients ultimately requiring definitive pull-through procedures. Additional concerns include the potential for sphincter dysfunction, soiling, or anal stenosis due to scarring, as well as a theoretical risk of Hirschsprung-associated enterocolitis (HAEC) if residual aganglionic or dysmotile segments persist (3-5).

The posterior rectal myectomy described by Lynn and later series shares conceptual similarities with the approach used in our patients, in that both target abnormal rectal neuromuscular tissue and aim to improve anorectal relaxation. In our series, however, FTRB was performed through an elliptical posterior incision, and the resected segment encompassed the full

thickness of the rectal wall over the aganglionic region, rather than a more superficial muscular strip. While FTRB and anorectal myectomy are distinct procedures, both may influence anorectal function by reducing outlet resistance. A practical limitation of classical anorectal myectomy is that the length of muscle resection rarely exceeds 5–6 cm, which may be insufficient in cases with more extensive short-segment involvement. In contrast, FTRB with an extended posterior incision allows excision of the full aganglionic segment (approximately 2–8 cm in our patients), which could theoretically reduce fecal stasis more effectively than superficial myectomy, though comparative data are lacking. Potential disadvantages of this extended FTRB include the risks of sphincter injury, posterior rectal scarring, and late stenosis, which must be carefully balanced against the possible therapeutic benefit (3-5).

In this case series, constipation resolved rapidly after FTRB, with clinical improvement already evident by the third postoperative day. Parents reported restoration of daily spontaneous bowel movements and resolution of abdominal distension, findings that were corroborated by physical examination at discharge. At follow-up evaluations in the outpatient clinic at 1 week, 1 month, and 3 months postoperatively, patients who attended scheduled visits maintained regular bowel habits, soft stool consistency, and absence of abdominal distension, without evidence of soiling or anal stenosis on examination. For patients who did not present to the outpatient clinic, the authors contacted parents by telephone; according to parental reports, these children experienced sustained improvement in defecation frequency and abdominal symptoms, with no episodes suggestive of HAEC or need for readmission during the initial follow-up interval. These observations support the possibility that FTRB with an extended posterior incision may provide not only diagnostic information but also short-term functional benefit in selected children with short-segment HSCR and severe constipation (3, 5).

We acknowledge the similarities between FTRB and posterior rectal myectomy (Lynn procedure), particularly in terms of targeting the distal aganglionic or dysmotile segment and aiming to improve anorectal relaxation. The key difference in our technique is the removal of the full-thickness distal aganglionic segment over a length of approximately 2–8 cm, rather than a limited muscular strip, which may enhance decompression while we sought to avoid circumferential scarring that could predispose to stenosis. At the same time, FTRB with an extended incision carries recognized risks, including potential sphincter damage, posterior fibrosis, symptom recurrence, and, in some cases, the later need for definitive pull-through surgery, similar to the long-term experience reported with myectomy. Current reviews of HSCR management emphasize that myectomy and related sphincter-targeted procedures are mainly reserved for ultrashort-segment disease or selected functional outlet disorders, and should not replace standard pull-through techniques in patients with more extensive aganglionosis. In line with this, we consider FTRB in our series as a combined diagnostic and potential therapeutic intervention for short-segment HSCR in carefully selected cases, rather than as a definitive alternative to pull-through

CONCLUSION

Full-thickness rectal biopsy performed using an elliptical incision allows effective sampling of aganglionic segments in short segment HSCR. In addition to its established diagnostic role, this approach may be associated with early symptomatic improvement in selected patients. Further studies involving larger cohorts and longer follow-up are required to confirm its potential therapeutic value and to define its role in clinical practice.

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REFERENCES

1. Wester T, Pakarinen M, Rintala R. Long-term outcomes of anorectal malformations and Hirschsprung disease. In: Pediatric colorectal and pelvic reconstructive surgery. Boca Raton: CRC Press; 2020. p. 127-36.
2. Mueller JL, Hotta R. Current and future state of the management of Hirschsprung disease. World J Pediatr Surg. 2025;8(1):e000860. doi:10.1136/wjps-2024-000860.
3. De Caluwй D, Yoneda A, Akl U, Puri P. Internal anal sphincter achalasia: outcome after internal sphincter myectomy. J Pediatr Surg. 2001;36(5):736-8. doi:10.1053/jpsu.2001.22948.
4. Gulcin N, Ersoy F, Erkoc G, Anadolulu AI, Caglar M, Ozkanli SS, et al. Full-thickness rectal biopsies in diagnosis of Hirschsprung disease: 13 years of experience of a single center. North Clin Istanb. 2025;12(1):138-43. doi:10.14744/nci.2025.97455.
5. Achugatla S, Redkar R, Singh S, Thakur S. Transanal rectal myomectomy following Martin left colon patch procedure—a proven therapeutic surgical technique with literature review. World J Colorectal Surg. 2025;14(2):76-81. doi:10.4103/wjcs.wjcs_9_25.