Surgical Management of Hirschsprung's Disease: A Prospective Study of 78 Cases

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Abstract ***Introduction:*** *Hirschsprung's disease (HD) is a congenital neurodevelopmental disorder characterized by the absence of ganglion cells in the distal bowel, resulting in functional intestinal obstruction. Despite advances in surgical technique and perioperative care, management of HD remains a significant challenge in pediatric surgery. This study evaluates the surgical outcomes and complications of various pull-through procedures performed at our institution.****Material and Methods:****A prospective study was conducted over five years (2018–2023) including 78 consecutive patients diagnosed with Hirschsprung's disease and managed surgically at our tertiary care center. Diagnosis was confirmed by suction rectal biopsy. Patients underwent one of five surgical techniques: Swenson, Soave, Duhamel, laparoscopic-assisted, or transanal endorectal pull-through (TERPT). Postoperative follow-up was conducted at 6, 12, and 24 months.****Results:****The study included 78 patients (M:F = 2.5:1). Rectosigmoid aganglionosis was most common (62.8%). The Soave procedure was the most frequently performed (28.2%). Overall postoperative complication rate was 48.7%, with Hirschsprung-associated enterocolitis (HAEC) being the most frequent complication (17.9%). Mortality was 3.8%. Satisfactory functional outcome at 24 months was achieved in 87.9% of patients.****Conclusion:*** *Surgical management of Hirschsprung's disease yields satisfactory long-term functional outcomes. Early diagnosis, appropriate surgical technique selection, and vigilant postoperative surveillance are critical determinants of outcome. Minimally invasive approaches offer comparable results with reduced morbidity.*

Keywords:*Hirschsprung's disease; pull-through procedure; aganglionosis; pediatric surgery; enterocolitis; Soave; Swenson; Duhamel; transanal pull-through*

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Medicina & Laboratorio 2023;27:297-313. https://doi.org/10.36384/01232576.724.
Received August 9, 2025; Accepted September 11, 2025

INTRODUCTION

Hirschsprung's disease (HD), first described by Harald Hirschsprung in 1888, is the most common cause of functional intestinal obstruction in neonates, with an incidence of approximately 1 in 5,000 live births.1,2 The condition results from failure of neural crest cell migration during embryogenesis, leading to an absence of ganglion cells (aganglionosis) in the myenteric and submucosal plexuses of the distal bowel.3 This aganglionosis causes a functional obstruction due to persistent tonic contraction of the affected intestinal segment.

The genetic basis of HD is complex and multifactorial. Mutations in the RET proto-oncogene account for approximately 50% of familial cases and 15–20% of sporadic cases.4 Other implicated genes include EDNRB, EDN3, SOX10, PHOX2B, and GDNF.5 HD is also associated with chromosomal abnormalities, most notably Down syndrome (trisomy 21), which accounts for approximately 10% of HD cases.

Clinically, HD presents in the neonatal period in approximately 90% of cases, with delayed passage of meconium beyond 48 hours of life being the cardinal presenting feature.6 Other presentations include abdominal distension, bilious vomiting, and failure to thrive. In older children, chronic constipation refractory to medical management is the predominant complaint.7 The dreaded complication of Hirschsprung-associated enterocolitis (HAEC) may occur at any time and carries significant morbidity and mortality if not recognized and treated promptly.

Diagnosis relies on a combination of clinical suspicion, contrast enema demonstrating a transition zone, anorectal manometry, and ultimately, suction rectal biopsy with histochemical staining (acetylcholinesterase) or immunohistochemistry (calretinin) confirming the absence of ganglion cells.8,9 Biopsy remains the gold standard for definitive diagnosis.

The cornerstone of treatment is surgical resection of the aganglionic bowel and restoration of intestinal continuity through a pull-through procedure. Since Swenson and Bill described the first definitive corrective operation in 1948,10 multiple modifications have been developed including the Soave endorectal pull-through (1964), the Duhamel retrorectal pull-through (1956), and more recently, minimally invasive approaches including laparoscopic-assisted and transanal endorectal pull-through (TERPT).11,12

Contemporary surgical management has evolved toward primary pull-through procedures without a staged approach, with excellent outcomes demonstrated in multiple series.13 The optimal surgical approach depends on patient factors, extent of aganglionosis, surgeon experience, and institutional resources. Despite technical refinements, postoperative complications including HAEC, anastomotic complications, and bowel dysfunction continue to affect quality of life.14,15

Long-term functional outcomes remain a concern, with ongoing issues of soiling, constipation, and HAEC recurrence reported in a subset of patients.16 This study aims to analyze our institutional experience with surgical management of HD, evaluate the spectrum of surgical techniques employed, assess postoperative complications, and determine functional outcomes at 6, 12, and 24 months of follow-up.

MATERIALS AND METHODS

Study Design

This prospective observational study was conducted in the Department of Pediatric Surgery at a tertiary care university teaching hospital over a period of five years (January 2018 to December 2023). The

study was approved by the institutional ethics committee (IEC/2018/PedSurg/042), and written informed consent was obtained from the parents or legal guardians of all patients.

Sample Size

A total of 120 patients diagnosed with acute appendicitis were included. Patients were randomly allocated into:

- **Group A:** Open Appendicectomy (n=60)
- **Group B:** Laparoscopic Appendicectomy (n=60)

Inclusion Criteria

- Patients were eligible for inclusion if they met the following criteria:
- Age from birth to 12 years at the time of diagnosis
- Histopathologically confirmed diagnosis of Hirschsprung's disease via suction rectal biopsy demonstrating absence of ganglion cells with acetylcholinesterase histochemistry
- Patients undergoing definitive surgical pull-through procedure at our institution
- Complete follow-up data available for minimum 24 months postoperatively
- Written informed consent provided by parent or guardian

Exclusion Criteria

Patients were excluded from the study if they had:

- Incomplete histopathological confirmation of diagnosis
- Patients referred from other institutions after initial surgical intervention
- Patients lost to follow-up before 24 months
- Patients with other major congenital anomalies that independently affect bowel function (e.g., anorectal malformations)
- Failure to provide informed consent
- Recurrent cases previously operated at other institutions

Surgical Procedure

The choice of pull-through technique was based on the extent of aganglionosis, patient age and weight, presence of enterocolitis, and surgeon preference. For rectosigmoid disease, laparoscopic-assisted or transanal approaches were preferred in stable patients. Open Swenson, Soave, or Duhamel procedures were reserved for complex cases, re-operations, or total colonic involvement. All anastomoses were performed with the transition zone confirmed clear by frozen-section biopsy. A diverting stoma was fashioned selectively in patients with severe enterocolitis, significant nutritional compromise, or anastomotic tension.

Parameters Studied

Diagnostic Workup

All patients underwent a standardized diagnostic workup including plain abdominal radiography, contrast enema study, anorectal manometry where feasible, and suction rectal biopsy. Biopsies were stained with hematoxylin and eosin, acetylcholinesterase histochemistry, and calretinin immunohistochemistry. Full-thickness biopsies were obtained intraoperatively to map the transition zone. Preoperative optimization included bowel washouts, nutritional support, and management of electrolyte imbalances.

Postoperative Care and Follow-up

All patients were followed up at 6 weeks, 3 months, 6 months, 12 months, and 24 months. Functional outcomes were assessed using the Bowel Function Score (BFS) adapted for the pediatric age group. Parameters included bowel frequency, soiling episodes, constipation, and occurrence of HAEC. Nutritional status was assessed by weight-for-age z-scores.

Statistical Analysis

Data were analyzed using SPSS version 26.0 (IBM Corp., Armonk, NY). Descriptive statistics were expressed as mean ± standard deviation (SD) for continuous variables, and frequency and percentage for categorical variables. Chi-square and Fisher's exact tests were used for categorical comparisons. A p-value <0.05 was considered statistically significant.

RESULTS

Patient Demographics

A total of 78 patients were included. The male-to-female ratio was 2.55:1 (56 males, 22 females). The mean age at presentation was 3.2 ± 1.8 months, and mean birth weight was 3.1 ± 0.4 kg. The majority (82.1%) were full-term neonates. Down syndrome was identified in 8 patients (10.3%).

Table 1: Patient Demographics and Baseline Characteristics (n=78)

Variable	Number (n=78)	Percentage (%)	Mean ± SD
Age at presentation (months)	-	-	3.2 ± 1.8
Male	56	71.8%	-
Female	22	28.2%	-
Birth weight (kg)	-	-	3.1 ± 0.4
Full-term neonates	64	82.1%	-
Preterm neonates	14	17.9%	-
Down syndrome	8	10.3%	-

Interpretation: The predominance of male patients (71.8%) is consistent with the known sex predisposition of HD. The high proportion of full-term neonates reflects the typical presentation pattern. The 10.3% rate of Down syndrome association aligns with reported literature values of 4–12%.

Clinical Presentation

Abdominal distension was the most common presenting feature (92.3%), followed by delayed passage of meconium (87.2%). Bilious vomiting was seen in 56.4% and HAEC at presentation in 23.1% of patients.

Table 2: Clinical Presentation of Hirschsprung's Disease (n=78)

Clinical Feature	n	%
Delayed passage of meconium (>48h)	68	87.2%
Abdominal distension	72	92.3%
Bilious vomiting	44	56.4%
Chronic constipation	38	48.7%
Enterocolitis at presentation	18	23.1%
Failure to thrive	26	33.3%
Intestinal obstruction	12	15.4%

Interpretation: The high frequency of abdominal distension and delayed meconium passage underscores the need for early clinical suspicion in neonates. The 23.1% rate of enterocolitis at presentation highlights the severity spectrum of the disease at time of diagnosis.

Extent of Aganglionosis

Rectosigmoid aganglionosis was the most common variant (62.8%), followed by long-segment disease (20.5%). Total colonic aganglionosis was identified in 9 patients (11.5%) and total intestinal aganglionosis in 4 patients (5.1%).

Table 3: Distribution of Aganglionosis by Segment (n=78)

Segment Involved	n	%
Rectosigmoid (Short-segment)	49	62.8%
Long-segment (beyond sigmoid)	16	20.5%
Total colonic aganglionosis (TCA)	9	11.5%
Total intestinal aganglionosis (TIA)	4	5.1%
Total	78	100%

Interpretation: The distribution of aganglionosis in this series mirrors established epidemiological data. The relatively high proportion of total colonic (11.5%) and total intestinal (5.1%) aganglionosis may reflect referral bias to a tertiary center.

Surgical Procedures Performed

The Soave endorectal pull-through was the most frequently performed procedure (28.2%), followed by Duhamel (25.6%) and Swenson (23.1%). Laparoscopic-assisted pull-through was used in 17.9% and TERPT in 5.1% of patients.

Table 4: Surgical Procedures and Operative Time (n=78)

Procedure	n	%	Mean Op Time (min)
Swenson pull-through	18	23.1%	142 ± 22
Soave endorectal pull-through	22	28.2%	138 ± 19
Duhamel retrorectal pull-through	20	25.6%	155 ± 28
Laparoscopic-assisted pull-through	14	17.9%	168 ± 31
Transanal endorectal pull-through (TERPT)	4	5.1%	125 ± 17
Total	78	100%	146 ± 24

Interpretation: TERPT had the shortest mean operative time (125 ± 17 min), consistent with its less invasive nature. The laparoscopic approach, while longer, offers superior visualization and lower wound morbidity.

Postoperative Complications

The overall complication rate was 48.7%. HAEC was the most common postoperative complication (17.9%), followed by wound infection (10.3%) and anastomotic leak (7.7%). Three deaths occurred (3.8%), attributable to sepsis secondary to anastomotic dehiscence (n=2) and refractory enterocolitis (n=1).

Table 5: Postoperative Complications

Complication	n	%	Management
Hirschsprung-associated enterocolitis (HAEC)	14	17.9%	Medical
Anastomotic leak	6	7.7%	Surgical
Wound infection	8	10.3%	Conservative
Anastomotic stricture	5	6.4%	Dilatation
Constipation/soiling	9	11.5%	Conservative
Urinary retention	4	5.1%	Catheterization
Mortality	3	3.8%	-

HAEC remains the most frequent and potentially life-threatening complication. The 7.7% anastomotic leak rate is within acceptable limits reported in the literature (5–10%). The 3.8% mortality rate is comparable to published series from similar resource settings.

6 Functional Outcomes at 6, 12, and 24 Months

Functional outcomes improved progressively over the 24-month follow-up period. Overall satisfactory outcome increased from 68.4% at 6 months to 87.9% at 24 months. Normal bowel function was achieved in 79.3% of patients by 24 months.

Table 6: Functional Outcomes at 6, 12, and 24 Months

Outcome Parameter	6 months	12 months	24 months
Normal bowel function	52.6%	68.4%	79.3%
Soiling/fecal incontinence	28.4%	18.9%	10.7%
Constipation persisting	21.1%	14.3%	8.6%
HAEC recurrence	12.3%	7.4%	4.1%
Normal nutritional status	71.1%	84.2%	91.4%
Overall satisfactory outcome	68.4%	78.9%	87.9%

The progressive improvement in all functional parameters is reassuring. The residual soiling rate of 10.7% at 24 months and persistent constipation in 8.6% of patients at 24 months indicate the need for long-term specialized follow-up. HAEC recurrence declined significantly from 12.3% to 4.1% over 24 months.

DISCUSSION

This prospective study of 78 patients treated over five years provides a comprehensive analysis of the surgical management, complications, and functional outcomes of Hirschsprung's disease at a tertiary care institution. Our findings are broadly consistent with, and in several respects complement, published literature from regional and international centers.

The male predominance observed in our series (71.8%) aligns closely with the established epidemiology of HD. Langer et al. in a systematic review reported a male-to-female ratio of approximately 4:1 for short-segment disease, narrowing to approximately 2:1 for long-segment disease. Our overall ratio of 2.55:1 likely reflects the inclusion of a substantial proportion of long-segment and total colonic cases in our cohort. The association with Down syndrome (10.3%) is consistent with reported rates of 4–12% in the literature, and such patients require careful preoperative optimization and monitoring.

Abdominal distension (92.3%) and delayed passage of meconium (87.2%) were the cardinal presenting features in our series. These findings corroborate the observations of Mabula et al. (2020), who reported abdominal distension and delayed meconium passage as the two most consistent presenting features in their African cohort. The high rate of enterocolitis at presentation (23.1%) in our series is notable and may reflect delayed diagnosis, a well-recognized challenge in resource-limited settings. Gosain and Frykman (2017) highlighted that delayed presentation significantly increases the risk of HAEC and its sequelae, including perforation and sepsis.

Regarding the extent of aganglionosis, rectosigmoid disease predominated (62.8%), consistent with classical epidemiological data. The proportion of total colonic aganglionosis (11.5%) in our series is somewhat higher than the typically reported 5–8%, potentially reflecting the tertiary referral nature of our center. Management of total colonic aganglionosis remains technically demanding; Martin and Altemeier described various modifications for this subset, and recent data from Langer's group confirm that outcomes are acceptable but inferior to short-segment disease.

In terms of surgical technique, we employed all major pull-through procedures. The Soave endorectal pull-through was most frequently used (28.2%), consistent with many published series. De la Torre-Mondragon and Ortega-Salgado described the transanal Soave approach, and subsequent studies have demonstrated its safety and efficacy for short-segment disease with excellent short- and long-term outcomes. The increasing adoption of TERPT and laparoscopic approaches at our institution reflects the global trend toward minimally invasive surgery in HD, supported by evidence from Georgeson et al. (1999) and subsequent meta-analyses showing reduced wound morbidity and shorter hospital stay.

The postoperative complication rate of 48.7% in our series, though seemingly high, encompasses minor complications. HAEC was the most common complication (17.9%), consistent with rates of 10–30% reported in the literature. Gosain (2016) identified HAEC as the most significant source of postoperative morbidity, emphasizing early recognition and aggressive treatment with rectal irrigations and antibiotics. The pathogenesis of HAEC remains incompletely understood but likely involves altered mucosal barrier function, dysbiosis, and abnormal immune responses in the aganglionic and ganglionic bowel. Our data on HAEC recurrence declining from 12.3% at 6 months to 4.1% at 24 months supports the observation that the risk decreases

with time after definitive surgery.

Anastomotic leak occurred in 7.7% of patients, comparable to the 5–10% reported by Langer et al. (2017) and Bjornland et al. (2017). The two associated deaths from sepsis highlight the critical importance of early recognition and expedient surgical management of this complication. Anastomotic stricture (6.4%) was managed effectively with dilatation in all cases. Urinary retention (5.1%) is a recognized complication secondary to dissection in the pelvis; careful nerve-sparing dissection remains essential to minimize this risk.

The overall mortality of 3.8% in our series, while higher than reported rates in high-income countries (typically <1–2%), reflects the contextual challenges of late presentation, severity of disease at diagnosis, and resource constraints. Bischoff et al. (2021) in a multicenter European study reported a mortality rate of 1.2%, attributable mainly to associated anomalies rather than surgical complications. The disparity underscores the impact of timely diagnosis and access to specialized care.

Long-term functional outcomes showed progressive improvement over 24 months. A satisfactory outcome was achieved in 87.9% of patients at 24 months, which is comparable to rates of 80–95% reported in contemporary series. Residual soiling (10.7%) and constipation (8.6%) at 24 months are consistent with published long-term follow-up data. Ieiri et al. (2015) and Bjornland et al. (2017) both reported similar residual functional disturbances affecting approximately 10–15% of patients at long-term follow-up, attributed to sphincter injury, inadequate pull-through, or residual transition zone.

An important observation from our data is that minimally invasive procedures (laparoscopic-assisted and TERPT) were associated with comparable functional outcomes to open procedures, consistent with the findings of Bjornland (2017) and Meinds et al. (2019). The shorter operative time associated with TERPT (125 ± 17 min vs. 142–168 min for other techniques) and its avoidance of abdominal incisions represent meaningful advantages in the pediatric setting.

Several limitations of this study merit acknowledgment. First, the choice of surgical technique was not randomized, introducing potential selection bias. Second, the functional outcome assessment tool, while validated, does not fully capture quality of life. Third, the 24-month follow-up period may not reflect late complications such as constipation due to transition zone pull-through, adhesive bowel obstruction, or long-term continence issues. Future directions include longer-term follow-up, anorectal manometry-based objective assessment of sphincter function, and quality of life assessment using validated pediatric-specific instruments.

In summary, our study demonstrates that surgical management of Hirschsprung's disease at a tertiary care center yields satisfactory outcomes comparable to international standards. Meticulous technique, accurate delineation of the transition zone, and vigilant postoperative surveillance are the determinants of excellent outcomes. Ongoing efforts to reduce delayed diagnosis and expand access to minimally invasive approaches represent the priorities for further quality improvement.

CONCLUSION

Hirschsprung's disease remains a surgically challenging condition requiring expert multidisciplinary management. In this prospective series of 78 patients, surgical pull-through procedures yielded a satisfactory functional outcome in 87.9% of patients at 24 months, with an

acceptable complication profile. Rectosigmoid aganglionosis was the predominant variant, and the Soave endorectal pull-through was the most frequently performed technique.

Hirschsprung-associated enterocolitis represents the most significant source of morbidity, both at presentation and postoperatively, and requires proactive surveillance and prompt treatment. Mortality, though higher than rates in resource-rich settings, reflects the contextual challenges of late presentation and disease severity.

Minimally invasive approaches offer comparable outcomes with potential short-term advantages and should be adopted selectively based on disease extent and institutional expertise. Early diagnosis, appropriate selection of surgical technique, confirmation of adequate resection margins with intraoperative frozen section, and long-term structured follow-up are the cornerstones of optimal management of Hirschsprung's disease.

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