A Retrospective Study of the Management of Hirschsprung's Disease at a Tertiary Healthcare Facility in Africa

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Abstract

Background: In low- and middle-income countries (LMICs), delayed diagnosis of Hirschsprung's disease (HD) can lead to severe complications, including malnutrition and chronic constipation. This study examines patient experience and surgical outcomes for HD diagnosed beyond the neonatal period in an LMIC. **Methods:** A retrospective review of HD patients at a tertiary institution in sub-Saharan Africa was conducted to evaluate clinical characteristics, surgical interventions, and outcomes in patients with HD, including Wilcoxon rank-sum tests for non-parametric comparisons and univariate logistic regression to assess predictors of stoma placement. **Results:** Among the 61 patients (72.1% male), the average diagnosis age was 4.4 years. Short-segment HD was most common (68.9%), with the most frequent symptoms being constipation (70.5%) and abdominal distension (45.9%). Before referral, 48.6% had stomas, with older patients more likely to have them (p = 0.002). Pull-through surgery complications included strictures (17%) and requiring anal dilation (18.9%, N=53). Unfortunately, seven patients died, three before the pull-through surgery due to HD complications. **Conclusion:** Delayed pull through surgery was a common occurrence for patients with HD presenting after the neonatal period. Early detection with referral to surgeons with adequate resources is essential.

Keywords: Hirschsprung Disease, Pediatric Surgery, Post-Neonatal, Complications, Guidelines

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Introduction

Hirschsprung's disease (HD) is a congenital abnormality in which neural crest cells fail to migrate to the full length of the colon during embryonic development, leading to functional, chronic constipation and obstruction (1, 2). Although curable through surgical intervention, outcomes depend heavily on the timing of diagnosis, perioperative management, and access to specialized pediatric care (3-8). In high-income settings,

early recognition in the neonatal period, often allows for timely procedures, minimizing morbidity (7).

Children in LMICs often have a delayed diagnosis due to a combination of factors including limited awareness among caregivers and providers, reliance on traditional remedies, and constrained access to diagnostic and surgical services (3-6). Diagnostic uncertainty is compounded by variability in pathologic capacity and inconsistent definitions of disease severity, both of which affect surgical decision-making and outcomes (9). A later diagnosis increases the need for a diverting stoma, requires more unplanned surgeries (e.g., anastomotic leaks, perforations, obstruction), is often complicated by malnutrition, and has a higher incidence of major complications, including redo pull-through procedures and death (8-12). The longer length of the aganglionic segment also increases complications and impacts the quality of life (2).

Reporting of LMIC experience with HD is limited and differs greatly from the experience described in high-income countries (HICs), with current guidelines not focused on addressing the older patient population that is often seen in LMIC settings (9). By characterizing the age at diagnosis, operative approaches, and perioperative complications, we aim to identify context-specific challenges and opportunities to improve care for children with HD in resource-limited environments.

Materials and Methods

Study population and institution details

Our 350-bed tertiary, multispecialty, teaching and referral facility is located in a small town in Kenya. Data were collected between January 2020 and June 2023. Patients with a histological confirmation of aganglionosis on a full-thickness rectal biopsy or leveling biopsy were included in the study (13). Patients were excluded if they were not confirmed to be aganglionic or required redo pull-through operations as they represent a separate higher-complexity population.

Cohort characteristics and classification

Studies from HICs have considered diagnosis past 30 days as late; however, studies in LMICs have used definitions of more than 1 or 2 years to fit their patient populations (3, 8, 14-17). In order to use the classification of post-neonatal patients, to allow for greater generalizability, and to grant opportunity for future comparison with HICs, we used the HIC definition of older than 30 days for "late diagnosis." Therefore, our study population included all but one patient who presented during the data collection period (a 16-day-old infant) whom we excluded to have a complete set of post-neonatal patients.

The presence of a functioning stoma can be used to confirm the presence of ganglion cells at or above the location of the stoma. Diverting or leveling stomas were placed as per the treating surgeon's preference based on nutritional or clinical factors. Stomas were placed in children with nutrition below 2 standard deviations below normal per Z-score on mid-upper-arm circumference measurement if nutritional status could not be improved with bowel management and nutritional augmentation. Stomas were also placed for those with bowel perforation, or in the case of severe abdominal distention that could not be resolved with a bowel management regimen including rectal washouts.

Long-segment HD was defined as a lack of ganglion cells in the colon proximal to the sigmoid colon. Short-segment HD was defined as aganglionosis limited to the rectum and sigmoid colon (13). Total colonic HD was defined as a level proximal to and including the ileum; however, the sample size was small, which would not provide sufficient power in separate analysis.

Data collection and analysis

Information from both paper and electronic medical records was entered into a Research Electronic Data Capture (REDCap) database, and data were managed using REDCap tools (19, 20). Variables analyzed retrospectively through chart review included: patient presentation, number and type of surgeries, date of surgery, weight, Z-score (nutritional status), complications, administration of antibiotics, etc. Single-stage or primary pull-throughs are definitive operations without a stoma placed before or during the pull-through procedure (18). The type of pull-through varied by approach—transanal or abdominoperineal—and by technique—Soave or Swenson. A normal Z-score for

nutrition is considered 0 to −1 (21). Long-term outcomes were not included in this study.

To compare age at definitive diagnosis between patients with and without complications, we conducted Levene's test to assess the homogeneity of variances, followed by a Wilcoxon rank-sum test for non-parametric comparison. In addition, separate logistic regression analyses were conducted to examine whether rectal washouts and aganglionic segment length were each individually associated with the likelihood of diverting stoma placement. Patients with complete data on washout status, stoma placement, and segment length were included in these models. The first model assessed the effect of rectal washouts on the odds of stoma placement, while the second model evaluated the association between the segment length and stoma placement. These univariate analyses allowed for the evaluation of the main effects of each factor on the likelihood of stoma placement.

Ethical review

Local ethical review acceptance was obtained from Kijabe Hospital (approval number: KH/ISERC/02718/0031/2022).

Results

With all of the patients in the postnatal period, the majority of the study population were older than 1 year (83.6%), with a median age of 2 [interquartile range (IQR): 1–6] years. Of the 61 total patients who met the criteria, most patients were male (72.1%) and had short-segment HD (68.9%).

Table 1. Patient characteristics (N=61)

Characteristic	**N**	**%**
Age		
Less than 1 year old	10	16.4
Between 1 year and 2 years old	12	19.7
2 years old and older	39	63.9
Sex		
Male	44	72.1
Female	17	27.9
Aganglionic segment		
Long segment	17	27.9
Short segment	42	68.9
Total colon	2	3.3
Presenting symptoms		
Constipation	43	70.5
Abdominal distension	28	45.9
Vomiting	8	13.1
Other	18	29.5

Thirty-nine patients (63.9%) were diagnosed at 2 years or older. The most common presenting symptom was constipation (70.5%), followed by abdominal distension (45.9%), as shown in Table 1. "Other" presentations included anorexia, epigastric pain, poorly functioning stoma, referral for pull-through, anastomotic stenosis, desire for a second opinion, foul-smelling anal discharge, Hirschsprung-associated enterocolitis (HAEC) diagnosed at another facility, transferred due to overload of patients, discomfort during bowel movements, and umbilical hernia. Notable comorbid diagnoses included trisomy 21 (N=3) and Waardenburg syndrome (N=1).

Twenty-one patients presented having undergone prior procedures at other hospitals; some patients had more than one procedure, amounting to a total of 37 procedures. Stomas were placed in 18 patients (48.6%), 12 (32.4%) patients had rectal biopsies, and 7 (18.9%)

had leveling biopsies. Eight patients had complications from their previous procedures, including stoma stenosis or persistent constipation.

Of the 61 patients, 53 (86.9%) had leveling biopsy procedures at our institution. Five patients needed repeated leveling biopsies (8.2%) because of inadequate results or imprecise transition zone location. Frozen section pathology is not readily available; 18 patients (29.5%) had a contrast enema to aid in diagnosis. Eight patients either had a leveling biopsy result from another hospital or a functioning stoma proximal to the presumed transition zone.

Table 2. Pull-through procedure variables versus presence of a complication (N=53)

Variable	Complication (%)	No complication (%)	Total (%)
Approach			
Transanal	10 (38.6)	16 (61.5)	26 (49.1)
Abdominoperineal	10 (41.7)	14 (58.3)	24 (45.3)
Laparoscopic assisted	2 (66.7)	1 (33.3)	3 (5.7)
Technique			
Soave	7 (35.0)	13 (65.0)	20 (37.7)
Swenson	11 (44.0)	14 (56.0)	25 (47.2)
Soave/Swenson	2 (33.3)	4 (66.6)	6 (11.3)
Unknown	2 (100)	0	2 (3.8)
Z-score			
−3	1 (100)	0 (0.0)	1 (1.9)
−2	6 (50.0)	6 (50.0)	12 (22.6)
−1	9 (52.9)	8 (47.1)	17 (32.1)
0	1 (14.3)	6 (85.7)	7 (13.2)
1	4 (36.4)	7 (63.6)	11 (20.8)
Unknown	1 (20.0)	4 (80.0)	5 (9.4)

Table 3. Timing of stoma placement (N=61)

Stoma placement	Short segment	Long segment	Total colon	Total (%)
Before coming to hospital	13	5	0	18 (29.5)
Before pull-through	1	3	2	7 (11.5)
During pull-through	1	1	0	2 (3.3)
After pull-through	4	1	0	5 (8.2)
No stoma	22	7	0	29 (47.5)

Patients received diverting stomas at different times in their course of treatment. Most stomas were placed prior to arrival (66.7%), with records indicating treatment for obstruction, perforation, or feeding intolerance. Fifty-nine percent of patients had rectal washouts. Twenty-one patients (34.4%) had signs of HAEC during their care.

A total of 53 pull-through surgeries were performed, with 32 (60.4%) being primary pull-through surgeries (no diverting stoma). The numbers and percentages can be found in Table 2, with notes of post-operative complications. The percentages for the complication versus no complication columns were determined by specific approach, technique, or Z-score (calculated horizontally), while percentages for the total column were determined out of all patients who received a pull-through (N=53).

Length of aganglionic segment analysis

The median age for the 17 patients with long-segment HD was 2 years (IQR: 1–3 years), while the median age for the 42 patients with short-segment HD was 3 years

(IQR: 1–6.75 years). A Wilcoxon rank-sum test was performed to compare the age distributions between the two groups, which indicated no significant difference (p = 0.1852). Table 3 describes which patients received stomas at which point during their treatment course by the length of the aganglionic segment.

Table 4. Univariate logistic regression analysis results for outcomes of stoma placement

Outcome	Log(OR)	95% CI	*p* value
Rectal washouts	0.10	0.02–0.40	0.002
Long segment	1.25	0.20–10.6	0.8
Washout[a] long segment	1.71	0.11–21.7	0.7

CI, confidence interval; OR, odds ratio.

Table 5. The impact of age at diagnosis on outcomes in Hirschsprung's disease: Wilcoxon rank-sum comparative analysis

	N [median age (IQR), years]		
	Present	**Absent**	***p* value**
Diverting stoma[a]	24 [5 (2–9.75)]	35 [2 (0.5–4)]	0.002
Rectal washout[a]	34 [2 (1–4)]	25 [4 (1–9)]	0.075
HAEC[a]	19 [3 (1.5–5)]	40 [2 (1–6.25)]	0.677
Stoma after pull-through	5 [1 (1–2)]	54 [3 (1–6.75)]	0.217
Deceased	7 [9 (1.5–12.5)]	52 [2 (1–5.25)]	0.167
Anastomotic dehiscence	4 [1.5 (1–3)]	48 [2.5 (1–5.25)]	0.532
Stricture (non-surgical)	9 [2 (2–2)]	43 [3 (1–6)]	0.608
Unintended further procedures	5 [1 (1–2)]	47 [3 (1–5.5)]	0.308
Abscess	4 [4 (1.75–7.5)]	48 [2 (1–5)]	0.532
Surgical anal dilation	10 [3.5 (2–5)]	42 [2 (1–5.75)]	0.174
Any complication	21 [2 (1–5)]	31 [3 (1–6)]	0.763

HAEC, Hirschsprung-associated enterocolitis; IQR, interquartile range.
[a]Unrelated to operation.

Of the short-segment HD patients with stomas placed during their care (N=19), 68.4% received stomas before presentation at our facility. In contrast, only 29.4% of patients with long-segment HD (N=17) received stomas before presentation at our facility. Both patients with total colonic HD had diverting stomas before undergoing a definitive pull-through procedure. Table 4 describes the univariate logistic regression analyses implemented to assess the association of rectal washouts and aganglionic segment length with the likelihood of diverting stoma placement. Patients who received rectal washouts had significantly lower odds of requiring a diverting stoma compared to those who did not receive washouts [odds ratio (OR): −2.11, 95% confidence interval (CI): −3.4 to −0.97, p = 0.0005]. Aganglionic segment length was not significantly associated with stoma placement; however, patients with long segments had slightly higher odds of having a stoma placed (OR=0.37, 95% CI: −0.78 to 1.5, p = 0.50).

Age analysis

The distribution of ages at the time of definitive diagnosis for various clinical factors, categorized by the "present" and "absent" status, was analyzed using the Wilcoxon rank-sum test (Table 5). Importantly, this analysis does not classify based on pre- or post-procedure characteristics. For diverting stoma, the "present" group had a median age of 5 years (IQR: 2–9.75 years), while the "absent" group had a median age of 2 years (IQR: 0.5–4 years), with a statistically significant difference observed between the two groups (p = 0.002). In contrast, rectal washout showed no significant difference, with the "present" group having a median age of 2 years (IQR: 1–4 years) and the "absent"

group a median age of 4 years (IQR: 1–9 years) (p = 0.075). The age distributions for HAEC, deceased, anastomotic dehiscence, stricture (non-surgical), unintended further procedures, abscess, surgical anal dilation, and any complication did not differ significantly between the groups. Specifically, the median ages for these factors were similar, with p values ranging from 0.174 to 0.763. Therefore, the only factor that showed a significant difference in age distribution was diverting stoma, while the remaining clinical factors did not demonstrate statistically significant differences in age.

Table 6. Logistic Regression Results: Effects of Age at Diagnosis on Clinical Outcomes

Outcome	**95% CI**	***p* value**
Segment length	−0.25 to 0.03	0.2
Complication, any	−0.22 to 0.05	0.3
Diverting stoma	0.04 to 0.30	0.016
Deceased	−0.02 to 0.23	0.090

CI, confidence interval; OR, odds ratio.

Table 6 presents the univariate logistic regression analyses assessing the relationship between age at diagnosis and clinical outcomes. Age at diagnosis was not significantly associated with segment length [log(OR)=−0.09, 95% CI: −0.25 to 0.03, p = 0.2] or the occurrence of complications [log(OR)=−0.07, 95% CI: −0.22 to 0.05, p = 0.3]. However, older age at diagnosis was significantly associated with a higher likelihood of requiring a diverting stoma [log(OR)=0.15, 95% CI: 0.04–0.30, p = 0.016]. Although a positive association was observed between age and mortality [log(OR)=0.10, 95% CI: −0.02 to 0.23, p = 0.090], it did not reach statistical significance.

Complication analysis

Overall, 22 patients (36.1%) had documented post-operative complications. Those of interest included anastomotic dehiscence (7.5%), stricture (non-surgical, 17.0%), unintended further procedures within 30 days of pull-through (9.4%), abscess (7.5%), and need for surgical anal dilation(18.9%). There was no significant difference in complications for type of approach, technique, or Z-score. Patients with Z-scores of 0 or 1 had higher chances of having no complications (85.7% and 63.6%, respectively) compared to those with lower Z-scores (Table 2). Patients with Z-scores of −2 or −3 were most likely to have a stricture (N=4) or require surgical anal dilation (N=3).

Five patients required stomas after the pull-through procedure. The reasons for stoma creation after pull-through included: abscess and anastomotic leak (N=2), small bowel obstruction and internal hernia related to the procedure (N=1), HAEC shortly after surgery (N=1), and persistent constipation after a year (N=1). Significantly, according to our records through June 2023, none of the 32 patients who received a primary pull-through required a repeat pull-through procedure.

In total, seven patients (11.5%) died. Four deaths were related to surgery, including a 10-year-old patient who had a hypoxic event in the post-anesthesia care unit leading to seizures and diffuse cerebral edema, a 2-year-old patient with cardiac arrest due to disseminated intravascular coagulation (DIC), a 1-year-old patient with post-operative aspiration, and another 1-year-old patient with sepsis, DIC, and multiorgan failure. The other three deaths occurred before pull-through and were determined to be caused by HD or other related diseases, including severe acute malnutrition (an 18-year-old patient), intestinal failure (a 15-year-old patient), and sepsis/HAEC (a 9-year-old patient).

Discussion

Our study highlights the significant challenges associated with the late diagnosis and management of HD in an LMIC setting. The majority of patients in our cohort were diagnosed well beyond the neonatal period, with a median age of 2 years, and more than 80% being

diagnosed after their first year of life. This delay in diagnosis was associated with an increased presence of diverting stomas and a higher likelihood of complications, including anastomotic leaks and requiring surgery for anal dilation. Our evidence, while not statistically significant, shows that older patients in our population more frequently have short-segment HD. Likely, these patients had less severe symptoms and controlled their constipation using enemas and laxatives (17, 22). Notably, older age at diagnosis was significantly correlated with stoma placement. Many of these patients likely could have received primary pull-throughs if diagnosed and referred to appropriately trained pediatric surgeons at a younger age. Patients with short-segment HD have shown success with primary pull-throughs even without a pathological confirmation (23). Therefore, later presentations for indicated definitive procedures affect patients' quality of life.

Decision-making regarding leveling biopsies in our setting relies on a combination of pathology reports and radiographic evidence (20). Contrast enemas are not routinely performed due to cost, intraoperative evaluation of bowel dilation, and stoma functionality. The need for repeat leveling biopsies in 8.2% of patients likely reflects concerns about the certainty and accuracy of pathology findings when pathology serves as the primary determinant of surgical planning (5, 6, 20). These challenges highlight the necessity of strengthening pathology services and refining diagnostic protocols in resource-limited settings.

Additionally, rectal washouts play a crucial role in our treatment approach, helping to manage constipation, avoid HAEC, and reduce bowel dilation to facilitate anastomosis (16). Our findings suggest that children receiving rectal washouts were generally younger and more likely to proceed directly to pull-through without fecal diversion, provided they had caregivers capable of administering consistent washouts. This reinforces rectal washouts as a preferred initial management strategy when diagnosed early, preventing disease progression and reducing the need for stoma formation.

Despite these challenges, our findings suggest that definitive surgical intervention remains feasible, even in delayed cases, with 60.4% of patients undergoing primary pull-through procedures without prior diversion (23). Encouragingly, none of these patients required a repeat pull-through procedure within the study period. However, the mortality rate of 11.5%, with both surgical and disease-related causes, underscores the critical need for earlier recognition and intervention. Three of the patients with disease-related causes of death were older than 9 years of age, likely indicating a severe burden of disease for an extensive period of time.

Limitations and future studies

This study had limitations, including a small sample size and reliance on patient or caretaker memory due to missing surgical and pathology records. The definition and reporting of HAEC were inconsistent, potentially leading to inaccurate data. Nutritional evaluations were only systematically conducted starting in 2021, making earlier Z-score data less reliable. The study did not track long-term complications or follow-up with patients who may have sought care at outside hospitals. Future studies should focus on long-term functional outcomes and quality-of-life assessments to guide further improvements in HD management.

Conclusion

This study contributes to the growing body of evidence on HD in LMICs and underscores the urgent need for strategies aimed at earlier diagnosis and optimized perioperative care. Strengthening provider education, improving access to diagnostic tools, encouraging appropriate referrals for definitive procedures, and incorporating standardized treatment guidelines tailored to the LMIC setting may help reduce morbidity and mortality.

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Author contributions

KRS led in formal analysis, investigation and in writing of the original draft. KRS and JA led in the reviewing & editing of the original draft. JA led in conceptualization.

All authors equally contributed to data curation, methodology, project administration, resources, supervision and validation.

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