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A Nine-Year Review of Clinical Presentations, Surgical Management and Outcomes of Hirschsprung's Disease in a Resource-Limited Setting

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Abstract

Background: Hirschsprung's disease is a common cause of distal intestinal obstruction in children. The patterns of clinical presentation and outcomes vary widely across different settings, influenced by available resources.

Objective: To review the clinical presentation, surgical management, and outcomes of children with Hirschsprung's disease.

Methods: This was a descriptive, retrospective, case series of children aged 0 to 15 years with Hirschsprung's disease who were treated between January 2015 and December 2023. Data on clinical presentation, surgical treatment, post-operative complications, and mortality were obtained from hospital records for analysis.

Results: The data of 23 children were retrieved for analysis. The male-to-female ratio was 6.7:1, and the median age at presentation was 11 months. Only about one-third presented in the neonatal period. Abdominal swelling (87%) and chronic constipation (70%) were the predominant symptoms, while the short-segment rectosigmoid disease type was most common. The transabdominal Soave–Boley pull-through was performed in 19 children, mainly as a two-stage procedure following colostomy. The most common complications were peristomal dermatitis (39%) and wound infection (30%), while mortality occurred in 13% of the patients. There was no association between age at presentation and stoma-related complications, pull-through–related complications, need for re-operation or mortality.

Conclusion: The clinical presentation of Hirschsprung disease was diverse, though most cases presented after the neonatal period, reflecting barriers to early recognition and referral. Post-operative outcomes were acceptable and comparable to those reported in similar resource-constrained settings.

Keywords: Constipation, Delayed Diagnosis, Hirschsprung Disease, Intestinal Obstruction, Post-operative complications.

Introduction

Hirschsprung's disease (HD) is a congenital disorder of the enteric nervous system characterised by the absence of ganglion cells in the submucosal and myenteric plexuses of the distal bowel, which are responsible for normal intestinal peristalsis. This absence results in functional intestinal obstruction at the level of the aganglionic segment due to impaired motility of the affected bowel.[1] It is a common cause of

intestinal obstruction in children, with an estimated incidence of 1 in 5,000 live births, and it shows a marked male predominance. [2] The disease mostly affects the distal colon but may extend proximally for varying lengths. The rectosigmoid segment is affected in approximately 80% of cases, while long-segment and total colonic aganglionosis occur less frequently. [3]

The clinical manifestations vary according to age, the length of the aganglionic segment, and the presence of associated comorbidities. [4] The clinical presentation ranges from neonatal intestinal obstruction to chronic progressive constipation in older children. Early diagnosis and timely surgical management are essential to prevent complications such as Hirschsprung-associated enterocolitis, intestinal perforation, and malnutrition. [5] In high-income countries, advances in neonatal care, diagnostic imaging, and pathology services have improved early recognition, reduced disease morbidity and led to favourable treatment outcomes.[6] In contrast, delayed presentation remains common in low- and middle-income countries. In Africa, only 20–40% of cases are diagnosed in the neonatal period compared with over 90% in developed countries.[7]

The diagnosis requires a high index of suspicion, supported by radiographic studies and histological confirmation from rectal or colonic biopsies. In many low-resource settings, limited awareness of the disease, scarcity of diagnostic tools, and socioeconomic constraints contribute to delayed referral for surgical care.[8] In addition, many healthcare providers have limited familiarity with its clinical presentation, while poverty and low health literacy among parents further delay care. Studies from sub-Saharan Africa have reported variable outcomes following surgery for Hirschsprung's disease, reflecting differences in patient age at presentation, disease extent, and resource availability. [7 - 10] However, published data from Nigeria remain sparse, whereas local experience is essential for understanding the burden and outcomes of HD within resource-limited health systems. This study aimed to describe the clinical characteristics and management outcomes of Hirschsprung's disease in a tertiary hospital in southern Nigeria.

Methods

Study design and setting

This was a descriptive, retrospective, case series conducted in a paediatric surgical unit of the University of Uyo Teaching Hospital, a tertiary hospital in Akwa Ibom, southern Nigeria. The hospital is a 500-bed referral centre that serves the state and neighbouring regions. The study covered 9 years, from January 2015 to December 2023.

Ethical considerations

Ethical approval for the study was obtained from the Health Research Ethics Committee of the University of Uyo Teaching Hospital (UUTH/AD/S/96/VOL.XXII/10). All data were de-identified at entry and managed in accordance with institutional confidentiality policies and the principles of the Declaration of Helsinki.

Study population and eligibility criteria

This case series included children aged 0 to 15 years diagnosed with Hirschsprung's disease who received treatment during the study period. Cases with incomplete records or those who did not complete treatment at the facility were excluded. Given the retrospective nature of the study, all eligible patients were included consecutively.

Data collection and variables

Clinical records were reviewed using a structured proforma. The collected data included demographic information [age at presentation, grouped into neonates (<1 month), infants (1–12 months), and children (>1 year)], and sex.

Clinical presentation was defined by symptoms or disease complications observed at first contact. Diagnostic work-up was recorded, including histopathological confirmation (rectal or colonic biopsy) and contrast studies (barium enema). The level of aganglionosis was classified as short-segment (rectosigmoid), long-segment, or ultrashort-segment disease based on operative and histological findings.

Surgical management was categorised as (a) two-stage procedures (a preliminary levelling colostomy followed by definitive pull-through), (b) one-stage procedures (either definitive pull-through without a diverting stoma or posterior myectomy for ultrashort disease), or (c) other procedures (diverting ileostomy).

Post-operative outcomes were assessed in terms of early complications (stoma-related and pull-through–related), need for re-operation, and mortality. Mortality was defined as death during the index admission or within 30 days of definitive surgery.

Statistical analysis

The data were analysed using IBM SPSS Statistics for Windows, version 24 (IBM Corp., Armonk, NY, USA). Continuous variables were summarised as medians and interquartile ranges (IQRs) as they were not normally distributed. Categorical variables were described using frequency and percentages. A sub-analysis was performed to explore the association between age at presentation and post-operative outcomes. Fisher's exact test was used because of small event counts. A p-value <0.05 was considered statistically significant. Results are presented in tables and charts.

Results

Patient demographics

A total of 23 children with histologically confirmed Hirschsprung's disease who had complete medical records were included in this review. There was a male predominance, with 20 males (87.0%) and three females (13.0%), giving a male-to-female ratio of 6.7:1. The age at presentation ranged from seven days to 14 years, with a median of 11 months (IQR: 2 weeks–18 months). Only 7(30.4%) cases presented in the neonatal period (Table I).

Table I: Age distribution of children at presentation

Age	Frequency (%)
< 1 month	7 (30.4)
>1 – 12 months	8 (34.8)
> 1 year	8 (34.8)
Total	23 (100)

Clinical presentation and diagnosis

The patients presented with clinical features and different complications of Hirschsprung's disease. Abdominal swelling was the most frequent symptom, occurring in 20(87.0%), followed by chronic constipation in 16 (69.6%). Enterocolitis was recorded in 5(21.7%) children. Among neonates, the most common features included abdominal swelling and delayed passage of meconium, while in older children, chronic constipation was the predominant symptom (Table II).

All the children had histological confirmation of Hirschsprung's disease. Sixteen children (69.6%) had colonic biopsies at the time of stoma creation, while seven (30.4%) had rectal biopsies. Barium enema was obtained in 9(39.1) children, showing features consistent with Hirschsprung's disease. The levels of aganglionosis identified at surgery

are described in Figure 1. Short-segment (rectosigmoid) disease was the most common type (19; 82.6%) (Figure 2).

Table II: Clinical presentations in children with Hirschsprung's disease distributed by age at presentation

Symptom/Complication	<1 month	>1-12 months	>1 year	Total (%)
Abdominal swelling	5	7	8	20 (87.0)
Chronic constipation	0	8	8	16 (69.6)
Delayed passage of meconium	5	5	4	14 (60.9)
Growth retardation	0	2	5	7 (30.4)
Acute intestinal obstruction	2	3	1	6 (26.1)
Enterocolitis	2	1	2	5 (21.7)
Intestinal Perforation/Peritonitis	2	0	0	2 (8.7)

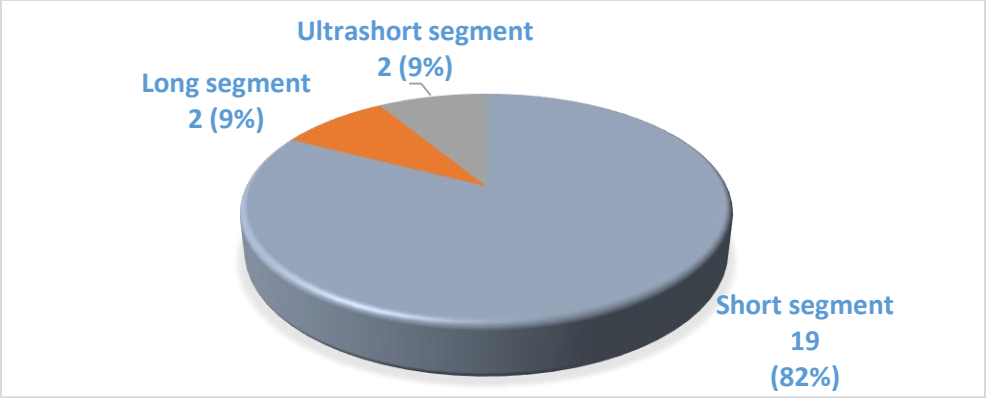

Figure 1: Level of aganglionosis among children with Hirschsprung's disease at diagnosis

Surgical Management

The median age at definitive surgery was 2 years (IQR: 1.5–3 years), with a range of 10 months to 15 years. The definitive surgical procedure was the transabdominal Soave–Boley endorectal pull-through, performed in 19(82.6%) children. Eighteen of these were performed in two stages, with a levelling colostomy before the pull-through, while one child had a single-stage procedure. Two neonates who presented with caecal perforation had diverting ileostomies (Table III).

Outcomes

Eighteen children developed stoma-related complications (78.3%), while 10 (43.5%) developed complications related to their definitive surgery. The most common stoma-related complication was peristomal dermatitis in 9 (39.1%), while abdominal wound infection was the most frequent complication after pull-through in 7 (30.4%), as shown in Table IV.

Four children (17.4%) required re-operation with complications following the pull-through surgical technique. Two (8.7%) had open drainage of pelvic abscesses, while two children had stomas created for an anastomotic leak and post-operative enterocolitis, respectively. The patient who had post-operative enterocolitis had a redo coloanal anastomosis nine months later for residual aganglionosis. Three patients died in this cohort, giving a mortality rate of 13.0%. Follow-

up information was available for 18 children (78.3%), with a follow-up duration of 1 to 2 years. Most patients achieved satisfactory bowel function, and none developed faecal incontinence or soiling (Table IV).

Table III: Types of surgical procedures performed in children with Hirschsprung's disease

Procedure type	Operation	Frequency (%)
Two-stage	Levelling colostomy + Soave-Boley endorectal pull-through	18 (78.3)
One-stage	Soave-Boley endorectal pull-through without stoma	1 (4.3%)
One-stage	Posterior myectomy	2 (8.7)
Other	Ileostomy without pull-through	2 (8.7)
Total		23 (100)

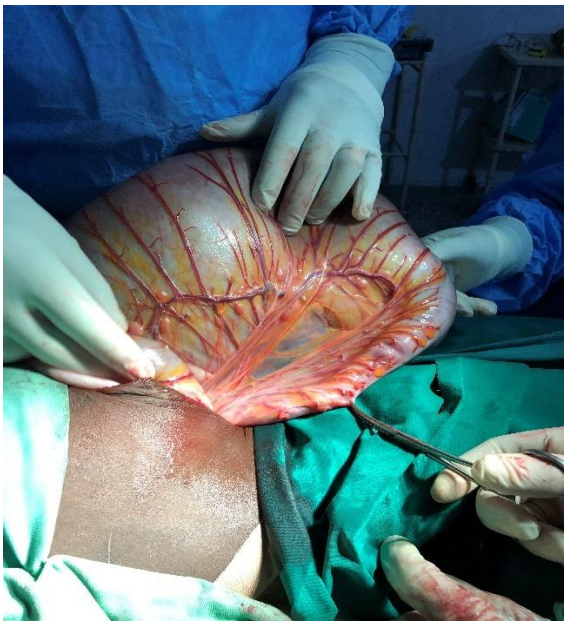

Figure 2: Intraoperative view showing the funnel-shaped transition between the dilated proximal colon and the narrowed aganglionic segment in rectosigmoid Hirschsprung's disease

There was no statistically significant association between age at presentation and stoma-related complications (p = 1.00), pull-through complications (p = 0.508), re-operation (p = 0.21), or mortality (p = 0.27) (Table IV).

Discussion

In this study, most children presented beyond the neonatal period, with abdominal swelling and constipation as the leading symptoms. The majority required staged surgery with a preliminary colostomy before definitive pull-through. Post-operative complications were mainly stoma-related or superficial wound infections, and a small number required re-operation. The mortality rate was low, and follow-up care showed favourable functional outcomes in the majority of children managed.

Delayed diagnosis of Hirschsprung's disease has been defined in various ways in the literature, with some authors considering any diagnosis made after the neonatal period as delayed. In contrast, others use the age of one year as the threshold. [8] Regardless of definition, late diagnosis remains a persistent problem in low- and middle-income countries, where many children escape recognition during infancy and where delays have been reported in up to 72% of cases. [11]

In the present study, only about one-third of patients were diagnosed in the neonatal period, which is consistent with findings from other centres in similar settings. [9,12]

Table IV: Post-operative outcomes in children with Hirschsprung disease categorised by age at presentation*

Complication/Outcome	< 1 month	>1-12 months	>1 year	Frequency (%)	p-value
Stoma-related Complications					1.000
Wound infection	2	2	2	6 (26.1)	
Peristomal Dermatitis	3	5	1	9 (39.1)	
Stomal Prolapse	0	2	4	6 (26.1)	
Diarrhoea	1	0	0	1 (4.3)	
Pull-through complications					0.508
Abdominal wound infection	1	3	3	7 (30.4)	
Pelvic Abscess	0	0	3	3 (13.0)	
Perianal Dermatitis	1	1	0	2 (8.7)	
Anastomotic leak	0	0	1	1 (4.3)	
Enterocolitis	1	0	0	1 (4.3)	
Constipation	1	0	0	1 (4.3)	
Soilage/Incontinence	0	0	0	0 (0.0)	
Re-operation					0.211
Abscess Drainage	`0	0	3	3 (13.0)	
Loop colostomy	0	0	1	1 (4.3)	
Ileostomy creation	1	0	0	1 (4.3)	
Redo coloanal anastomosis	1	0	0	1 (4.3)	
Mortality					0.273
Neonatal Sepsis	`0	0	3	3 (13.0)	
Post-pull through	0	0	1	1 (4.3)	

Note: Age groups reflect age at presentation, not age at treatment

The literature, however, contains several reports of adult or late-diagnosed Hirschsprung's disease, highlighting how delayed presentation remains a global phenomenon. [13-15]

The reasons for delayed presentation in children with Hirschsprung's disease are multifactorial. In many low- and middle-income countries, a large proportion of births occur outside hospitals, often attended by traditional birth attendants who may not recognise delayed passage of meconium as abnormal or requiring medical attention. [11,16] The symptoms may also be mild, especially in ultrashort-segment disease, allowing affected children to go undiagnosed for years. [13,14] In some infants, stooling may appear normal during exclusive breastfeeding, with constipation becoming evident only after weaning, further delaying recognition. Health system factors compound these delays. Access to paediatric surgical care is limited, and constipation, being a common childhood complaint, is often managed conservatively with diet, laxatives, or behavioural modification at the primary care level. [13] Moreover, the widespread cultural practice of using enemas for constipation and other ailments leads many caregivers to rely on home remedies before seeking medical care. [8,17]

Consequently, many children present later in childhood, and some even in adolescence or adulthood, with long-standing severe constipation that has never been properly investigated. [13,16,18]

Despite these challenges, there are clinical features that should raise suspicion for Hirschsprung's disease. These include failure to pass meconium within 48 hours of life, progressive abdominal distension, failure to thrive, persistent constipation without encopresis, dependence on enemas and failure to respond to conservative therapy. [19] In such cases, a rectal biopsy remains the gold standard for diagnosis. However, some authors have suggested that a barium enema, which can demonstrate a transition zone, 24-hour barium retention, and a measurable rectosigmoid index, may be an appropriate first-line investigation in resource-limited settings, particularly for neonates, due of its non-invasive nature and wider availability. [20]

Children with Hirschsprung's disease may present with varying degrees of preoperative morbidity, depending on the disease severity and the healthcare context. Several studies have shown that symptoms such as chronic constipation, malnutrition, enterocolitis, and toxic megacolon are frequent among children at the time of diagnosis, especially in settings where recognition occurs later. [16] In our cohort, more than two-thirds of children experienced chronic constipation before presentation. This finding is consistent with the series by Osterig-Hill *et al.*, who also reported constipation as the predominant symptom. [21] In contrast, Bradnock *et al.*, in a multicentre study in the UK and Ireland, did not encounter constipation but reported abdominal distension, bilious vomiting, and delayed passage of meconium as the most frequent presentations.[22] These differences are likely explained by the earlier age at diagnosis in high-income settings, where most children are identified and treated in the neonatal period or early infancy, leading to a different spectrum of presenting symptoms.

Nearly all patients required a diverting stoma before definitive pull-through. In older children, postponing the pull-through technique until the massively dilated colon has decompressed and regained normal tone reduces the risk of complications and allows for a safer operation. Creating a temporary stoma remains an accepted strategy in such cases, particularly in resource-constrained environments. [7, 23] It also allows time for nutritional rehabilitation in children presenting with malnutrition and failure to thrive. Similar high colostomy rates, often exceeding 80%, have been reported from other centres in Africa and Asia. [10, 24-26] In contrast, single-stage pull-through procedures are increasingly favoured in high-income countries, where early diagnosis and better preoperative stability allow definitive surgery without diversion. [22,27]
In our local practice, colostomies are usually created 5–10 cm proximal to the transition zone. Because frozen-section biopsies were unavailable, we relied on intraoperative visual assessment of peristalsis after stimulation, supplemented by full-thickness colonic biopsies for histological confirmation. The biopsies are taken at three points: proximal to the transition zone, at the transition zone and distal to the transition zone. This approach is similar to what has been described in other resource-constrained settings. [7] The goals of surgery in Hirschsprung's disease are to excise the aganglionic segment and restore continuity with normally innervated bowel down to the anus while preserving sphincteric function. [23]

In this series, children with rectosigmoid disease typically had simultaneous pull-through and stoma closure. This approach was well tolerated, with few early post-pull-through complications. The complication rates were comparable to those

reported in other studies of transabdominal pull-throughs in similar settings. [7,10,23,24] Interestingly, while most of the patients in this series presented late, there was no significant association between age at presentation and the occurrence of early post-operative complications or other short-term outcomes. Other studies have also observed that late presentation does not necessarily correlate with increased early post-operative morbidity. [28,29]

Following pull-through surgery, children are expected to achieve regular bowel opening and continence. Unlike patients with anorectal malformations, the sphincter mechanism is generally normal in Hirschsprung's disease. [30] Long-term outcomes reported in the literature indicate that most children achieve good or normal bowel function following Soave's pull-through, with rates ranging from 60 to almost 100 per cent. [9, 31, 32] Persistent problems, however, are not uncommon. Constipation, soiling, and faecal incontinence are frequently reported, though these often improve with age. [33,34] A minority, estimated at around 15%, may have ongoing symptoms requiring interventions such as bowel management programmes, regular enemas, or redo surgery. [9,35] In our cohort, functional outcomes were favourable. None of the patients who were followed up developed faecal incontinence or soiling. One patient with persistent constipation and subsequent enterocolitis due to residual aganglionosis required redo of the coloanal anastomosis, with a good outcome.

The mortality in this series was attributed to severe faecal peritonitis in two neonates who presented with caecal perforation and to one post-operative death in an older child following the pull-through. Reported mortality for Hirschsprung's disease is generally low across published series.[4,8,9,26] The finding in this study is comparable to that of Rahmo *et al.*, who recorded three neonatal deaths (9.4%), Trovalusci *et al.*, who also reported four deaths (4.2%), and Sholadoye *et al.*, who reported three post-operative deaths. [4,8,9] In contrast, Ekenze *et al.* [26] documented a single death (2.4%). The higher proportion in the present study most likely reflects the acute nature of our patients at presentation, especially bowel perforation and established sepsis, as well as constraints in perioperative and neonatal intensive care at the hospital.

The strength of this report is that it represents one of the few series on Hirschsprung's disease from the southern region of Nigeria, contributing local data from a resource-limited setting. The nine-year span allows for the capture of a wide spectrum of cases, including neonates and older children. It provides a comprehensive view of the challenges and results of managing Hirschsprung's disease in this context.

The study is limited by its retrospective design, which relied on the completeness of hospital records. Functional outcomes were assessed from routine post-operative follow-up notes (where no standardised scoring tools were applied). The long-term evaluation was restricted because most patients did not return beyond the first two years after surgery. This limited follow-up introduces the possibility of attrition bias. In addition, the small number of patients reduced the ability to perform subgroup comparisons or identify predictors of outcome. Despite these limitations, the report provides information on patterns of presentation and outcomes that reflect the realities of practice in low-resource settings.

Conclusion

This case series showed that most children presented beyond the neonatal period, reflecting persistent challenges with early recognition and referral in resource-limited settings. Despite these delays, the outcomes achieved in this study were satisfactory, with early and late outcomes

comparable to those reported from similar settings.

Strengthening diagnostic capacity, improving community health awareness, and providing continuous education for primary care physicians and frontline health workers remain important strategies to promote early diagnosis and timely surgical intervention. Further prospective studies are needed to define long-term functional outcomes better and to evaluate post-operative quality of life among children treated for Hirschsprung's disease in resource-limited settings.

Authors' Contributions: AEI and IMA conceived the study. AEI did data curation and wrote the draft of the manuscript. IMA, EME, and EAE participated in data curation and analysis. AEI, AIC and EME performed the literature review and contributed to data interpretation. All the authors revised the draft for sound intellectual content and approved the final version for publication.

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