



Original Article

Clinicopathological Characteristics of Hirschsprung's Disease: A Single-Center Study in Bangladesh

Chakraborty D¹, Rahman SMM², Sufian A³, Rahaman MA⁴, Alam MM⁵

Abstract

Background: Hirschsprung's disease (HD) is the congenital absence of parasympathetic ganglionic cells within the distal intestine. This leads to a lack of gut motility, stool stasis, and constipation. HD is relatively common in children. This study aims to identify the clinic-pathological characteristic of HD patients in a tertiary care hospital in Bangladesh. **Materials and Methods:** This cross-sectional observational study was carried out in the department of pediatric surgery, Mymensingh Medical College Hospital, Mymensingh from January 2018 to April 2019 with ethical clearance from respective IERB. Sixty cases of clinically, radiologically consistent with Hirschsprung's Disease not in acute stage, chronic constipation with assisted defecation in older children and children who came with colostomy for subsequent management without tissue diagnosis were purposively included in the study. In each case, information about the patients was obtained in a predesigned questionnaire after obtaining written consent of the parents/guardians in the consent form. All the patients underwent rectal punch biopsy. Variables were expressed as mean \pm SD, frequency and percentage. **Results:** The age range of the study group was 3 days to 7 years with a mean age of 1.20 ± 1.90 years. Among the 60 patients 40 (66.6%) patients were male and the rests 20 (33.4%) were female with a ratio of 2:1. Most of the patients were in neonatal period (25, 41.66%) followed by infants (18, 30%). In the study cases delayed passage of meconium was in 42 patients, constipation was in 58 patients, abdominal distensions were in 42 patients, vomiting was in 25 (41.66%) patients. **Conclusion:** The clinical presentation of Hirschsprung's disease ranges from neonatal intestinal obstruction to chronic progressive constipation in older children. It is expected that early presentation and diagnosis of the disease enhance early surgical correction and prevent fatal complications among the children.

Keywords: Hirschsprung's disease, Clinicopathological characteristics, X-ray diagnosis.

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Introduction

Hirschsprung's disease (HD) is a congenital disorder identified by the absence of ganglion cells at Meissner's plexus of the submucosa and Auerbach's plexus of the muscularis. Typically, it is characterized by vague symptoms like persistent constipation¹. The absence of ganglion cells causes the colonic segments to remain permanently contracted and tonic state. The presence of feces prevents the internal anal sphincter from relaxing. This leads to a lack of gut motility, stool stasis, and constipation². In 65% of cases, the diagnosis is made before the patient turns one month old, and in 95% of cases, it is made before the patient turns one-year old¹.

HD was first described in 1691 by Frederik Ruysch, a Dutch anatomist and surgeon, as a phenomenon associated with very dilated colon disorders^{3,4}. Nonetheless, at the German Pediatric Society

conference in Berlin in 1886, the disorder was given its name in honor of Harald Hirschsprung, where Hirschsprung discussed the cases of two babies who passed away from intestinal blockage-related complications^{4,5}. HD, another name for congenital aganglionic megacolon, which produces a functional obstruction and proximal dilatation of the affected segment^{6,7}.

About 1 in 5000 live births have Hirschsprung's disease. It is divided into 2 types based on the degree of innervation. The short segment, which only affects the rectosigmoid region of the gut, is the most common and is seen in 80-85% of patients with HD. Up to 20% of cases of long segment disease have aganglionosis that extends proximally to the sigmoid colon. Aganglionosis of the entire colon is less common, affecting only 3%-8% of HD patients. Another rare variant is ultra-short segment disease,

¹Debashis Chakraborty, Assistant Professor, Dept. of Paediatric Surgery, Eastern Medical College & Hospital, Cumilla, Bangladesh.

²SM Mahfuzur Rahman, Assistant Professor, Dept. of Surgery, Eastern Medical College & Hospital, Cumilla, Bangladesh.

³Abu Sufian, Assistant Professor, Dept. of Paediatrics, Eastern Medical College & Hospital, Cumilla, Bangladesh.

⁴Md. Arifur Rahaman, Assistant Professor, Dept. of Paediatric Surgery, Central Medical College & Hospital, Cumilla, Bangladesh.

⁵Md. Mazharul Alam, Assistant Professor, Dept. of Paediatric Surgery, Mainamoti Medical College & Hospital, Cumilla, Bangladesh.

Address of Correspondence: Dr. Debashis Chakraborty, Assistant Professor, Department of Paediatric Surgery, Eastern Medical College and Hospital, Cumilla, Bangladesh. Mobile: +8801722722636. Email: dcborty22@gmail.com

affecting only the distal rectum (≤ 2 cm)⁸⁻¹⁰. Short-segment disease is 2 to 4 times more likely in males than females, though long-segment disease has equal gender ratios¹¹. Abdominal distension, vomiting, and delayed meconium passage are the traditional triad of symptoms associated to HD. The latter symptom is seen within 80% to 90% of afflicted patients¹². A digital examination may reveal a tight anal sphincter and an explosive bowel movement. Enterocolitis can manifest in approximately one-third of patients. Fecal stasis causes mucosal inflammation and bacterial overgrowth, which results in leukocytosis, vomiting, fever and severe diarrhoea¹³. If untreated, perforation of the bowel, septic shock, and death can result. Overall, 18% to 50% of patients with HD develop enterocolitis in their lifetime¹².

Abdominal X-ray, contrast enema (CE) and anorectal manometry (ARM) can be used for the diagnosis of HD. But the gold standard for an HD diagnosis is a rectal suction biopsy (RSB), which shows an elevated cholinesterase activity and may even show aganglionosis in case of HD¹⁴. Surgery is the definitive management for patients with HD. The objective of the surgical procedure is to excise the segment of the colon lacking ganglion cells and establish a connection between the portion of colon and the rectum with the aim of reinstating normal physiological function¹⁵. This study aims to identify the clinic-pathological characteristic of HD patients in our institution in Bangladesh.

Materials and Methods

This cross-sectional observational study was conducted in the department of pediatric surgery, Mymensingh Medical College Hospital, Mymensingh from January 2018 to April 2019. 60 cases of suspected HD were purposively selected for this study fulfilling the exclusion & inclusion criteria for the study. Ethical approval of the study was taken from the Ethical Review Board (ERB) of MMCH (ref: MMC/IRB/2018/70). Clinically, radiologically consistent with HD not in acute stage, chronic constipation with assisted defecation in older children and Children who came with colostomy for subsequent management without tissue diagnosis were included in the study. Patients with clinically HD with acute intestinal obstruction who needed immediate surgical intervention and whose legal guardian will not provide valid consent were excluded from the study.

In each case, information about the patients was obtained in a predesigned questionnaire after obtaining written consent of the parents/guardians in the consent form. All the patients underwent rectal punch biopsy due to its easy availability instead of RSB and specimen were sent to histopathological examinations. The biopsy was performed in the

lithotomy position with no anaesthesia. A biopsy tube was inserted into the rectum with the side hole facing the posterior wall of the rectum. Post biopsy we followed the patient for any per rectal bleeding, abdominal distension, temperature or any local infection. Every patient was advised to use no suppository or enema for 48 hours. All the data was processed and analyzed using Microsoft Excel and IBM-SPSS v23.0 for Windows. Statistical inference was based on 95% confidence interval and p-value <0.05 was considered statistically significant. Variables were expressed as mean \pm standard deviation (SD), frequency and percentage. The summarized data were presented in the form of tables and figures.

Results

In this cross-sectional observational study, a total of 60 clinically and radiologically consistent with HD not in acute condition patients were included. The age range of the study group was 3 days to 7 years with mean age 1.20 ± 1.90 years (table-I). Table 1 also shows that among the 60 patients 25 (41.66%) were within the neonatal period which were maximum and 18 (30%) were infants with the second incidence. In this study 40 patients (66.6%) were male and the rests 20 (33.4%) were female with a ratio of 2:1 (figure-1).

Table-I: Age distribution of the study cases (n=60)

Age Group	Frequency (%)	Mean \pm SD
Neonate	25 (41.66%)	1.20 \pm 1.90
Infant	18 (30%)	
Older Children	17 (28.33%)	

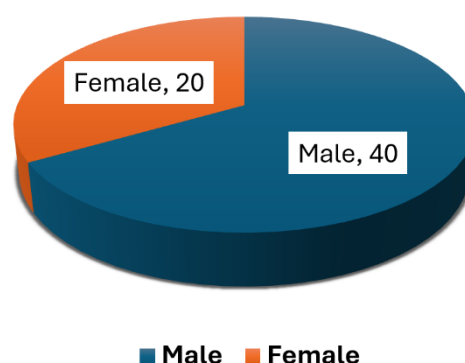


Figure-1: Distribution of Gender among the study cases (n=60)

In table-II shows that delayed passage of meconium was in 42 patients among which 25 (41.66%) patients within the neonatal period, 10 (16.66%) were infant and 7 (11.66%) were older children. Constipation was in 58 patients among which 25 (41.66%) were neonate, 16 (26.66%) were infants

and 17 (28.33%) were older children. Abdominal distensions were in 42 patients among which 25 (41.66%) were neonate, 10 (16.66%) were infants and 7 (11.66%) were older children. Vomiting was

in 25 (41.66%) patients in which all were neonate. Assisted defaecation were in 34 patients among which 17 (28.33%) were infants and 17 (28.33%) were older children.

Table-II: Presenting complaints of the study cases (n=60)

Age group	History of delayed passage of meconium	Constipation	Abdominal distension	Vomiting	Assisted defaecation
Neonate	25 (41.66%)	25 (41.66%)	25 (41.66%)	25 (41.66%)	0
Infant	10 (16.66%)	16 (26.66%)	10 (16.66%)	0	17 (28.33%)
Older Children	7 (11.66%)	17 (28.33%)	7 (11.66%)	0	17 (28.33%)
p-value	0.003*	0.001*	0.003*	--	0.093*
Level of significance	Significant	Significant	Significant	--	Not significant

*p-value obtained from χ^2 test

Discussion

Hirschsprung's Disease is a troublesome surgical problem in children especially in neonates which cause neonatal intestinal obstruction¹⁶. In this study, age ranges were 3 days to 7 years with a mean age of 1.20 ± 1.9 years. Other studies like Yoshimaru, et al.¹⁷ showed an age range from 1 day to 13 years, Henna, et al.¹⁸ showed the mean age was 1.9 years and Haricharan, et al.¹⁹ showed mean age was 4.4 years. Age distribution in this study is more or less consistent with these studies. This is the cause of cultural differences, neglect of initial symptoms poverty and prolonged treatment with enema and laxatives. In this study 40 patients were male and the rests 20 were female with a ratio of 2:1. The ratio of male: female was 4:1 in a study by Henna, et al.¹⁸. Male predominance in the gender ratio in the present study were also consistent with the study by Muise, et al.²⁰

Classical signs of HD are delayed in passage of meconium, abdominal distension and vomiting. Delayed in passage of meconium beyond 48 hours after birth is thought to be an important diagnostic clue and more important if it is associated with chronic constipation. In our study 42 (69.98%) population had a history of delayed passage of meconium and 18 (30.02%) had no history of delayed passage of meconium. Islam, T²¹ study showed 30 (63.8%) had history of delayed passage of meconium and Hannan, et al.²² study showed 72.23% had history of delayed passage of meconium. So, this study finding conforms to that of other studies and validates the fact that the passage of meconium beyond 48 hours of life is an important diagnostic clue of HD.

Constipation is another presenting feature of HD. In this study 96.66% of cases had constipation. Among them 41.66% were neonate, 26.66% were infants and 28.33% were children. Islam, T²¹ study showed 63.8% patients present with the history of

constipation and Hannan, et al.²² study found 78% cases of constipation. So, constipation is considered as a very important diagnostic clue of HD. A total of 42 (70.00%) cases of the study had mild abdominal distension and 25 (41.66%) had vomiting which was bilious in nature. After a single episode of per-rectal wash abdominal distension regressed and vomiting subsided. Islam T²¹ study showed abdominal distention and vomiting were 57.4% and 29.8% respectively. Henna, et al.¹⁸ study found abdominal distension was 91.7%, vomiting was 50.3% which is similar to this study. Assisted defecation was another finding to diagnose HD. In this study 34 (56.66%) had a history of assisted defecation with the aid of glycerin suppository.

Even though the exact etiopathogenic pathways underlying HD remain unclear, recent advancements have yielded noteworthy outcomes. Notwithstanding partial knowledge about the etiopathogenic mechanisms, studies have unveiled parts of the embryonic development²³. The most distinctive factor involved in the etiopathology of this disease is the neural crest disorder, causing the absence of these neural cells²⁴.

Conclusion

The clinical presentation of Hirschsprung's disease ranges from neonatal intestinal obstruction to chronic progressive constipation in older children. It is expected that early presentation and diagnosis of the disease will enhance early surgical correction and prevent fatal complications among the children.

Conflict of Interest

The authors declared that they have no conflicts of interest.

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