

Hirschsprung's Disease: Pattern of Clinical Presentation

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ABSTRACT

Objective To report clinical symptoms and signs and age at initial presentation of patients with Hirschsprung's disease.

Study design Descriptive case series.

Place & Duration of study Department of Paediatric Surgery, National Institute of Child Health Karachi, From September 2008 to December 2013.

Methodology Patients who were proven cases of Hirschsprung's disease on rectal biopsy report were included. The data was collected prospectively. Variables recorded included clinical presentation including symptoms, signs, age, gender, mode of presentation either elective or acute etc. The data was analyzed using descriptive statistics like frequency, percentage and proportions in relation to quantitative and qualitative variables.

Results A total of 84 patients with Hirschsprung's disease were managed during the study period. There were 63 male and 21 female patients with male to female ratio of 3:1. Fifty-five (65.5%) patients belonged to Karachi. Fifty percent (n=42) patients presented by day 6 of life. The commonest presentation was delayed passage of meconium but it was reported in 39 (46.4%) patients only. All these patients were operated and extent of the disease was up to rectosigmoid level in 78 (93%) patients.

Conclusion Clinical presentation in present study had similarities to reported literature but important differences were noted with regards to passage of meconium and extent of aganglionosis.

Key words Hirschsprung's disease, Clinical profile, Meconium: delayed passage.

INTRODUCTION:

The incidence of Hirschsprung's disease from Western countries is reported as 1:5000 births.¹ It is an outcome of failure of migration of ganglion cells in craniocaudal direction though much information has been gained in relation to its etiology over the past few decades than this simpler explanation.² Most of the babies with this disease presents with failure to pass meconium in first 24 hours after birth. Some may have acute presentation with intestinal obstruction and gut perforation.³

Hirschsprung's disease has been further classified based upon extent of absence of ganglion cells in gut wall, a hallmark of the condition, into classical short segment (up to sigmoid colon) and long segment disease the extent of which may include whole colon and at times variable extent of small bowel.⁴ Internal sphincter achalasia is another type with distinct clinical presentation and histological criteria.⁵

Hirschsprung's disease is not an uncommon condition in oriental race. Though condition is well reported from our part of the world but still number of patients with classical symptoms and signs are not recognized early.^{6,7} The delayed presentation adds to morbidity and at times mortality. It is therefore essential that all those medical and allied health professionals who are involved in the care of children, must be aware of varied presentation of this condition so as to make early referral to appropriate facility for further

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investigations and treatment. This study describes our experience of patients with biopsy proven Hirschsprung's disease with regards to pattern of clinical presentation.

METHODOLOGY:

This descriptive case series was conducted at the Department of Paediatric Surgery National Institute of Child Health Karachi, from September 2008 to December 2013. Patients suspected of Hirschsprung's disease who on further investigations and rectal biopsy, reported as having absence of ganglion cells, were included. The data related to age at presentation, gender, clinical symptoms and signs, mode of presentation as elective cases in outpatient department or acute presentation in Emergency Room, findings on barium enema, extent of aganglionosis at colostomy, were recorded.

The data were entered into a perform and SPSS version 17. Descriptive statistics were used for analysis of quantitative variables like age and qualitative variables like gender, clinical symptoms, in frequency and percentages.

RESULTS:

A total of 84 patients were included. There were 63 (75%) males and 21 (25%) females. Male to female ratio was 3:1. Fifty-five (65.5%) patients belonged to Karachi while others were brought from other parts of Sindh and Baluchistan provinces. Overall 39 (46.6%) patient gave history of passage of meconium after 24 hours while in 35 (41.7%) patients baby did pass meconium in variable quantity within 24 hours of birth. In ten cases parents could not recall as to the timing of passage of meconium. Abdominal distension was noted in 20 (23.8%) patients while five (6%) patients presented with acute obstruction. In addition nine (10.7%) patients had pneumoperitoneum at the time of admission to ER. Constipation was the presenting symptom in 27 (32.1%) patients. Many patients had more than one symptom at the time of presentation (table I).

Two patients with pneumoperitoneum presented on day 3 and two on day 4 of life. These patients did not pass meconium and developed abdominal distension. Patients who presented with acute symptoms were mostly below three weeks of age (n=51,60.7%). Constipation was the predominating complaint in older neonates and infants (n=27, 32.1%).

A wide variation in age distribution was noted at the time of presentation to our hospital. Only 11 (13%) patients presented on day-2 of life while 50% (n=42) patients were brought by day-6 of birth. Out of these delayed passage of meconium was the complaint in 15 (17.8%) patients while 27 (32.1%) patients had abdominal distension which was of concern to the parents. Barium enema was performed in 66 (78.5%) patients. In four (4.8%) cases it was not suggestive of Hirschsprung's disease.

Partial thickness rectal biopsy was performed in all the patients and stoma made as a stage procedure. In four cases at stoma formation the whole colon was found collapse. Ileostomy was made in these cases. Biopsy report suggested total colonic aganglionosis. In two cases extent of aganglionosis was up to transverse colon (table II).

DISCUSSION:

Hirschsprung's disease has varied presentation thus many patients may not be identified early in the course of the disease. Same was the observation in the present study. The number cases managed in six years duration in present study was higher than a reported series from United States where in eleven years, 91 patients were managed.⁸ But our data is quite different from that reported from two hospitals in Lahore, where number was far more than present study.⁷ These numbers do not convey the actual population based statistics as present study, as well that from US, provide hospital based data.

Table I: Modes of Presentation

Symptoms	Frequency (n)	Percentage (%)
Not passed meconium in first 24 hours after birth	39	46.4
Abdominal distension	20	23.8
Pneumoperitoneum	09	10.7
Acute intestinal obstruction	05	6.0
Constipation	27	32.1
Chronic abdominal distension	03	3.6

Table II: Extent of Aganglionosis and Gender

Extent of Aganglionosis	Gender		Total n (%)
	Male (n)	Female (n)	
Up to Rectosigmoid	57	21	78 (92.8%)
Up to Transverse colon	02	00	2 (2.38%)
Whole colon	04	00	4 (4.76%)
Total	63	21	84

Hirschsprung's disease has a known predilection for male gender. It occurs five times more commonly in males than females.⁹ Our data is quite similar to previously reported series but number of females affected was slightly on higher side. The male to female ratio in our series was 3:1.

Hirschsprung's disease is one of the common causes of intestinal obstruction in neonatal period and must be considered in the differential diagnosis.¹⁰ Failure to pass meconium does not convey information holistically. It is therefore important to ask probing questions related to timing, quantity and quality of meconium passed. Passage of small quantity of light green meconium should be of concern. Same was noted in present series as non passage of meconium was found in only 20 patients. Other patients did pass meconium but in variable quantity. This symptom, therefore, must be interpreted in combination with abdominal distension which was noted in equal number of patients in present series. With adequate amount of meconium passage, abdominal distension must not occur early.

Symptoms in infants included constipation with chronic abdominal distension.¹¹ In our series many of these older children were malnourished. Various drugs and home remedies were used in them to relieve constipation. Seven children in our study were older than one year at the time of presentation with fecal impaction and pot belly. In recent literature from western countries diagnosis is established early, due to increased awareness about the disease.

Pneumoperitonium is a known complication of Hirschsprung's disease in neonatal period that may have grave consequences.¹² In this series nine (10.7%) patients presented with this complication. All of them underwent laparotomy with stoma formation. The reported incidence of this complication is 4%.¹³ One of our patients with pneumoperitoneum was 2 months of age. A high index of suspicion for Hirschsprung's disease must be exercised in cases of neonatal appendiceal and cecal perforation.¹⁴ All such patients must have

multiple colonic and rectal biopsies. Same was practiced in present series.

Contrast barium enema was done in 66 patients. This was not of much help in terms of diagnosis though it did help in augmenting clinical impression. Variable findings were noted on x ray from classical description of Hirschsprung's disease to only few findings like colonic dilation (n=13, 15.5%), and retained contrast after 24 hours (n=22, 26.2%). Clear transition zone was noted in only 17 (20.2%) cases. X rays did help in planning surgery. The role of diagnostic barium enema remained controversial. A high diagnostic yield of 87% - 89% was reported in some series which is duplicated in experiences of others.^{15,16}

Rectal biopsy remains the gold standard for making definitive diagnosis.¹⁷ In this series all patients had partial thickness rectal biopsy. Suction rectal biopsy is a patient friendly procedure with high diagnostic accuracy.¹⁸ This facility was not available at our hospital. All patients in this series had leveling colostomy. Endorectal pull through procedure was scheduled for later date.

In overwhelming majority (93%) of patients in this series the disease was limited to rectosigmoid area, the short segment variety. This is quite different from other series.¹⁹ The reason for this pattern cannot be ascertained. This may be a reflection of regional uniqueness but no similar report could be found in literature. Only four (4.76%) patients had total colonic aganglionosis, the incidence of which varies from 0% to 8%.²⁰ Two patients had Shah Waardenburg syndrome which is an autosomal recessive neurocristopathy.²¹ In this condition there is an association of Waardenburg syndrome with Hirschsprung disease. This is grouped under Waardenburg type IV. Till 2008 less than 50 cases were reported in literature.²²

This study highlighted varied pattern of Hirschsprung's disease. This uniqueness leads to

frequent delay in diagnosis. By creating awareness the late diagnosis can be minimized with its related morbidity and at time mortality.

CONCLUSIONS:

History of delayed passage of meconium, though characteristic presentation of Hirschsprung's disease, may not be obtained in all the patients. Majority of the patients in present series had short segment variety of the disease. Pneumoperitoneum was found in 10% patients at presentation.

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