Hirschsprung's Disease: Experience Of 145 Patients

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This is a report of our experience of 145 patients of Hirschsprung's disease. In this series the male to female ratio was 3.83:1 . Majority (76.55%) were born to mothers less than 30 years of age. Less than 12% were first born. Fiftyfive patients presented with in first month of life. Thirteen patients had associated anomalies. Down's syndrome and umbilical hernia were the commonest associated anomalies. Progressive abdominal distension, chronic constipation and acute intestinal obstruction were common presenting symptoms. No premature patient was seen in this series. Majority of the patients belonged to poor socioeconomic group. In 131 patients, level of aganglionosis was determined , with 102 (77.27%) cases of short segment disease. Out of 145 patients, only 77 patients reported for definitive operation during the study period. In 66 cases an Endorectal pullthrough and in 11 patients Duhamel's procedure was performed. More than 70% patients were fully continent in this series with both the procedures. Overall mortality in this series was 14.48%.

Key Words: Hirschsprung's disease, Duhamel's procedures, Boley's Modification of Soave's endorectal pullthrough, Swenson's pulthrough.

Hirschsprung's disease is not an uncommon cause of constipation in children. Diagnosis and treatment Hirschsprung's disease is well established in the west but still poses a major problem in our country. Majority of our population is uneducated and the knowledge of our doctors about Hirschsprung's disease as a major cause of chronic constipation is also limited. These are the major reasons for delay in diagnosis and mismanagement of these cases.

There are many surgical procedures presently being performed for Hirschsprung's disease. This study compares the results of earlier cases managed by an Endorectal pullthrough and the results of Duhamel's procedure performed in the later part of this study. These are the two most popular surgical procedures available at present [1,2]. The clinical significance and relative importance of different clinical factors have also been evaluated critically.

Materials and Methods:

A prospective study was started at Mayo Hospital (Jan. 1988 to Dec. 1992), The Services hospital (Jan. 1996 to Oct. 1997) and The Children's Hospital Lahore (Jan. 1997 to Dec. 1997). Detailed history was taken in the patients who presented with symptoms suggestive of Hirschsprung's disease. A plain X-ray of the abdomen and barium enema were the main diagnostic modalities. The diagnosis of Hirschsprung's disease was made only if aganglionosis was confirmed on histopathology of a full thickness rectal biopsy taken 2 cm above the dentate line. Some cases were diagnosed at laparotomy for intestinal obstruction and confirmed by serial segmental biopsies. Enterostomy was created at the most distal normally ganglionic bowel in the dilated part above the transitional area or cone. Post operatively the patients were reviewed monthly at follow-up. Every patient was offered the definitive procedure at a minimum of 6 months after colostomy/ileostomy or at the age of six months or older if colostomy was done in the neonatal age. A regular follow up after every month was carried out to note the complications and results of the operation.

The continence of the patients was assessed by a quantitative method which is based on the suggestions of Soper and Figuera (1971)[2] and Templeton and Diteshein (1984)[3]. A score of 4-5 is considered good, 2-3 is fair and a score of less than 1.5 is poor.

Results:

During the study period a total of 145 patients were diagnosed as suffering from Hirschsprung's disease. In a total of 145 cases, 115 were male and 30 were females giving M:F ratio of 3.83:1.

Only one patient had family history of Hirschsprung's disease, some had history suggestive of the disease in their family. Out of these 145 patients, 49 were product of a consanguineous marriage, 29 parents were 2nd cousins , whereas sixty nine parents were unrelated (Table 1).

Table 1. consanguinity of parents: (n = 145)

Relation	n=	%age
First cousin	49	33.79%
Related but not first cousins	27	18.62%
Unrelated	69	47.58%

We also noted the maternal age at the time of birth of these patients .Majority of the patients [76.55%) were born to mothers less than 30 years of age and one patient was born after 40 years (Table 2).

Majority of our patients(70%) were breast fed. More than 70% of our patients belonged to poor socioeconomic group.(Table 3)

In our series 13 (8.96%) patients had associated anomalies. Down's syndrome and umbilical hernia were the dominant associated anomalies seen in.(Table 4).

Table 2.Iaternal age at birth of patients:(n = 145)

No	Age of mother	No of patients	%age
1	.<20 years	15	10.34%
2	20-25 years	55	37.93
3	25-30 years	41	28.27%
4	30-40 years	33	22.75%
5	> 40 years	01	0.69%
Total		145	100

Table 3: Socioeconomic status of parents: (n = 145)

Earning	n=	%age
<2000Rs	46	31.72%
2000-5000Rs	78	53.79%
5000-10000Rs	12	8.27%
> 10000Rs	9	6.20%

Table 4: Associated anomalies:(n = 145)

Anomaly	No of patients	%age
TEV	02	1.38%
Umblical hernia	03	2.07%
Down's syndrome	04	2.75%
Inguinal hernia	02	1.38%
Hydrocephalus	01	0.69%
Malrotation of Gut	01	0.69%
Total	13	8.96%

Majority of the patients (38%) presented before the age of 1 month and 51% with in the first year of life. The early presentation dominated in the late years. The oldest patients in our series presented at the age of 11 and 13 years respectively(Table 5).

Table 5:age at time of presentation:(n = 145

Age	No of patients	%age
<1 month	55	37.93%
1 month-1year	19	13.00%
1-5 years	47	32.40%
5-10 years	24	16.55%
>10 years	02	1.38%
Total	145	99.98%

Time lapse between appearance of symptoms and presentation is reflected in (Table 6).

Table 6: Time lapse between appearance of symptoms and presentation:(n=145)

Duration	n=	%age
< 1 month	55	37.93
1 month to 1year	48	33.10
1-4 years	34	23.34
>4 years	08	5.52

The most common presenting symptoms were progressive abdominal distension (71), chronic constipation(55), acute intestinal obstruction(49), Vomiting(46) and fecal soiling due to overflow incontinence(16). History of delayed passage of meconium was present in 47.58% of the patients. Hirschsprung's associated enterocolitis (HAEC) manifested by diarrhea, fever, abdominal distension and prostration was seen in 5 patients in this series.(Table 7)

Table 7: Presenting complaints(n = 145)

Presenting complaints	n=	%age
Chronic constipation	55	37.93%
Acute intestinal obstruction	49	33.79%
Vomiting	46	31.72%
Progressive abdominal distension	71	48.96%
Loss of appetite and failure to grow	36	24.83%
Delayed passage of meconium	69	47.58%
Enterocolitis (fever, abdominal distension, diarrhea and prostration)	05	3.45%
Fecal soiling	16	11.34%

Level of enterostomy:
Total 131 patients agreed for laparotomy/colostomy . Transverse colostomies were performed in the early period in 42 patients. Subsequently pelvic end colostomies were performed in 74 patients, Cecostomy in one patient and Ileostomy was performed in 11 patients suffering from total colonic aganglionosis and skip lesion .

Extent of aganglionosis:
The upper level of aganglionosis was assessed by naked eye appearance at laparotomy and confirmed by segmental histopathology. It was noted in 132 patients. In 102 patients, it was present at the level of rectum or rectosegmoid and 7% had total colonic aganglionosis (TCA) with or without ileal involvement. The very rare variety of skip lesion was seen in 2 patients in our series. One patient had ultra short segment disease, he was relieved of symptoms with rectal biopsy and myectomy.

Pullthrough operations:
A total of 77 patients reported for definitive operation during the study period. Out of these in 56 patients Boley's modification of Soave's operation was performed. These also include three patients of total colonic aganglionosis who were treated with ileoanal Endorectal pullthrough and two patients neediing a repeat pullthrough. In 10 patients original Soave's operation was performed. In eleven patients Duhamel's procedure was performed.

Complications and mortality:
131 patients had an initial laparotomy/ enterostomy. Major complications encountered after laparotomy and colostomy were burst abdomen(4), paracolostomy evisceration(5), skin excoriation(28) and recurrent prolapse of stoma(30) . All patients with TCA suffered from diarrhea and fluid and electrolyte imbalances and 6 of 9 patients died before definitive surgery. Seven patients developed septicemia and 6 of them died. Two patients died in the early post operative period due to major transfusion reaction and the other one from pneumonia.

There was one death due to complications of anaesthesia during induction..

Perianal skin excorriation(15), burst abdomen[4] and wound infection12 were the early complications seen with Endorectal pullthrough. Mucosal prolapse was seen in two patients. In one patient bleeding from the stump occurred and needed cautry. The early complications encountered with Duhamel's procedure were wound infection[1], perianal skin excorriation[2] and bleeding from the spur which was relieved with cauterization in one patient.[Table 8)

Table 8:Complcations of pullthrough: (n = 77)

Complications	Endorectal procedure n=66		Duhamel's n=11	procedure
EARLY	n=	%age	n=	%
Wound infection	12	18.18	01	9.09
Burst abdomen	04	6.06	00	
Mucosal prolapse	02	3.03	00	
Perianal skin excorriation	15	22.72	02	18.18
Bleeding from stump/ spur	01	1.51	01	9.90
Stitch abscess	01	1.51	00	0.0
LATE				0.0
Anal stenosis & constipation	15	22.72	00	0.0
Constriction ring	10	15.15	00	0.0
Adhesion obstruction	02	3.03	01	9.90%
Enterocolitis	02	3.03	00	0.0
Fecal fistula	01	1.51	00	0.0
Recurrence of symptoms	02	3.03	00	0.0

Anal stenosis(15), and constriction ring formation(10) were the common late complications seen with Endorectal pullthrough. Anal stenosis responded to anal dilatation whereas a constriction ring needed surgical division. Postoperative enterocolitis was seen in two patients with Endorectal pullthrough with 100% mortality, they had presented late and parents of one of them refused treatment. Another patient developed an abscess on the right buttock 3 months after ERPT which was drained by a doctor at a DHQ hospital, with unexplained death after one week.. Adhesion obstruction developed in three patients, two with ERPT and one with Duhamel's procedure. First two patients responded to conservative measures while the third patient managed with Duhamel's procedure, presented late at a DHQ hospital and was operated there but did not survive.

Continence:

The follow up ranges from 4 months to 7 years. In case of ERPT 76.19% of cases were fully continent while 10 % had a fair control over defecation. Only 5 patients (7.93%) had poor control over defecation and are declared as incontinent.

With Duhamel's procedure 70% of the patients are fully continent while 20% had fair control over defecation. Only one patient (10%) has a poor control with a follow-up of only 6 months.[Table 9)

Table 9: Continence score:

Quality of continence	Endorectal pullthrough n= 63		Duhamel's pullthrough n=10	
	Number	%age	Number	%age
Fully continent	48	76.19	07	70
Fairly continent	10	15.87	02	20
Incontinent	05	7.93	01	10

Discussion:

The exact incidence of Hirschsprung's disease for various reasson cannot be calculated in our unit. The incidence reported in the literature is 1/5000 live birth[4].

Age at the time of diagnosis has an important bearing on the prognosis of the disease, with delay in diagnosis , the chances of HAEC are increased many fold[5]. The patients of Hirschsprung's disease may present at any age. Majority of our patients presented in the neonatal age with acute intestinal obstruction, which is rather a less uncommon presentation of the disease. In this series two patients presented at the age of 11 and 13 years. There is on average a lapse of one year between the onset of symptoms and presentation in hospital. Usually the parents seek treatment from other sources during this period.

Patients usually present with chronic constipation, abdominal distension and failure to thrive. In this series loss of appetite and failure to thrive were encountered in 24.83% of patients. Acute intestinal obstruction was predominantly seen in patients below one month of age . History of delayed passage of meconium was present in 53% of the cases as opposed to 86 to 94 % of the cases in contemporary reports.[6,7]

The overall male to female ratio has been generally reported as ranging from 3:1 to 5:1[1,8]. In our series the ratio was comparable to other series. The incidence of family history is generally reported to be 3.2 to7%[4,5,19] in deferent series The familial incidence was not prominent in this series.

Majority of parents were first cousins as is the prevalent custom in Pakistan... Schiller et al (1990) reported a very high incidence of long segment disease (95%) and familial Hirschsprung's disease(27%) in the offspring of consanguineous parents[9]. In this series no increased incidence of long segment or familial Hirschsprung's disease was noted in those patients who had related parents. This finding correlates well with that of Ahmad[10].

It is reported that 5-21% of the patients of Hirschsprung's disease suffer from associated anomalies[11]. In our series 13 (8.96%) had associated anomalies. Down's syndrome and umbilical hernia were dominant associated anomalies seen in our patients. Maternal age does not appear to be a causative factor in this disease. It is also reflected in our series, as majority of the children were born to mothers between 20--30 years of age.

The order of birth became important after the work of Ryan et al[12]. In our series rarely the patients were first

born. This fact may have some significance regarding some immunologically mediated mechanism in the etiology of this disease[12].

Majority of the patient(70%) in our series were breast fed. There is some evidence that breast feeding protects against HAEC[13], probably accounting for low incidence of preoperative enterocolitis in our patients.

In early part of study we usually performed transverse loop colostomies but due to high incidence of fecoloma formation and prolapse of distal loop, we shifted to divided pelvic colostomy. Colostomy is performed at the distal most ganglionic bowel to preserve the maximum length of normal colon.

Short segment disease has the highest incidence in our series [77.27%), this is comparable to the reports in the literature, which is 64 to -80% in different series[1,5]. Total colonic aganglionosis with or without ileal involvement was seen in 9 (6.81%) cases, which is slightly higher than the reported figures[5,11].

There are three commonly used definitive surgical procedures with many modifications for Hirschsprung's disease .The basic aim is to bring the healthy normally ganglionic colon down to the anus rectum just above the dentate line.

These are:

1. Swenson's rectsigmoidectomy with coloanal anastomosis described by Swenson in 1948[14].
2. :Duhamel's retrorectal pullthrough described in 1964[15]. Martin modified the procedure by performing both upper and lower anastomosis to eliminate the spur and blind rectal pouch[16].
3. The Endorectal pullthrough was first described by Soave in 1964[17]..In original Soave's procedure normally ganglionic colon is pulled through the seromuscular tube after mucosa has been stripped from the rectum. A 5 to 10 cm colonic stump is left out of the anus which is resected later on. Boley modified the procedure by performing primary anastomosis at the dentate line thus avoiding the third stage[18].

Swenson believed that the results of definitive surgery depend more on the personal experience rather than the actual procedure.

"Resection of aganglionic colon .....is a difficult operation. Yet if a well trained surgeon has an opportunity to observe the technical details of the operation and then perseveres, good results can be obtained.[20]"

There are however inherant weaknessess and strengths of different procedures. Duhamel's procedure and ERPT need either minimal or no pelvic dissection respectively as opposed to Swenson's procedure which needs extensive pelvic dissection and can result in damage to pelvic nerves. Duhamel's and ERPT are widely accepted and practiced throughout the world.

Conclusions.

We have noted that the patients acceptability of Duhamel's procedure is superior as we perform the operation in two rather than three stages, the definite procedure beiing performed under caecostomy cover rather than a covering colostomy, which does not need formal closure. Moreover continanace is achieved almost instantaneously in most patients. There are no complication of anal stenosis and costriction ring. In our opinion Duhamel is superior method of treatment of Hirschssprung's disease.

References

1. Ikeda K; Goto S : Diagnosis and treatment of Hirschsprung's disease in Japan. An analysis of 1628 patients Ann. Surg. 1984; 199(4): 400-405.
2. Soper T; Figueroa PR : Surgical treatment of Hirschsprung's disease. Comparison of modifications of the Duhamel and Soave operations. J. Pediatr. Surg. 1971; 6(6): 761
3. Templeton JM; Diteshein JA : Quantitative result of long term fecal continence. J.
4. Pediatr. Surg. 1984; 20: 445-452.
5. Passarge E: The genetics of Hirschsprung's disease. N. Engl. J. Med: 1967:276:138-143.
6. kleinhaus S, Boley SJ; Sheran M; Sieber WK : Hirschsprung's disease. A survey of the members of the surgical section of American academy of pediatrics. J. Pediatr. Surg. 14(5): 588-597.
7. Lister J; Tam PKH : Hirschsprung's disease. In. Lister J; and Irving IM (eds).
8. Neonatal surgery, 3rd Ed. London Boston Sydney Butterworth and Co. (publisher)
9. Ltd. 1990; 523-546.
10. Swensen O, Raffensperger JG: Hirschsprung's disease. In Raffensperger JG:(ed) Swenson's Pediatric Surgery; 5th Edition. Norwalk connecticut. Appleton Lange,1990:555-577.
11. Swensen O, Sherman J; Fisher JH: Diagnosis of congenital megacolon: An analysis of 501 patients. J. pediatr. Surg. 1973: 8: 587.
12. Schiller M; Levy P; Shawa RA; Abu-Dau K; Gorenstein A; Katz S : Familial Hirschsprung's disease. A report of 22 affected siblings in four families. J. Pediatr. Surg. 1990; 25 (3): 322-325.
13. Ahmad S: A study on Hirschsprung's disease: MS Thesis; Jinnah Post Graduate medical centre, Karachi: 1989:109.
14. Sieber WK: Hirschsprung's disease: In Randolph SG; Ravitch MM; O Neil JA; and Rowe MI; (ED) Pediatric Surgery , 4th edition. 1986:995-1020.
15. Ryan ET; Ecker JL; Christakis NA; Folkman J : Hirschsprung's disease. Associated abnormalities and demography. J. Pediatr. Surg. 1992; 27(1): 76-81.
16. Harrison MW; Detiz DM; Campbell JR; Campbell TJ : Diagnosis and management of Hirschsprung's disease. A 25 year perspective. Am. J. Surg. 1986; 152: 49-56.
17. Swenson O : My early experience with Hirschsprung's disease. J. Pediatr. Surg. 1989; 24(8): 839-845.
18. Duhamel B : Retrorectal and transanal pull through procedure for the treatment of

19. Hirschsprung's disease. Dis. Colon. Rectum. 1964; 7: 455-459.
20. Martin L.W: Caudill Dr. A method for elimination of the blind rectal pouch in the
21. Duhamel's operation for Hirschsprung's disease. Surgery 1967; 62: 951-953.
22. Soave F : Hirschsprung's disease. A new surgical technique. Arch. Dis. Childh. 1964; 116-124.
23. Boley SJ : An endorectal pull through operation with primary anastomosis for
24. Hirschsprung's disease. Surg. Gynaecol. Obstet. 1968; 98: 353-357.
25. Stannard VA; Fowler C; Robinson L; et al : Familial Hirschsprung's disease. Report of autosomal dominant and probable recessive x-linked kindrds. J. Pediatr. Surg. 1991; 591-594.
26. Swenson O; Sherman JO; Fisher JH; Cohan E: The treatment and Post operative complications of congenital Megacolon: Ann. Surg: 1975: 182:3:266-273.