



HIRSCHSPRUNG'S DISEASE; DIAGNOSIS AND MANAGEMENT: EXPERIENCE AT IBN-E-SIENA AND NISHTAR HOSPITAL, MULTAN.

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ABSTRACT... Objective: To determine the frequency of Hirschsprung's Disease as a cause of neonatal intestinal obstruction, to compare the results of treatment with others and to suggest means and ways to improve the deficiencies in our circumstances. **Place and duration of study:** Nishtar hospital and Ibn-e-Siena Hospital, Multan from January, 2008 to June, 2012. **Methodology:** A total number of 115 patients from neonatal age to more than 5 years were diagnosed and treated for Hirschsprung's Disease. **Results:** Hirschsprung's Disease was a cause of neonatal obstruction in 26 (22.6%) patients. Hirschsprung's Disease was a cause of recurrent diarrhea in 37 (32.17%) patients. 76 (66.08%) patients presented with chronic constipation. 27 patients underwent for pull-through operation. **Conclusions:** The overall complications rate and stooling pattern after Soave's procedure are not significantly different from others.

Key words: Hirschsprung's Disease, Soave's procedure, Intestinal obstruction.

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INTRODUCTION

Congenital megacolon still commonly known as Hirschsprung's Disease was named after the name of Dr. Harold Hirschsprung, who first gave the description of the disease. He thought that the colonic dilatation was the cause of the problem, although the actual pathology was in the distal contracted segment. Hirschsprung's Disease has many modes of presentation varying from complete intestinal obstruction to chronic constipation with the classical picture of pot belly, poor health and emaciation¹.

Many workers have contributed towards its etiology, pathology, epidemiology, methods of investigations and finally different methods of therapy². The etiology of this disease is still unknown. The pathological picture is well established i.e. the disease is due to the absence or lack of ganglion cells leading to persistent spasm of aganglionic segment which fails to allow contents to flow freely, resulting in massive

dilatation of the proximal segment³.

Subsequently secondary changes of hypertrophy of muscles, oedema and ulceration of mucosa and at times lethal enterocolitis ensue⁴.

Diagnostic aids include barium enema on unprepared bowel, suction biopsy for acetyl cholinesterase staining. Anorectal manometry; as a preliminary screening test also has a place⁵. However the diagnosis has to be confirmed in all suspected cases with the H/E staining of rectal muscle biopsy. Treatment depends on the mode of presentation, age and condition of the patient. Briefly the children with chronic constipation give adequate time to follow a predefined protocol being practiced in paediatric surgical unit but neonatal obstruction may not allow the preliminary diagnostic procedures to be performed routinely and the diagnosis is established at exploratory laparotomy with multiple serial biopsies.

Various surgical procedures like Swenson's, Soave's, Duhamel's and their modifications have their own merits and demerits⁶. The fact is that this treatable congenital condition should have more attention of family physicians, paediatricians and surgeons. A high index of suspicion should be maintained in all cases of neonatal intestinal obstruction and chronic constipation^{7,40}.

METHODOLOGY

A total number of 115 patients with symptoms resembling Hirschsprung's Disease were admitted to the department of paediatric surgery Nishtar Hospital and Ibn-e-Sina Hospital, Multan during the period of January 2008 to June 2012. The patients were diagnosed and managed with the sequence i.e. history and clinical examination, plain x-ray abdomen, barium enema, rectal biopsy and internal sphincterotomy, laparotomy and serial biopsy and colostomy and finally the pull-through operation. In certain number of neonates diagnosis was established after laparotomy, when operated in emergency for neonatal intestinal obstruction.

RESULTS

A total number of 115 patients aged from neonates to 5 years and above were admitted and treated (Table 1). 26 neonates with Hirschsprung's Disease presented with intestinal obstruction. In others 88 patients of varying ages rectal biopsy and internal sphincterotomy was performed. There was one mortality after rectal biopsy and internal sphincterotomy. After rectal biopsy and internal sphincterotomy 42 patients were excluded from the study as 20 were relieved after internal sphincterotomy (ultra short segment) and 25 were not of Hirschsprung's Disease as ganglion cells were detected in their rectal biopsies. Among the remaining 42 patients 5 were not willing for colostomy. 63 patients underwent for laparotomy and colostomy/ileostomy. 1 patient expired after laparotomy/colostomy procedure. Among 61 patients 27 underwent for endorectal Soave's pull-through operation and 34 are waiting for surgery. Overall mortality was 5. In this study, 10 patients (8.6 %) had a positive family history. In this study parents gave the history of inadvertant passage of

lose faeces into clothing in 10 patients, out of these in 5 patients it appeared only when gut was massively loaded. Despite severe constipation and distention, abdominal pain and discomfort was either mild or did not occur. Out of 115 cases, 9 (7.8%) patients complained of daily abdominal pain, 15 (13%) patients felt pain occasionally. Majority of cases i.e. 45 patients had abdominal pain only when gut was loaded. In most of the cases i.e. in 52 (45.21%) patients there was history of constant abdominal distension which varied from mild to massive. In other 30 (26%) cases, the distension which was episodic, was reported frequently. Paradoxical diarrhea alternating with constipation was observed in 21 (18.26%) patients. 16 (13.91%) patients gave history of occasional diarrhea.

Age	No. of patients
Less than one month	38
Less than one year	49
2 to 5 years	22
Above 5 years	06

Table-I. Age at the time of diagnosis.

DISCUSSION

Congenital megacolon is still popularly known as Hirschsprung's Disease after the name of Danish Paediatrician, who was the first to give the description of the disease. No doubt the detailed discussion of this clinical entity is due to the contribution of Heral Hirschsprung, professor of paediatrics in Copen Hegen University. Ever since many workers from all over the world have contributed towards better understanding of various aspects of the disease including its etiology, pathology, role of enteric nervous system, neural control of gut motility, histoanatomy, histochemistry, diagnostic procedures and the best form of treatment.

Extent of the lesion may vary from low rectum to the entire colon extending to small intestine and even to duodenum as in total aganglionosis of the gastrointestinal tract which is the most extreme

and rare form of Hirschsprung's Disease^{8,9}. Histochemically it has been confirmed that along with the absence of the ganglion cells in the distal contracted segment, there is increase of acetyl cholinesterase in the cholinergic nerve fibres of aganglionic segments^{10,37}. Recent studies on the peptinergic innervation of the affected intestinal segment have demonstrated a marked reduction in the density of nerve fibres storing vasoactive intestinal peptide (VIP), substance P (SP), Enkephalin and gastrin releasing peptide (GRP)¹¹. The exact etiology of Hirschsprung's Disease is unknown. The positive factors may be vascular, genetic, viral, maternal hyperthermia and micro environmental changes¹². As far as the genetic aspect is concerned, sufficient evidences have been accumulated in favor of the role of genetic factors in the etiology of congenital aganglionosis. Autosomal recessive inheritance is considered to be possible in some families, it is suggested that aganglionosis may be inherited as X-linked recessive or transmitted on a pattern of autosomal inheritance^{13,14,15}. The characteristic features of gross pathology is the proximal dilatation of the colon with contracted effected segment. Narrowing of the effected segment is the result of the intrinsic inhibitory innervations. Dilatation proceeds gradually proximally. Length of the contracted segment varies according to the extent of aganglionosis^{3,8,16}. Table II shows the extent of lesion as seen in the present study.

G-cells negative	No. of patients
Rectum	26
Rectosigmoid	23
pelvic colon	07
Desc. colon	02
Trans. Colon	02
Total Colon	03
Total	63

Table-II. Extent of Lesion

In the present study, there were 90 male and 24 female patients making Male to female ratio to be

5:1. This is comparable with Suita's¹⁷ study, which reported male to female ratio to be 3:1. In the present study, 10 patients had positive family history. 38 (33%) patients were under the age of one month and 39 (34%) patients were under the age of one year. Although another 88 patients had onset of symptoms in the neonatal age. 26 (22.6%) patients presented as neonatal intestinal obstruction. The remaining 89 patients had the history of recurrent constipation being relieved by laxatives or enemas. A detailed history and careful physical examination can elicit the clinical diagnosis. A number of diagnostic procedures are available to establish the diagnosis e.g. radiological, histochemical, and manometric^{18,19,20,24}. Neonates and infants who present with incomplete intestinal obstruction, abdominal X-rays are taken from Lateral anterior-posterior and up-right positions. Colonic distension with absence of air is the indicative of congenital megacolon. Plain abdominal X-ray was suggestive in 33 patients with massive faecal loading and colonic dilatation. However in 70 (65%) patients it showed multiple gas filled loops. This preliminary investigation gives some idea of the underlined pathology, which has to be confirmed on barium enema^{21,22,25,26}. Barium enema was done in 100 patients. In 26 patients it was strongly positive with well defined transitional zone and in 56 patients it was only suggestive of Hirschsprung's disease. The results are comparable with Klein et al²³. The accuracy of barium enema as a diagnostic tool is reported to the 63% by Hussain et al¹⁶. Ideally all the patients should have mucosal suction biopsy with acetyl cholinesterase staining of the nerve fibres²⁷. In our institution we are not fortunated in this regard and do not have this facility. Therefore we rely mostly on full thickness rectal muscle biopsy with H/E staining²⁸. We take a longitudinal strip of rectal muscle of variable length, depending on the age. This not only helps in the diagnosis of congenital aganglionosis but it also relieves the symptoms of chronic constipation particularly in ultra short segment disease. Out of 88 patients who underwent rectal biopsy and internal sphincterotomy, in 20 patients constipation was relieved. The results are comparable with

Neilson²⁹, who strongly recommended internal sphincterotomy as the sole treatment for ultrashort segment disease.

Serial biopsies at laparotomy with frozen section to determine the extent of lesion is the prerequisite for diversion in the developed countries. Unfortunately we do not have the facilities of frozen section. Therefore we limit ourselves to visual impression of contracted and the beginning of dilated segment and take full thickness muscle biopsy for histopathological staining from various points. The results of extent of lesion are given in Table II. Generally it is agreed that the interval between colostomy and definite surgery should be six months to one year or when the infant gains weight up to 10 Kg. however the interval depends on the age at presentation for colostomy. The recent trend to perform definite surgery i.e. pull-through at an early age and even in the neonatal period^{30,33,34}. In our circumstances the problems associated with the early pull-through operations without colostomy are: in adequate diagnostic facilities in our hospitals and the cases usually presented late. So we still remain very conservative to perform two stage pull-through operations as many of the patients are above the neonatal age and pre-operatively the colon has been dilated. We still prefer the two stage pull-through operation considering certain safeguards to adopt; 1. Confirmation of pathology while performing colostomy and absolutely be certain that normal ganglion cells are there at the level of dilated gut, 2. The patients who underwent for two stage operation can be well monitored for weight gain and adequate decompression prior to definite surgery. In this Study 27 Soave's pull-through procedures were performed. In all 27 cases, colostomy was resected with the aganglionic bowel at the time of definite surgery. The idea to sacrifice some portion of ganglionic gut and to pull down the colostomy in one stage was to save the patients from another surgical procedure. Moreover it was also adopted to decrease the hospital stay and financial burden on parents. In this study two types of post operative complications i.e. early and late were noted. Anastomotic leak represents a real hazard, this

was encountered in one (3.7%) patient. Faecal fistula and perirectal abscess developed in early post-operative period in one (3.7%) case. There was retraction of stump in three (11.11%) patients. One (3.7%) patient wound dehiscence requiring secondary suturing, the same patient developed incisional hernia later on. As far as the late complications are concerned, two patients (7.4%) were readmitted with intestinal obstruction after pull-through operation. In one (3.7%) multiple adhesions and gangrene of terminal ileum was found. Resection and anastomosis was done. The second case was a female patient who was admitted one year after pull through with intestinal obstruction and internal hernia was found. It was reduced, the rent was repaired and the patient made uneventful recovery.

Anastomotic stricture is often considered a late major complication of pull-through operation³¹. In this study, 4 patients developed anastomotic stenosis. This happened mostly due to poor compliance of post operative dilatation. Mothers were advised to dilate the anal canal with dilator at home until the anocolic anostomosis remained at satisfactory caliber. The mothers were advised to report for regular follow up. Failure to follow the instructions resulted in the above complication. The late temporary soiling is an important problem³² and was observed in one patient after pull through operation. All these complications are shown in Table III.

It is difficult to compare this study to those Duhamel and Swenson's procedure with their modifications as all of the cases in this study underwent for endorectal pull-through. However the results were compared with different studies. Comparing the results of data collected by Chumpitazu, Coran and Protor^{26,35,36}, the overall incidence of complications observed in our study is somewhat less. The overall complication rate and one year follow up, the results are not significantly different from others. Looking at the bad results of different centres adopting different procedurs, Gunra and Coran^{36,37} believed that "these are more often the consequences of technical errors than inherent fault in the methods

Complications	No. of patients
Early:	
Paracolic Abscess	01
Wound Dehiscence	01
Retraction of stump	03
Fecal fistula	01
Bleeding stump	01
Anastomotic leak	01
Late:	
Constriction ring	04
Intestinal obstruction	02
Soiling	01
Diarrhea	02

Table-III. Complications after pull-through operation (early and late), total pull-through operations = 27.

chosen.” The following facts are evident from the present study: 1- Hirschsprung's Disease was a cause of neonatal intestinal obstruction in 26 cases. Similar observations are documented in other reports where the frequency of Hirschsprung's disease as a cause of neonatal intestinal obstruction is relatively high as compared to this study^{38,39,40}. The reason behind this might be the fact that in our circumstances all the cases of neonatal intestinal obstruction are not brought and treated in paediatric surgical units. A significant number of these patients are treated elsewhere, 2- Hirschsprung's Disease was a cause of recurrent diarrhea in 32.17% of cases, 3- 76 (66.8%) patients presented with chronic constipation of variable severity as judged by the tie interval between stools. The stooling pattern was observed as stool was formed in 15 patients, loose in 8 cases and constipation in 4 cases. As for the Frequency of defecation after pull-through operation was noted as 15 patients were passing one or two bowel movements per 24 hours. 8 patients passing three bowels per 24 hours while 4 patients were observed for passing four bowel movements in 24 hours. Looking at the results of

our post-operative study, we suggest that: 1- A strong suspicion of aganglionosis should be made in the neonates presenting with abdominal distension, 2- the delayed passage of meconium is lightly to have Hirschsprung's Disease, 3- infants and children with the history of recurrent diarrhea and chronic constipation may also need to be investigated in order to rule out Hirschsprung's Disease, 4- the public general practitioners, paediatricians and general surgeons should be made conscious of this disease. So the goal of treating a child with Hirschsprung's disease should be to attain normal anorectal functions as near to normal as possible. After one year follow up all the patients after definite surgery attain the normal anorectal functions. Stooling pattern and stooling frequency was noted to assess the surgical results. In our circumstances, endorectal pull-through is a procedure of choice. The overall complication rate and long term stooling patterns are not significantly different from the other studies.

CONCLUSIONS

This study implicates that in our circumstances, the use of classical endorectal pull-through conventionally approached with the placement of decompressive colostomy in all age groups resulted in almost equivalent early/late complications and stooling pattern as compared to other studies.

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PREVIOUS RELATED STUDY

Muhammad Zafar Iqbal, Muhammad Jahangir, Muhammad Anwar, Hammad Azam, Muhammad Tahir, Shumaila Irum. HIRSCHSPRUNG'S DISEASE; MODIFIED DUHAMEL (MARTIN MODIFICATION), A PROCEDURE OF CHOICE (A STUDY AT SHEIKH ZAYED HOSPITAL RAHIM YAR KHAN) (Original) Prof Med Jour 17(2) 223-231 Apr, May, Jun 2010.

*“If you can dream it,
you can do it.”*

Walt Disney