Original Article

Clinical manifestations of Hirschsprung's disease: A 6-year course review on admitted patients in Guilan, north Province of Iran

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Background: Hirschsprung's disease (H.D) is a congenital disease in which intestinal ganglionic cells are absent and can cause intestinal obstruction. The disease has various clinical manifestations and different lengths of bowel may be involved. Our aim was to study Hirschsprung's clinical presentations and its rate of intestinal involvement in hospitalized patients in a 6-year course study in Guilan, north Province of Iran.
Methods: We studied the patients with Hirschsprung's referred to Guilan surgery referral center, Poursina Hospital, from 1995 to 2001. In this cross sectional descriptive- analytic study all data were collected from patients' files and questionnaires including demographic data, clinical presentations, diagnostic methods, involved segments, surgical procedure techniques, surgical complications
Results: We detected 58 cases of HD during these 6 years, who underwent surgery. 19 patients were female and 39 were male with age range from one day to 18 years old. Clinical findings were variable such as: constipation, abdominal distension, failure to pass mecunium, diarrhea and other less common manifestations. We found 3 different pathological types: rectosigmoid, ultra short-segment and total–colonic involved segment. There wasn't any difference between presentations of disease in these 3 groups. Surgical procedures which were performed were: Swenson, Soave–Boley and Lynn. Early complications, which occurred during the first month after operation, contain anastomotic leakage, wound infection, hemorrhage and stenosis of anastomosis. Late complications, occurring at least one month after operation, comprise long-term constipation, enterocolitis, incontinence, obstruction due to adhesion. We didn't find any significant difference between the complications of these surgical procedures and any accompanying anomalies except 3 ones.
Conclusions: most of patients had a chronic course of constipation and abdominal distension. without any difference between types of involvements and age and clinical presentations. There are few associated anomalies in the patients. In some cases, surgical complications were less than those in other studies; so, it seems that the performed procedures had fewer complications or they may have been performed in appropriate time.

Keywords: Hirschsprung's disease; Abdominal distension; Anomalies

Introduction

Hirschsprung's Disease (HD) or aganglionic megacolon is a congenital disorder characterized by absence of enteric ganglia along a variable length of the intestine, resulting in a functional obstruction[1]. Most cases of HD are diagnosed in infancy, although some may not be diagnosed until adolescence or early adulthood. There is a familial background for it that seems hereditary more often in mothers[2]. Incidence of HD ranges from 1 per 4400 to 1 per 7000 live births[3]. There is a male predominance of about 4 to 1 in HD, 4 boys versus each girl[4]. The disease has no race predilection and is a

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relatively common problem in gastroenterology and pediatric surgery departments.

The main symptom of HD is constipation, which cannot be treated using laxatives or softeners. Clinical features of the disease and even its diagnostic approaches and treatment are various. Some cases of the disease have been seen with the associated anomalies.[5] that account for 25% of familial cases and 10% of non-familial cases[2]. H.D can be life-threatening or a chronic disorder.

Only when the disease has been early diagnosed, these patients will be referred to the specialized surgery units and undergo a proper treatment trend. Because of different anatomic forms and manifestations of HD, a large number of the patients are misdiagnosed and mistreated even for years and their proper treatment are delayed. HD can involve different length of intestine from a few centimeters of distal rectum to entire colon and even a part of the terminal ileum, and clinical manifestations can vary from a mild or severe constipation in different ages to failure to pass meconium stool at 24-48 hours at birth and prominent distension, even perforation of proximal colon and peritonitis during the first few days after birth. On the other hand, it appears that Iranian researchers have not greatly focused on this disease. In a study in Iran, high percentage of patients with HD presented with constipation and abdominal distension; incontinence and peritonitis were the least common symptoms. Among these patients, different anomalies such as G6PD deficiency, congenital heart disease, microcephaly, and hypostasis were observed[5]. In other parts of the world, in a study by Bonomo *et al*., it was stated that HD is usually diagnosed during the neonatal period. In this study, the diagnostic prevalence of HD in patients was studied with natural barium enema as one of the diagnostic procedures for this disease and by using two other diagnostic procedures (manometry and rectal biopsy)[6]. In a study by Larini *et al*., the major postoperative complications were known related to different extents of dysganglionosis and neuronal bowel dysplasia[7]. In other study in 2000, next effective treatment of the disease has been attributed to the histology of proximal segment[8]. Also, in a study in 2001, two surgical treatments of Duhamel and Swenson procedures were compared[9].

We have conducted the study to discover the various aspects of H.D and the relations between the length of bowel involvement and different types of clinical manifestations in patients with HD presenting to Poursina Hospital, one of the important referral centers of surgical treatment in the northern Iran, during a six-year period.

Materials and Methods

Study samples were patients presenting to specialized referral medical center of Poursina hospital in Guilan, the north province of Iran, between 1995 and 2001. The patients were followed-up with the diagnosis of H.D. In this cross-sectional descriptive-analytical study, the data were extracted from the patients' medical records. Collected data were classified in tables. The data consisted of age, sex, disease manifestations, diagnostic procedures leading to the definitive diagnosis, affected segment, the type of surgery performed, the complications of the surgery performed, and the

Table 1: Distribution of age and sex in studied patients

Sex	Male		Female	
Age	**Frequency**	**Percent**	**Frequency**	**Percent**
<48 hr. after the birth	5	8.6	0	0
> 48 hr and < 1 month	5	10.3	2	3.5
> 1 month and <6 years	26	44.8	14	24.1
> 6 years	2	3.5	3	5.2
Total	39	67.2	19	32.8

outcomes of the treatment. Ultimately, data were analyzed with statistical software of SPSS /10. For statistical analysis of qualitative data, the Chi Square and Fisher Exact tests were used. P value less than 0.05 was considered significant.

Information was obtained from the hospital records and the patients' files available in the private centers following up the patients like the physician's offices. In order to study the postoperative complications, there was a limitation for patients whose follow up periods were not enough yet to study late postoperative complications. Thus, it is likely that the surgery complications were underreported in this study.

Results

In this study, 58 hospitalized patients with HD were enrolled. Of these, 19 patients (33%) were females and 39 males (67%) (Sex ratio of 1:2). The majority of patients (N=40) were classified in the age group of 1-6 years (Table 1) with age range of 1 day to 18 years .With respect to symptoms and clinical manifestations, the most prominent symptoms were

Table 2: Distribution of clinical signs and symptoms in studied patients

Signs and symptoms	Frequency	Percent
Delay of meconium defecation	10	17.3
Diarrhea and enterocolitis	3	5.2
Chronic constipation	46	79.3
Abdominal distension	39	67.3
Perforation of diverticolitis	1	1.7
Soiling	2	2.5
Over-flow-incontinence	1	1.7
Prolapse	1	1.7
Imperforated Anus	1	1.7
Vomiting	5	8.6

Table 3: Distribution of different types of involved segment

Type of involved segment	Frequency	Percent
Rectosigmoid	49	84.5
Ultra-short-segment	3	5.2
Total-colonic	5	1.7
Uncertain	1	8.6
Total	58	100

Without any different types of clinical signs and symptoms, no statistically significant difference was found among the different age groups and between the two sexes (P>0.41, p>0.24 respectively). Other symptoms of the disease-included failure to pass a meconium stool, constipation, diarrhea, vomiting, overflow incontinence and some symptoms with less prevalence (Table 2). Diagnosis for all patients (N=51) was based on rectal biopsy and 38 individuals had a prior barium enema and one person had undergone manometry of rectum and colon. The length of the involved segment was divided to three types; recto-sigmoid segment, very short segment and total colon, and one person were placed in the uncertain segment group due to the sporadic involvement site (Table 3). Clinical manifestations of the patients didn't show any difference in respect of type and the time of presentation. Surgeries were performed in Swenson [abdomino perineal pull-through],[10] Soave-Boley [endorectal pull-trough],[10] and Lynn [posterior anorectal myectomy to correct short-segment agangliosis][10] procedures that in any groups, there were 44, 9 and 5 patients ,respectively and all Lynn surgical operations were related to the patients with the involvement of ultra short segment. Early complications (within one month after the surgery) and late complications (after one month postoperatively) are observed in tables 4 and 5. Concerning the incidence of post-operative complications, no statistically significant difference was seen (p>0.05). No associated anomalies were observed in these patients, for instance, Down syndrome, major cardio-vascular diseases and urogenital abnormalities. Associated Anomalies with HD in present study are as follows:

In gastro-intestinal (GI) tract, one case of imperforate anus was observed which was improved by surgery. There were two cases of premature neonates with a birth time age lower than 38 weeks. One neonate had polydactyly in hands and feet and other one was with cleft palate .Both newborns were treated by appropriate surgery during this period. Regarding the post-operative complications, one patient died due to the undetected enterocolitis as a post-operative complication after one year. The patient presented with diarrhea and severe dehydration due to delay in refers to medical center.

Table 4: Distribution of surgical early complications

Complications	Frequency	Percent
Leakage of anastomotic fistula	5	10.2
Wound infection	1	2.0
Cuff abscess	1	2.0
Anastomotic stenosis	2	4.1
Bleeding of anastomosis	1	2.0

Table 5: Distribution of surgical late complications

Complications	Frequency	Percent
Incontinence	3	6.12
Constipation	5	10.2
Enterocolitis	4	8.16
Soiling	1	2.04
Adhision Band		

Discussion

As it can be inferred from the results, in our study like other studies, frequency of girls are less than the boys (sex ratio of 1:2). However, according to Lee *et al*. study with long-segment disease, the incidence in females increases. HD is uncommon in premature infants[4] that are similar to our study. HD is manifested mostly within the first weeks of life, and it is diagnosed in those aged 5 years or younger. Occasionally, HD is diagnosed during adulthood[11].

In our study regarding tables 1 and 2, we found out the frequency of patients with ages more than 1 year and even 6 years is relatively high that can be attributable to the presence of only minor symptoms in them, has caused HD to be neglected. So, chronic constipation and abdominal distension should be considered the most prominent symptoms of HD in these ages. Failure to pass meconium stool within 24-48 hours after birth, and other acute symptoms like vomiting and peritonitis due to severe distension of proximal colon and its perforation, or diarrhea and enterocolitis as the severe signs of the disease is not considered in this age group as well. Thus it is not unexpected that cases with more extensive involvement are more likely to be familial[1].

Although we know that the problems a child will experience with HD depend on how much of the intestine has normal innervation, but in our study, there was no statistically correlation between the type of the involved segment in length and the type of clinical manifestations among patients, it can not be seen in other studies, though here this issue should be noted that the sample size of this study is too small to reach this conclusion that “In HD, severe symptoms should not be attributed to the more prolonged involved segment”. On the other hand, the post-operative complications in these three groups of Swenson, Soave–Boley and Lynn procedures were the same. The application of Lynn surgery, however, is only in the cases of ultra short segment that can be due to a low sample size. The same result is mentioned in Harjai paper: there are no important differences with regard to outcome and long-term function in patients treated with these surgical procedures[12].

In a study by Sarioglu (2001), stenosis of anastomosis in Swenson surgery was considerably higher than Duhammel surgery; unfortunately we didn’t have a sample of Duhammel technique in our study. Authors of that article have suggested Duhammel procedure as a safe and easy technique for HD treatment[9].

A review of 229 cases of adult HD in the literature suggested that the Duhamel procedure is the operative choice because of the lower rate of morbidity and better functional outcome.[10] Duhamel’s operation in Ikeda-Sopor’s modification is according to authors’ opinion very effective in treatment of congenital megacolon, being associated with a minimum of complications and is the prerequisite for achieving excellent functional results[13]. The Swenson operation is the most physiological but is a difficult operation and the extensive pelvic dissection leads to trauma to the pelvic nerves. The Duhamel operation requires less pelvic dissection and is safe in infants as well as in adults but its problem is of residual spur. The Soave’s operation is easy to perform with

no pelvic dissection required but the disadvantage is that it needs repeated dilatations[12]. According to other studies, common complications include HD-associated enterocolitis (HAEC) after the Swenson operation, diarrhea and incontinence after the Soave endorectal pull-through procedure and constipation after the Duhamel procedure[11]. Associated anomaly was seen in only 3 cases (5.1%) of the patients. In some scientific resources, associated anomalies of H.D have been estimated between 10%-30%[14] that this ratio was not apparent in the patients of our study. Furthermore, it is different from the associated anomalies in other studies in which H.D is a frequent finding in cases of Down syndrome[1]. Among 179 children first born H.D were less likely. 22% of these children had one or more associated abnormalities involving the neurological, cardiovascular, urogenital, and gastrointestinal systems[15].

4 cases had diarrhea and enterocolitis, and a case led to death .It is said that the diarrhea is seen in one third of children in whom the diagnosis was made at the age before 3 months,[16] and in 12-58% cases with HD, if remained untreated, enterocolitis will be occurred[17-21]. Enterocolitis is the most severe complication of HD that occur either pre or postoperatively,[22] and in our study, it has occurred postoperatively. Primary Laparoscopic Pull through Procedure (PLPP) in infants with HD as a minimally invasive surgery is an effective, problem-free approach for treating neonatal HD. It has replaced the standard two stage open procedure in babies with uncomplicated presentation[23]. Unfortunately, we didn't have any case of PLPP in our study.

We concluded that a great number of patients with HD can present a chronic symptom of abdominal distension and constipation that among them; the affected segment extent has no much effect on the onset and type of symptoms. The common anomalies observed in other studies were not seen in our patients. Thus, the association of this disease with anomalies in our study appears rather less important. Post-operative complications in our patients are less than those in other studies. It is probably because of the performed surgical techniques with lower complications or maybe due to select a suitable time for operations that have been carried out in our patients. At the end, further studies with a larger sample size are needed to assess the relations, discussed in this study.

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