The Mild Form of Hirschsprung's Disease (Short Segment)

Fourteen-Years Experience in Diagnosis and Treatment

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From 1966 through 1980, 39 children were treated with anorectal myectomy for the mild form of Hirschsprung's disease. The mild form of Hirschsprung's disease is differentiated from the severe one mainly on clinical grounds. Of the entire group, only four patients needed a Duhamel procedure, one to three years after myectomy. One patient still uses occasional enema (11 years after operation). The remaining 34 patients are symptom-free (1–14 years after operation). Anorectal myectomy is the treatment of choice in the management of the mild form of Hirschsprung's disease.

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THE MILD FORM OF Hirschsprung's disease, also known as short segment Hirschsprung's disease, was first described by Hurst.[1] This variant of congenital megacolon is better known to pediatricians and surgeons.[2-12] The authors' experience with 11 patients was published in 1969.[13] During the past 14 years, 39 patients were diagnosed and treated on the authors' service.

Materials and Methods

From 1966 through 1980, 39 children were treated with anorectal myectomy for the mild form of Hirschsprung's disease. Four of the children were temporarily relieved by this procedure but later had to undergo a Duhamel operation for what turned out to be a moderate, rather than mild, form of this disease. Of the remaining 35 children, 25 were males and 10 females. The mean age ± was 5.8 ± 0.7 years when the diagnosis was made. A brother and sister were in the group. No other family history was found.

The diagnosis of Hirschsprung's disease was based on clinical, roentgenographic, manometric and histologic studies. The differentiation between the mild and the more severe forms was based on clinical grounds. The clinical findings in the mild form were: constipation, abdominal distention, presence of stool in the rectal ampulla, soiling and late onset of symptoms (Table 1). In the severe form, the rectal ampulla is empty, soiling does not occur, and the onset of symptoms is early, in most patients in the first month of life. Constipation was the main presenting symptom in 34 out of 35 children, lasting from two to 12 days. Soiling occurred in 19 out of the 30 patients who were beyond the age of toilet training. Abdominal distention was found only in ten patients, and stool was present in the rectal ampulla in 29 patients.

In Table 2, the clinical findings in the large group of patients who permanently benefited from anorectal myectomy are compared with those in the four patients who had the moderate form of the disease and later underwent Duhamel procedure. In the latter group there was a higher ratio of male to female, the onset of symptoms was in the neonatal period in all four patients, feces were present in the rectal ampulla in three and abdominal distention was observed in three. The roentgenographic, manometric and histologic data are presented in Table 3.

Barium enema was performed in all patients. Delayed evacuation was found in 22 children, assessed by 24 and 48 hour x-ray examinations of the abdomen. All patients treated after 1970 underwent manometric studies by a simplified method which was developed on our service[14] as a modification of Schuster's technique.[15] Absence of relaxation of the internal sphincter was seen in 21 children and equivocal results in three. Absence of ganglion cells and increased activity of choline-esterase were diagnostic of Hirschsprung's disease. Between the years 1970–1976 the diagnosis was made by manometric and x-ray studies only. From 1976 to date,

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Submitted for publication: March 10, 1981.

0003-4932/81/1200/0767 $00.70 © J. B. Lippincott Company

TABLE 1. *Clinical Findings in Patients with the Mild Form of Hirschsprung's Disease*

No. of Patients	Sex		Mean Age at Operation yrs.	Onset of Neonatal	Symptoms Infancy	Consti-pation	Stool in Rectal Ampulla.	Soiling	Abdominal-Distention
	Male	Female							
35	25	10	5.8 ± 0.7	7	28	34	29	19*	10

* Five patients before the age of toilet training.

mucosa-submucosal biopsy specimens for histologic and choline esterase studies were added. A specimen material, obtained in anorectal myectomy, confirmed the diagnosis in all patients. In 20 of 39 patients treated with anorectal myectomy, ganglion cells were not found in the entire specimen that was removed.

Anorectal myectomy was performed as described in previously[13] following preparation by sulfa, neomycin and lately by erythromycin. The strip of muscle that was removed measured 6-10 cm × 1 cm and included portion of the internal sphincter and both layers of the muscularis. After operation, mineral oil was administered during a week or more, if necessary, until bowel movements were regular. Follow-up study was accomplished by regular visits to the out-patient clinic, by interviews, telephone calls or correspondence. Special attention was paid to bowel habits and the need for laxatives in the immediate and late postoperative periods.

The results were classified into four groups: unsatisfactory—if there was only transient improvement followed by the need for a more extensive procedure; equivocal—if there was a need for occasional enema three years after operation; good—if there was a need for stool softeners and occasional enema not later than one year after the operation and, excellent—if there was an immediate and permanent relief of all symptoms.

Results

Anorectal myectomy was performed in 39 children in whom the initial diagnosis of mild form of Hirschsprung's disease was made. All symptoms were immediately relieved following operation. In one patient, the operation was complicated by retrorectal infected hematoma that was surgically drained. No other immediate postoperative complication occurred. There was one patient in whom a stricture developed, and he responded well to dilatations.

Four patients developed symptoms of moderate form of Hirschsprung's disease and had to undergo a Duhamel procedure one to three years after anorectal myectomy, without undue technical difficulties. One of these four patients, presented in our initial report,[13] in whom the diagnosis of mild Hirschsprung's disease was made at the age of seven months, had an empty rectal ampulla with an extremely poor general condition—all pointing to the diagnosis of rather severe form of Hirschsprung's disease. In spite of this, the patient underwent anorectal myectomy. His condition markedly improved, his weight reached normal level and he was well for all practical purposes. It was evident only at the age of 2½ years that the recurrent symptoms were the manifestation of a moderate form of Hirschsprung's disease requiring a Duhamel procedure. This was done with immediate and permanent relief of all symptoms. The histologic examination showed that the aganglionic segment reached the splenic flexure of the colon.

In Table 4, long-term results and the length of follow-up study in the remaining 35 patients are presented. Remyectomy had to be performed in two children, two and four years after operation. The results of anorectal myectomy were: equivocal in one patient, good in seven and excellent in 27 patients. At present all patients are well and lead normal lives.

TABLE 2. *Clinical Findings in Patients with Mild Form of Hirschsprung's Disease and those who Turned out to be with the Moderate Form*

	No. of Patients	Sex		Onset of Symptoms		Constipation		Stool in Rectal Ampulla		General Condition		Abdominal Distention	
		M	F	N	IC	+	−	+	−	good	poor	+	−
Mild form	35	25	10	7	28	34	1	29	6	35		10	25
Moderate form	4	3	1	4		4		3	1	3	1	3	1

M: male. F: female. N: neonatal period. IC: infancy or childhood.

Discussion

In the mild form of Hirschsprung's disease impairment of motility of the distal alimentary tract extends only over a short segment of the rectum, including the internal sphincter. This explains the presence of stool in the rectal ampulla and the relief of symptoms by anorectal myectomy. It is evident that the length of the aganglionic segment does not correlate with the propulsive capacity of the bowel.[16] The term "short segment" or mild Hirschsprung's disease is, therefore, functional rather than anatomic. In 16 of 35 patients with mild Hirschsprung's disease there was total aganglionosis throughout the myectomy specimen, yet all of the patients responded well to anorectal myectomy. The patient who was first presented as a case of mild Hirschsprung's disease[13] and later needed a Duhamel procedure for aganglionic colon extending to the splenic flexure demonstrates the lack of correlation between the extent of aganglionosis and severity of symptoms. Anorectal myectomy was not only temporarily beneficial in this child but also enabled him to regain his weight and to reach the age of 2½ years with no need for major operation and without a colostomy. Soper describes a patient with total aganglionosis of the colon in whom the diagnosis was made as late as 9½ years of age.[17]

The mild form of Hirschsprung's disease is usually diagnosed after the age of toilet training,[7,12] unlike patients with moderate or severe Hirschsprung's disease, where symptoms appear early in the first year of life. Only seven patients out of 35 presented their symptoms before the age of one year in this series.

The four patients who turned out to have the moderate form of Hirschsprung's disease presented their symptoms in the neonatal period. One of them was in a poor general condition. The early onset of symptoms and poor health are suggestive of the rather severe form of the disease. Three of the four had stool in the rectal ampulla, thus making the differentiation between these borderline cases and patients with mild form of Hirschsprung's disease somewhat difficult. The ratio of male to female in the mild form of Hirschsprung's disease was 25:10, as compared with the male predominance of 9:1, seen in the severe form of Hirschsprung's disease.

TABLE 3. *Roentgenographic, Manometric and Histologic Data in Patients with Mild Form of Hirschsprung's Disease*

Patient	X-ray Evidence	Positive Manometry	Histology Total Aganglionosis	Histology Transitional
1	−	+		+
2	+	ND		+
3	+	+		+
4	−	+		+
5	−	+	+	
6	+	+	+	
7	+	ND		+
8	+	+	+	
9	−	+		+
10	+	+	+	
11	+	+		+
12	+	+		+
13	+	ND	+	
14	+	ND		+
15	−	ND	+	
16	−	ND	+	
17	+	+	+	
18	+	ND		+
19	−	+	+	
20	−	ND		+
21	+	+ −		+
22	+	ND		+
23	+	+	+	
24	+	+	+	
25	+	+ −		+
26	+	+		+
27	−	ND		+
28	+	ND	+	
29	−	+		+
30	−	+		+
31	−	+		+
32	−	+ −	+	
33	+	+	+	
34	+	+	+	
35	+	+	+	
Total 35	+22 −13	+21 + − 3 ND 11	16	19

ND: not done.
+ −: equivocal.

At present, the diagnosis of Hirschsprung's disease is made on our service by anorectal manometry, x-ray studies and examination of a mucosa–submucosal biopsy specimen that includes choline-esterase examination.[18] All of these can be done on an ambulatory basis.

TABLE 4. *Long-term Results and the Need for Remyectomy in Patients with Mild Form of Hirschsprung's Disease Treated with Anorectal Myectomy*

No. of Patients	Follow-up (yrs)	Remyectomy	Results		
			Excellent	Good	Equivocal
35	Mean ± SE: 9.4 ± 0.5 Range: 1–4	2	27	7	1

The risks attendant upon these examinations is small as the patients do not have to undergo general anesthesia or an extensive procedure such as full thickness rectal biopsy. During the same 14 years, 76 children were treated for moderate to severe form of Hirschsprung's disease as compared with 35 treated for the mild form. This high proportion of 76 to 35 is attributed to the readiness of pediatricians to refer children with severe constipation and soiling for investigation and treatment.

Follow-up studies during 14 years showed that almost all children with the mild form of Hirschsprung's disease (34 out of 35), who underwent anorectal myectomy, are, at present, free of symptoms. This and the experience of others[7] justifies the use of the above procedure in all patients with evidence of mild form of Hirschsprung's disease.

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