Surgical Treatment in Familial Visceral Myopathy

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In a kindred with a familial visceral myopathy, seven patients had operations seeking relief of chronic abdominal pain and other symptoms of intestinal obstruction; one patient had an 80% cystectomy and a Y-V-plasty of the bladder neck for urinary retention. Five patients with megaduodenum had bypass operations; a side-to-side duodenojejunostomy was done in four and a retrocolic gastrojejunostomy in one. Two of these died of postoperative complications, and one developed symptomatic adhesions. Two other patients who had duodenojejunostomy have done well for 6 years and 1½ years respectively. One patient with dilation of the distal jejunum and proximal ileum had relief of intestinal obstructuve symptoms from jejunostomy to decompress the distal jejunum. One patient who had a resection of the descending and sigmoid colon for sigmoid volvulus has done well for four years. Three of these seven patients developed peritonitis postoperatively, and two had symptomatic adhesions after operations. Duodenal aspiration from a patient who developed postoperative peritonitis grew *E. coli*, 10^{13} colonies per ml. After review of the results of operations in other families and in our kindred, we favor side-to-side duodenojejunostomy in megaduodenum. Duodenal aspirate must be cultured before operation. Evidence of bacterial overgrowth in the aspirate should prompt appropriate antibiotic treatment to reduce the likelihood of sepsis.

FAMILIAL VISCERAL MYOPATHY is an entity that is characterized by atrophy of smooth muscle throughout the gut and in some organs outside the gastrointestinal tract.[2] The patients with this disorder usually have symptoms of chronic intestinal pseudo-obstruction.[1] Six other families with a similar disorder have been described previously.[4-7,10-11] Schuffler and his colleagues were the first to describe atrophy of smooth muscle in the dilated duodenum of such patients.[9] Various operations have improved obstructive symptoms in several of these patients. The purpose of this study is to review the types of operations that have been used, the postoperative complications and the results of operation in the family with familial visceral myopathy that we have described recently.[2]

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Patients and Methods

The propositii (cases 15 and 16) and their mother (case 11) were admitted to the University of Iowa Hospitals and Clinics in August 1977. Information was obtained from the other family members, from their family doctors and from hospital records. A family tree was constructed (Fig. 1). Of 93 members in five generations of this family, at least 18 members have the disease. The diagnosis was confirmed by radiographic or histologic studies in 10 patients, and the diagnosis was very strongly suggested by the history in another eight family members. Eight of the ten confirmed cases were operated on in several different hospitals. The operative reports, postoperative course, and the follow-up reports were obtained from those local hospitals.

Results

Clinical Manifestations and Pathologic Findings

Table 1 summarizes the clinical manifestations in 14 patients. Clinical information was not available in cases 1, 2, 3, and 7. Recurrent abdominal pain has been the most common complaint. Characteristically, the pain is described as a cramping, located in the epigastrium or right upper quadrant and usually occurring in the postprandial period. Nausea, vomiting, abdominal distention, diarrhea, constipation, urinary retention and heartburn have also occurred, but less commonly. The symptoms have been characteristically intermittent; between attacks most of the patients have felt quite well. Mydriasis has been noticed in three patients. Two patients (cases 5 and 11) have been asymptomatic. Table 2 lists the lesions found in 10 confirmed cases. A typical upper gastrointestinal roentgenogram shows a dilated duodenum with apparent termination of the dilatation just proximal to the ligament of Treitz (Fig. 2). The stomach appears to be

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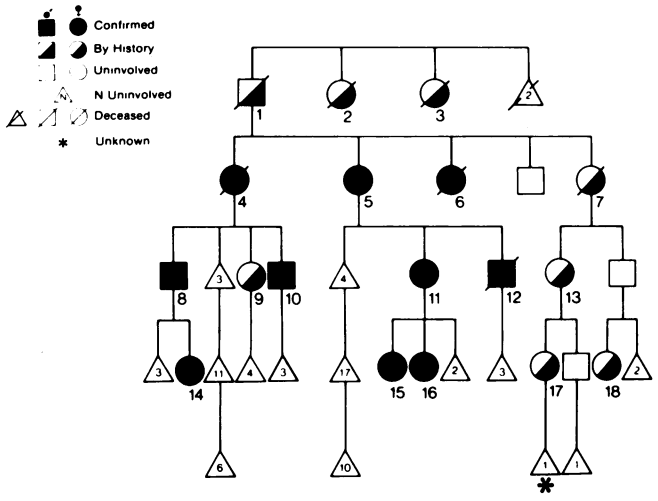

FIG. 1. Family pedigree.

normal. Barium remained in the dilated duodenum of one patient for 14 days (Fig. 3). A barium enema examination in three patients revealed an elongated, redundant colon with loss of haustral markings in two patients and a megacolon in one. Four patients were found to have megacystis on infusion pyelography.

Esophageal manometric studies were performed on three patients and showed abnormality in two. The gastroesophageal sphincter pressure was only 3–5 mmHg (normal 20 mmHg). It relaxed with swallowing, although the relaxation was difficult to see since the resting pressure was so low. There were no contractions in the lower two-thirds of the body of the esophagus, but the upper one-third showed normal contractions with swallowing. The upper esophageal sphincter was normal.

Histological specimens were available for examinations in five patients. These included specimens from the stomach, the dilated duodenum, the junction between duodenum and jejunum, jejunum, ileum, colon and urinary bladder. All of the tissues showed evidence of significant damage to smooth muscle. There was marked attenuation of the muscle and replacement with collagen. In the small intestine, these changes appeared to affect outer longitudinal muscle layer almost exclusively. These changes were most severe in the megaduodenum (Fig. 4), and least severe in the ileum. Electron microscopy demonstrated a large amount of extracellular collagen. Individual smooth muscle cells displayed areas where the plasma membranes appeared to be discontinuous. There appeared to be some degree of mitochondrial swelling, and myofilaments were present in decreased numbers. Ganglion cells were readily demonstrable in all areas, apparently in normal numbers.

TABLE 1. *Clinical Manifestations**

Symptom or Sign	Total
Recurrent postprandial abdominal pain	10
Nausea and vomiting	5
Abdominal distension	3
Diarrhea	3
Constipation	3
Alternating constipation and diarrhea	1
Urinary retention	3
Mydriasis	3
Heartburn	2
Asymptomatic	2

* Clinical data was available on 14 patients.

TABLE 2. *Lesions of Ten Confirmed Cases*

Lesion	Number
Megaduodenum	7/8*
Megacystis	4/5
Megacolon or redundant colon	3/3
Dilated distal jejunum and proximal ileum	1/8
Abnormal esophageal motility	2/3

* Numbers positive over numbers examined.

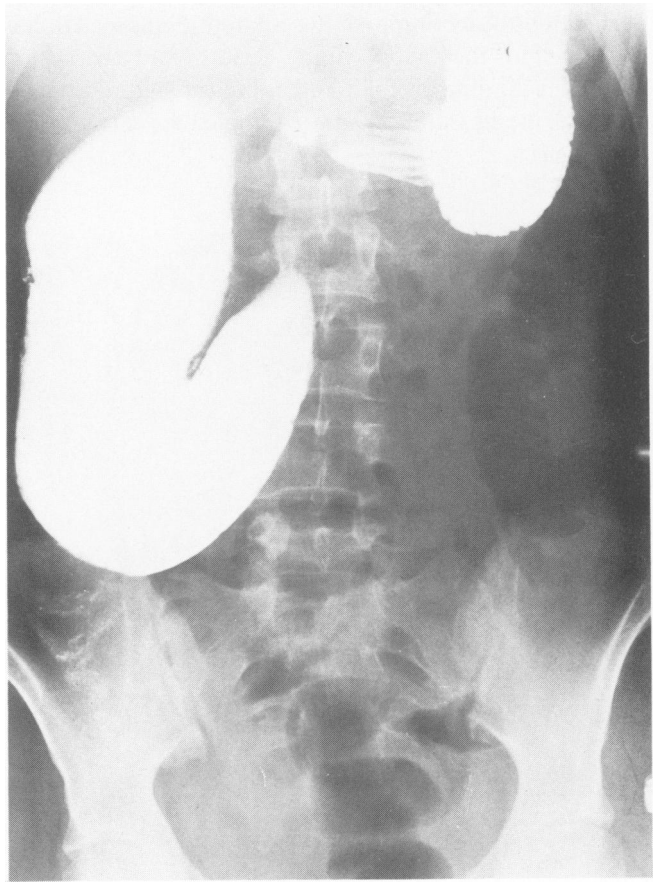

FIG. 2. A typical upper gastrointestinal roentgenogram shows a dilated duodenum (Case 16).

Surgical Treatment, Postoperative Complications and Results of the Operations (Table 3)

Seven patients had operations to relieve chronic abdominal pain and other symptoms of intestinal obstruction. One was operated on for urinary retention. Five patients with megaduodenum had bypass operations, side-to-side duodenojejunostomy in 4 and retrocolic gastrojejunostomy in one. Two patients (Case 4, duodenojejunostomy and Case 6, gastrojejunostomy) died immediately after operation from pulmonary emboli and septic peritonitis. Case 12 (who had duodenojejunostomy) was complicated by symptomatic adhesions, and died after several unsuccessful operations to cut adhesions. Cases 15 and 16 have done well for the past 6 years and 1½ years respectively after side-to-side duodenojejunostomy. Case 16 developed mild peritonitis postoperatively, perhaps as a consequence of peritoneal soiling, for the presence of duodenal bacterial overgrowth (*E. coli*, 10^{13} colonies per ml) was not recognized before operation. Case 10, with dilatation of distal jejunum and proximal ileum, developed peritonitis after an exploratory laparotomy. In the following 4½ months, he had three additional operations to cut adhesions. During the last operation, a Stamm jejunostomy was performed to decompress the dilated jejunum. He has done well for the past 12 years. Case 14 had a resection of the descending and sigmoid colon for sigmoid volvulus, and she has done well for the past 4 years. Case 8

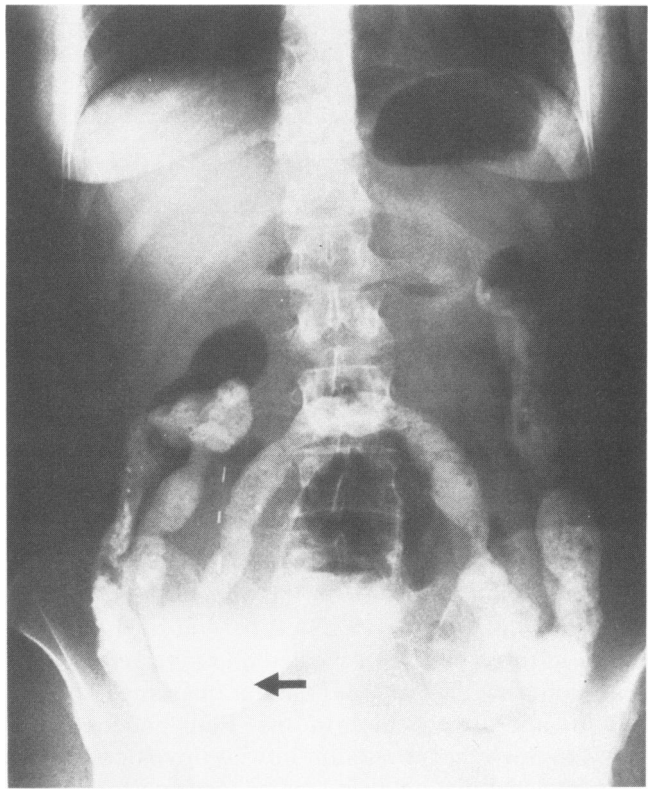

FIG. 3. A plain abdominal x-ray taken 14 days after a contrast upper gastrointestinal roentgenogram. There was still some barium in the dilated duodenum (arrow) and colon (Case 11).

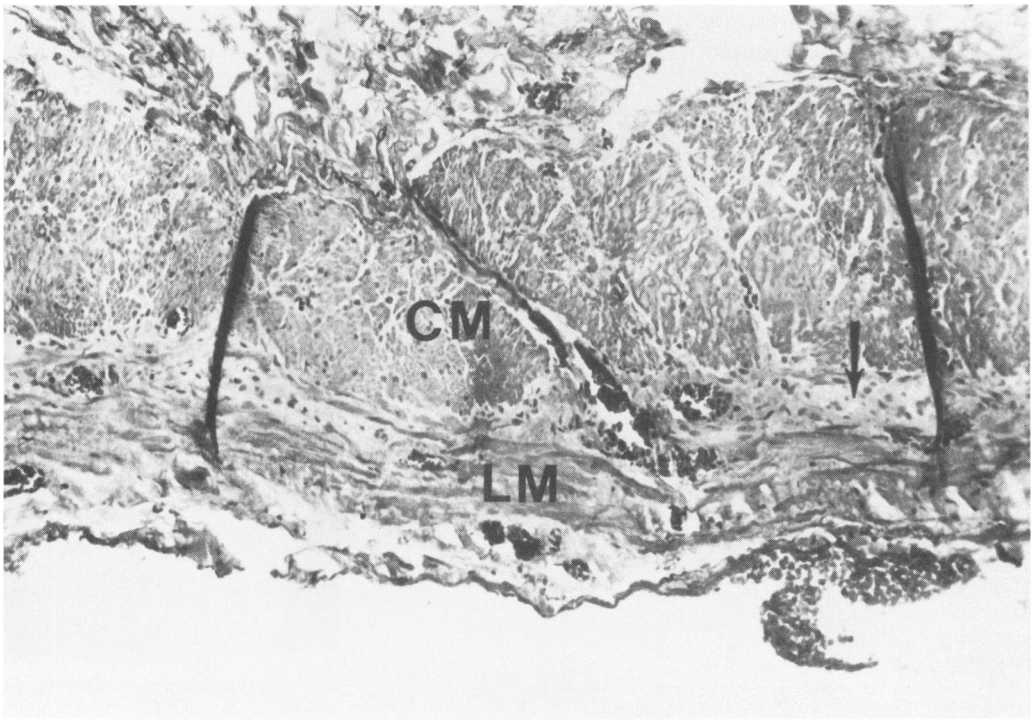

FIG. 4. Duodenal biopsy. The circular muscle layer (CM) is intact, but there is marked attenuation of the longitudinal muscle layer (LM). Ganglion cells (arrow) are present (H&E stained, ×40).

TABLE 3. *Types of Operations and Results in Seven Patients in Kindred with Familial Visceral Myopathy*

Patients	Age at Time of First Operation	Lesion	Indication for Operation	Type of Operation	Results
Case 4, F	47	Megaduodenum	Barium impaction following an UGI X-ray	Side-to-side duodenojejunostomy	Died from pulmonary emboli postoperatively
Case 6, F	72	Megaduodenum	Symptoms of intestinal obstruction	Retrocolic gastrojejunostomy	Died from septic peritonitis and upper gastrointestinal bleeding postoperatively
Case 8, M	51	Megacystis	Urinary retention	80% cystectomy and Y-V plasty of bladder neck.	Has done well for the past 6 years
Case 10, M	18	Dilatation of distal jejunum and proximal ileum	Symptoms of intestinal obstruction after playing football	1st operation: exploratory laparotomy.	Had septic peritonitis postoperatively, followed by severe adhesions.
			Symptoms of intestinal obstruction	2nd operation: lysis of the adhesions (3 weeks after the 1st operation)	No symptomatic improvement
			Symptoms of intestinal obstruction	3rd operation: repeat lysis of the adhesions (7 weeks after the 2nd operation)	No symptomatic improvement
			Symptoms of intestinal obstruction	4th operation: Stamm jejunostomy and lysis of the adhesions (8 weeks after the 3rd operation)	Jejunostomy was closed few months later. The patient has done well for the past 12 years
Case 12, M	35	Megaduodenum	Symptoms of intestinal obstruction	1st operation: Side-to-side duodenojejunostomy	Had done well for 8 months
			Symptoms of intestinal obstruction	2nd operation: Lysis of the adhesions (8 months after the 1st operation)	Had done well for 6 months
			Symptoms of intestinal obstruction	3rd operation: Lysis of the adhesions (7 months after the 2nd operation)	Had done well for 18 months
			Symptoms of intestinal obstruction	4th operation: Lysis of the adhesions and child plication (18 months after the 3rd operation)	No symptomatic improvement
			Symptoms of intestinal obstruction	5th operation: Widening duodenojejunostomy anastomosis and lysis of the adhesions (3 weeks after the 4th operation)	Complicated by pulmonary emboli and upper gastrointestinal bleeding
			Symptoms of intestinal obstruction	6th operation: Lysis of the adhesions (10 weeks after the 5th operation)	The patient had some symptomatic relief but died a year later from malnutrition and pneumonia
Case 14, F	11	Dilatation of descending colon	Sigmoid volvulus	Resection of 37 cm of descending colon	Has done well for the past 4 years
Case 15, F	18	Megaduodenum	Symptoms of intestinal obstruction	Side-to-side duodenojejunostomy	Has done well for the past 6 years
Case 16, F	20	Megaduodenum	Recurrent severe postprandial abdominal pain	Side-to-side duodenojejunostomy	Had mild peritonitis postoperatively. Has done well in the past 1½ years

had an 80% cystectomy and Y-V-plasty of the bladder neck for urinary retention, and he has done well for the past six years. Out of the seven patients who had abdominal surgery, three developed peritonitis postoperatively and two developed symptomatic adhesions.

Discussion

In members of the other families with conditions of this kind, several types of operations have been performed (Table 4). Eight patients in these other families have been described with a total of 12 operations. Three patients (Cases A, C, and D) had duodenojejunostomy; one died postoperatively and the other two had a good result. Three patients (Cases B, F and H) had retrocolic gastrojejunostomy with or without partial gastrectomy; all had poor results. Two patients (Cases F and G) had relief from their symptoms after resection of megaduodenum. One patient (Case H) had relief after partial resection of the anterior duodenal wall and a Roux-en-Y

TABLE 4. *Types of Operations and Results in Other Families*

References	Case	Lesion	Type of Operation	Results
Weiss[11]	A	Megaduodenum	Duodenojejunostomy and ligation of duodenojejunal loop	Had done well for at least 7 years postoperatively
	B	Dilated stomach, duodenum and proximal jejunum	Gastric resection with retrocolic gastrojejunostomy	No relief of symptoms and died less than a year later
	C	Megaduodenum	Duodenojejunostomy	The patient died 17 days postoperatively because of mechanical ileus
Law et al.[4]	D	Megaduodenum	Duodenojejunostomy	Had done well for at least 2 years
	E	Megacystis and urinary retention	Y-V plasty of the bladder neck	Had done well postoperatively
Newton[7]	F	Megaduodenum	1st operation (age 11): mobilization of jejunum at the ligament of Treitz	No improvement
			2nd operation (age 23): partial gastrectomy with retrocolic gastrojejunostomy, and side-to-side duodenojejunostomy using the afferent loop of jejunum	Partial improvement of symptoms
		Bleeding from marginal ulcer	3rd operation (age 31): resection of megaduodenum, and vagotomy	Had done well for at least 6 years postoperatively
	G	Megaduodenum	Duodenal resection, partial gastrectomy and vagotomy	Had done well for at least 3 months postoperatively
Schuffler et al.[9]	H	Megaduodenum	1st operation (age 14): exploratory laparotomy	No improvement
			2nd operation (age 15): retrocolic gastrojejunostomy and Stamm gastrostomy	No improvement
			3rd operation, 2 months after the 2nd operation, partial resection of anterior duodenal wall. Distal end of the afferent loop of gastrojejunostomy was anastomosed end-to-side to the jejunum (Roux-en-Y)	Had done well for at least 23 months postoperatively

anastomosis of the afferent loop of the gastrojejunostomy to the jejunum.

If one considers all the familial syndromes reported, there have been 12 patients who have been operated on for megaduodenum. Seven had side-to-side duodenojejunostomy; two of these died postoperatively from pulmonary emboli and mechanical ileus. Four had relief of the symptoms. One patient developed severe adhesions postoperatively and he died following multiple subsequent operations. Resection of the megaduodenum and Roux-en-Y anastomosis of the Billroth II gastrojejunostomy also gave good results. Side-to-side duodenojejunostomy has been widely used for "superior mesenteric artery syndrome"[3] and "acquired megaduodenum".[8] The opening of the duodenojejunostomy anastomosis has to be big to allow intestinal contents to pass easily to the jejunum.[8]

The high incidence of postoperative complications in our patients is disturbing. Three patients developed peritonitis postoperatively. This may have been due to soiling of duodenal contents at operation.

We conclude that side-to-side duodenojejunostomy is the operation of choice for megaduodenum in familial visceral myopathy. Gastrojejunostomy seems to be less effective. Duodenal aspiration and culture for aerobic and anaerobic bacterial cultures must be performed before operation. Significant bacterial overgrowth should prompt giving of appropriate antibiotics to avoid postoperative peritonitis and the development of adhesions.

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