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NEW CONCEPTS OF THE ETIOLOGY, DIAGNOSIS AND TREATMENT OF CONGENITAL MEGACOLON (HIRSCHSPRUNG'S DISEASE)

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THE clinical syndrome now known as Hirschsprung's disease, or congenital megacolon, was recognized and described over 100 years ago.[1] A careful and complete description of two patients, including their clinical course and postmortem findings, was reported by Hirschsprung in 1886.[2] Since that time numerous publications have been issued regarding this syndrome and while there has been a general agreement in the clinical description and course of this disease, there has been considerable difference of opinion concerning the underlying pathology. Most observers agree that the Hirschsprung disease eponym should be reserved for those patients with hypertrophy and dilatation of the colon without mechanical obstruction. In this limited group of patients with congenital megacolon in which no mechanical obstruction can be demonstrated, the pathogenesis of the dilatation and hypertrophy has been debatable.

A study of 26 patients with congenital megacolon, made during the last five years at The Children's Hospital, has clarified to the authors the nature of this disease and a new treatment based on this concept has been instituted. There are certain clinical features of this disease that should be stressed because they support the authors' concept of the etiology of congenital megacolon. In all but one of the 26 patients, the onset of symptoms occurred in the first few weeks of life. In the remaining patient, the history was obscure as the child was adopted at six months of age and the early background was unknown. However, when the child was six months old, a diagnosis of congenital megacolon was made. Other reports on congenital megacolon have stressed the early onset of the disease. Both patients described by Hirschsprung[2] had symptoms beginning shortly after birth. Barrington-Ward[3] studied 19 patients with congenital megacolon in which the diagnosis was confirmed at autopsy. In 16 of these the inception was at birth. In the remaining three, constipation dated from two months, six months and 3½ years. One patient in this series was operated on during the first week of life. Dilatation and hypertrophy of the colon were found but no mechanical obstruction was present. All of this evidence suggests that the disease is congenital in origin. The marked preponderance of males reported in the literature is also evident in this series. In this group of 26 patients, only four were females.

A careful analysis of the histories and physical examinations of the group of patients reveals the whole clinical picture and course to be consistent with partial obstruction of the large bowel. All of the patients in this group had severe constipation and bouts of obstipation as the presenting complaints. In the periods of exacerbation of the disease,

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the children, particularly the older ones, complained of abdominal cramps and vomiting. Several mothers commented on the fact that audible gurgles could be heard emanating from the child's abdomen. The degree of abdominal distention also varied from time to time. In the children with distention it was possible to see the pattern of the dilated bowel through the thin abdominal wall and to observe peristalsis in the dilated colon. On palpation of the abdomen, large fecal impactions were frequently outlined, most commonly in the left lower quadrant. Despite these impactions in the colon, rectal examination demonstrated an empty ampulla in all patients except two. It is interesting that Hirschsprung[2] commented specifically on the absence of fecal material in the rectum, despite a large impaction in the colon. Thus it can be stated that the histories and physical findings in congenital megacolon are perfectly consistent with partial obstruction of the lower part of the colon, the severity of symptoms and signs being related to the degree of obstruction.

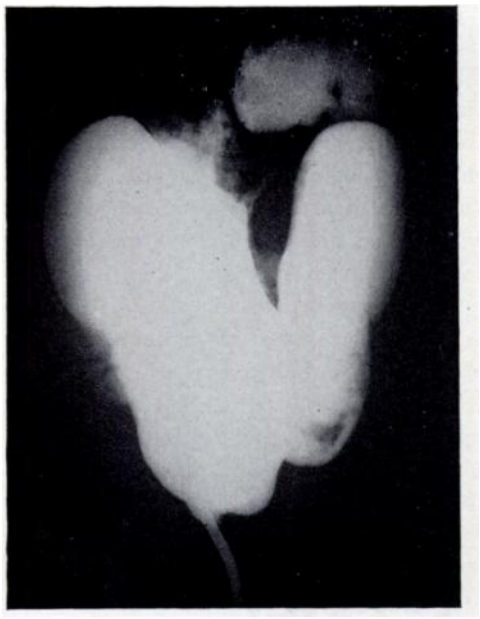

FIG. 1. Routine barium enema. Whole colon has been filled and abnormal area of rectosigmoid is obscured.

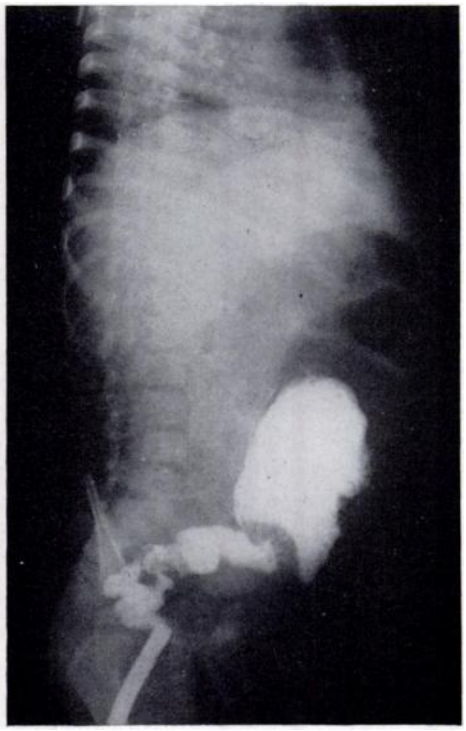

FIG. 2. Barium enema correctly performed which gives detailed outline of rectosigmoid and demonstrates narrowing in colon.

While the clinical picture has been consistent with partial mechanical obstruction, postmortem studies and findings in the operating room have never demonstrated any form of mechanical obstruction. However, it is well known that occasionally the clinical picture of megacolon can be produced by a true organic obstruction, such as a congenital web or stenosis of the sigmoid or anus. All the patients in this series were subjected to fluoroscopic examination of the colon during barium enema. Each presented the picture

of functional obstruction produced by a dyskinesia of the rectum or rectosigmoid and this could usually be detected only when the barium mixture was allowed to run slowly and in small quantities into the rectum. For satisfactory visualization of the rectosigmoid, it is essential to examine the patient in the oblique projection, because if this is not done the appearance produced is that which is so familiar in much of the literature—that of a colon which appears dilated to the anus (Fig. 1).

In these patients, a portion of the rectum or rectosigmoid, often a segment up to 10 cm. in length, was consistently less than normal in caliber. The contour was frequently irregular and often a turbulent and purposeless peristaltic activity could be seen. Occasionally there appeared to be reverse peristalsis, so that the barium mixture was carried cephalad into the sigmoid (Fig. 2). More often no peristaltic activity can be seen.

After this portion of the bowel was satisfactorily visualized, a small amount of barium was allowed to run through this area of abnormal function of the colon into the portion that exhibited dilatation. As soon as even a moderate amount of barium passed into the dilated portion of the bowel, it was usually no longer possible to see the area of dyskinesia as the markedly dilated and often redundant loop of sigmoid and proximal colon obscured the rectosigmoid completely.

This experience gave concrete evidence of a physiologic, partial obstruction of the rectum and rectosigmoid. If this conclusion was correct, colostomies above this point in the colon should give complete relief of the patient's symptoms.

Colostomies were performed in the normal bowel in three of the patients—that is, in the dilated portion of the colon—and in each instance all symptoms of Hirschsprung's disease disappeared. These patients behaved precisely as any patient would who had been relieved of large bowel obstruction by a colostomy. Closure of the colostomies was performed in these three patients and in each instance there was recurrence of the complaints which were not relieved until the colostomies were reopened. Colostomies have been performed on 12 other patients. Each one has been completely relieved of his symptoms and the large hypertrophied colon has decreased markedly in size.

The roentgen evidence of obstruction in conjunction with the clinical findings and experience with colostomies in these patients has led to the conclusion that the abnormality in Hirschsprung's disease rests exclusively in that portion of the rectum or rectosigmoid where the caliber of the bowel has been normal or less than normal but is the seat of a severe dysfunction, with disorganized peristalsis or failure of relaxation. The dilated and hypertrophied portion of the colon is believed to be simply the result of the longstanding partial obstruction. Therefore, it would seem that all therapeutic measures should be directed toward this short segment of malfunctioning bowel rather than to the obvious dilated and hypertrophied colon.

During the performance of transverse colostomies in the patients, the sigmoid loop was delivered in order to correlate the finding on direct examination of the sigmoid with what had been demonstrated in the roentgenograms. In all instances where the abnormal bowel extended up into the sigmoid, an abrupt change in diameter of the bowel was found. In some patients, the abnormal bowel was low in the pelvic cavity or below the pelvic peritoneum and, therefore, could not be seen. That such was the case could always be predicted from the roentgen films. Proximal to the point of narrowing, the colon would measure as much as 20 cm. in diameter. In the course of 5 cm., this tapered into bowel which was normal, or smaller than normal, in size (Fig. 3). In appearance and

to palpation, this bowel seemed far more normal than the dilated bowel above. However, the roentgen findings, as well as the experiences with colostomies in these patients, led to the conclusion that it was functionally deficient.

The next problem was to restore continuity of the intestinal tract so that the colostomy could be closed successfully. In most instances it was demonstrated by barium enema that the abnormal bowel extended down to the anus. Therefore, resection of the rectum would of necessity have to be performed down to the anus to assure removal of all of the malfunctioning bowel. No standard surgical procedure could be employed in this resection for none of them would leave the anus functionally intact.

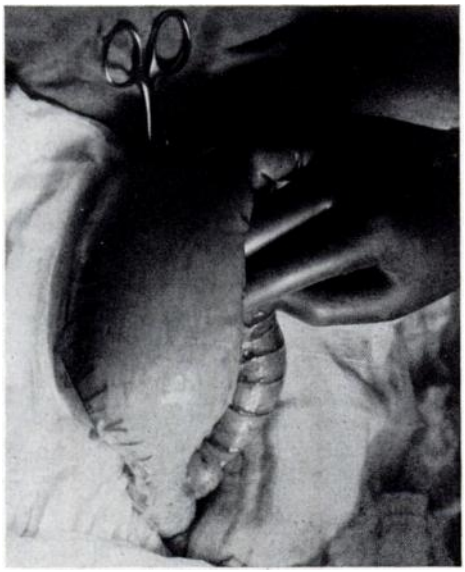

FIG. 3. Photograph of sigmoid loop taken during performance of colostomy. Markedly dilated bowel is proximal sigmoid. This dilatation terminates abruptly in distal sigmoid and bowel becomes smaller in caliber than normal.

The usefulness of a special pull-through form of resection, which had been devised and tested in the laboratory, has been demonstrated in 23 patients.[4] This special type of resection, which preserves the anal sphincter, is performed as follows: A left paramedian suprapubic incision is made, extending from the symphysis to a point a few centimeters above the umbilicus (Fig. 4). The demarkation between the functionally inadequate bowel and the normal colon is evident by the abrupt change in diameter of the colon, or, if a colostomy had been performed previously, this point had been marked by several black sutures. This is important, for after a diverting colostomy the dilated bowel becomes normal and the point of transition from the functionally normal to abnormal bowel is indistinguishable by examination of the bowel. The resection is carried from above this point of change down to the anus.

The first part of the operation is the same as a combined abdominal-perineal resection. The sigmoid and rectum are freed from all attachments in the pelvis down to the levator

ani muscle. The bowel is then divided proximal to the abnormal portion and both ends are closed with continuous catgut sutures (Fig. 5). The distal end is then pulled out through the anus, and the mucosal surface, which is now presenting, and the perineum are prepared with Zephiran scrubs (Fig. 6). Drapes are placed and the bowel is opened transversely 2.5 cm. from the anal skin margin. The proximal bowel is pulled down through this opening (Fig. 7).

The anastomosis is made, using first a row of interrupted black silk Lembert sutures through the muscular coats of the two segments of bowel. Up to this point the procedure has been relatively aseptic. The proximal bowel is then opened and the mucosa of the two ends are approximated with interrupted chromic catgut sutures. The anastomosed bowel is then pushed back through the anus and the suture line assumes a position about 2½ cm. above the anal skin margin.

This operation has now been performed on 23 patients. There have been no deaths. In 15, preliminary colostomies had been performed because of the advanced state of the disease. Closure of the colostomy has been performed on all of these patients except two. Abdominal distention has disappeared. No special diets, laxatives of any form, or enemas have been used postoperatively and all of the patients now have normal bowel movements. As far as can be determined, they are normal children. Three infants, the youngest two months old, were subjected to this type of resection without preliminary colostomies. These youngsters were all operated upon before malnutrition had set in; they all tolerated a one-stage operation and have done well postoperatively.

Following operation, the children have an urge to defecate, which is reported by two of the older patients (8 and 9 years) to be normal, as far as can be determined from their descriptions. All of them have normal sphincter control, both as to performance and to findings on digital examination. Most of these patients have been followed only a few months. However, one has been followed one year and another for 10 months and both continue as normal children.

These children have had such satisfactory results following operation that barium enema studies have not been performed in all subjects. However, for purposes of clinical investigation follow-up roentgen studies were made on five of them. The colon remains slightly larger than normal in these children. However, the emptying is excellent (Fig. 8).

Discussion

As has been stated, a true organic stenosis of the anus or rectum may occasionally be encountered in a patient with the clinical picture of congenital megacolon, and such a lesion should be looked for in patients suspected of having Hirschsprung's disease. Reports have been made of patients in whom there was a segment of narrow bowel distal to the megacolon but no significance has been placed on these anatomic observations. During the past five years, 26 patients have been studied with clinical diagnosis of Hirschsprung's disease, and in each one it was possible by careful examination to demonstrate a spastic segment of colon in the rectum or rectosigmoid with dilated bowel proximal to this. If such an appearance can be disclosed in a patient with a clinical picture of Hirschsprung's disease, it is felt that excision of the malfunctioning distal portion of the bowel should be performed with preservation of the anal sphincter, providing medical therapy has been unsuccessful. Two patients in this group are being managed successfully on a medical regime with mecholyl bromide. There is a third patient in

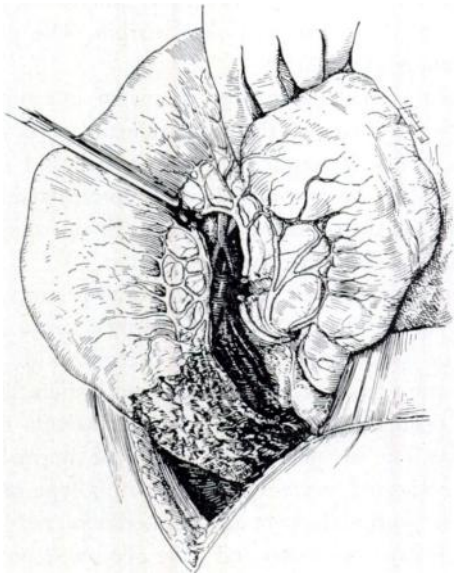

FIG. 5. Sigmoid loop has been delivered, rectosigmoid freed down to levator ani. Note change in caliber at peritoneal reflection in this patient. Sigmoid is now divided at about its midpoint.

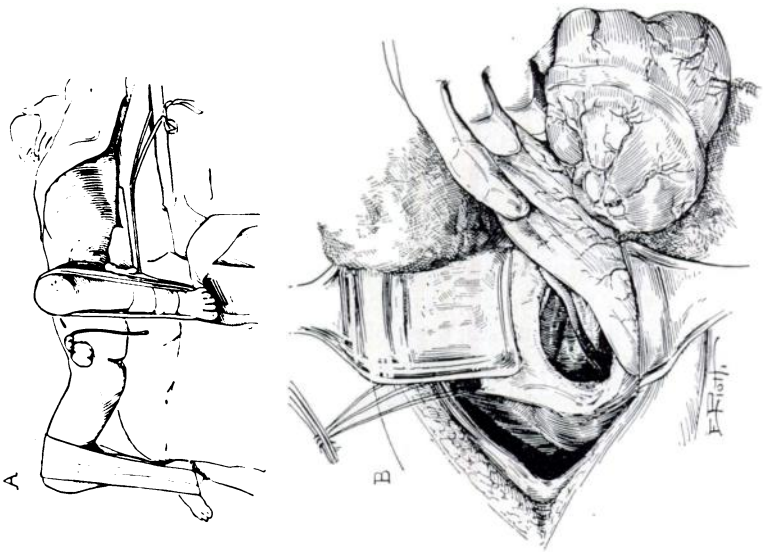

FIG. 4. A. Position on table is so arranged that 2 teams can work during 2nd half of operation. When pull-through is done, 1 team performs anastomosis; other team closes abdomen. B. Pelvis is exposed through midline incision. Right ureter has been identified.

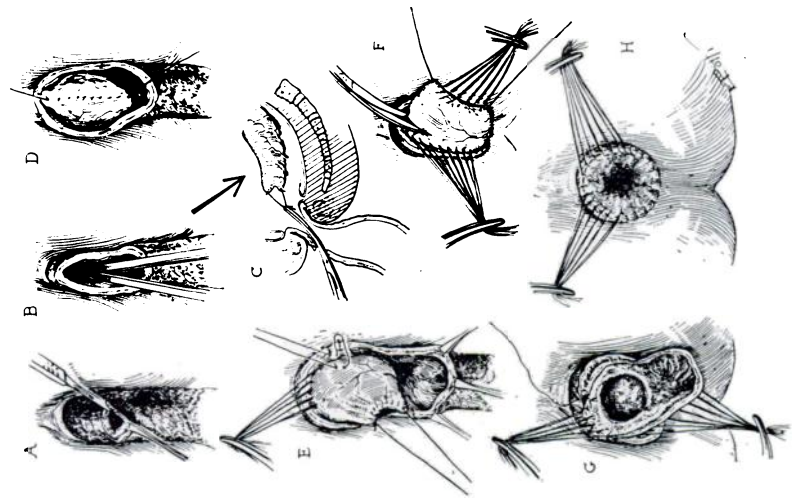

FIG. 7. A. Distal bowel is cut across 2.5 cm. from skin margin. B. Clamp is inserted through opening. C. Sutures on proximal bowel are grasped. D. Proximal bowel is pulled through distal bowel. E. Muscular surfaces are approximated by interrupted silk sutures. F. End is cut off proximal bowel. G. Mucosa is approximated with chromic catgut sutures. H. Suture line is completed and suture line is ready to be pushed through sphincter.

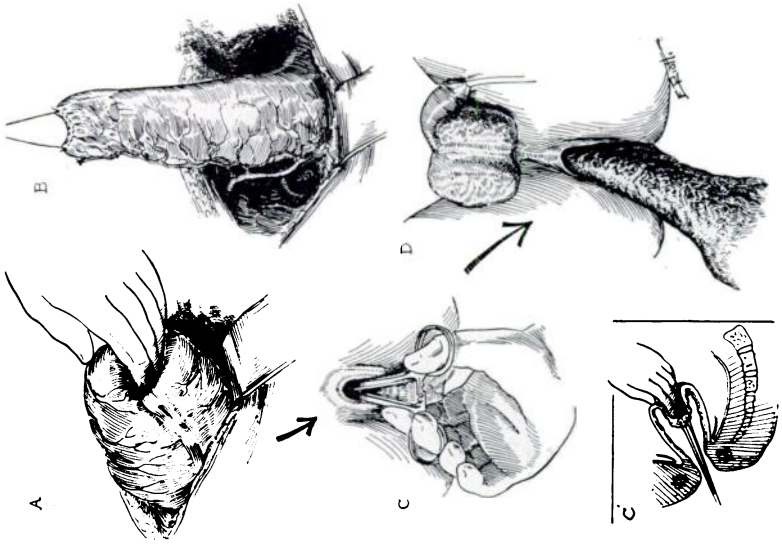

FIG. 6. A. Beginning of inversion of distal bowel is begun from above. B. Proximal colon is ready to be pulled through. C. Long curved clamp is inserted through anus. C.' Distal bowel is grasped and pulled through. D. Distal bowel protrudes from anus with its mucosa on surface.

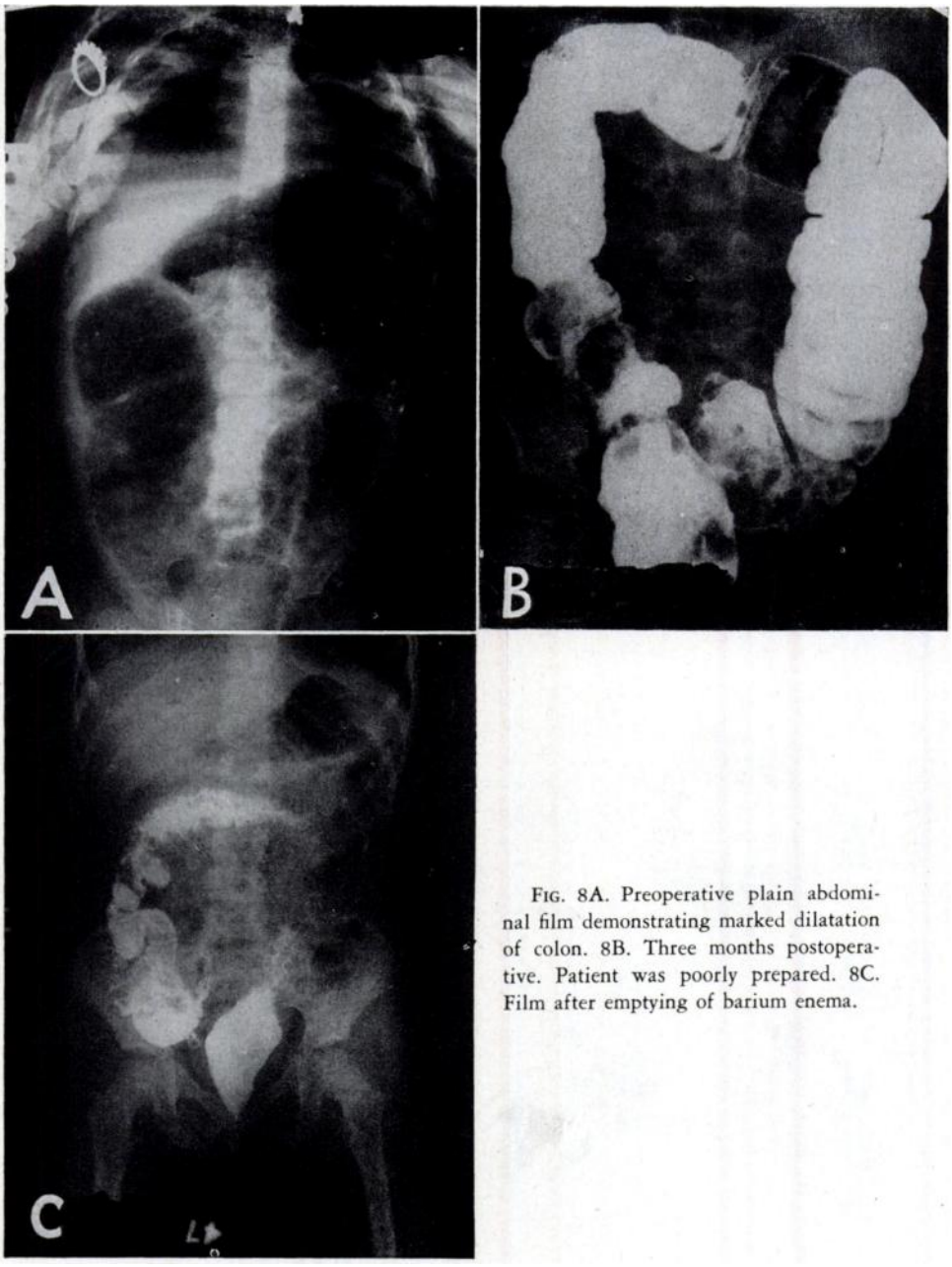

FIG. 8A. Preoperative plain abdominal film demonstrating marked dilatation of colon. 8B. Three months postoperative. Patient was poorly prepared. 8C. Film after emptying of barium enema.

whom the lesion is at the anus (or just above it) and daily rectal dilatation has been sufficient to control the disease.

Summary

Hirschsprung's disease, or congenital megacolon, is due to a functionally abnormal segment of bowel in the sigmoid or rectosigmoid. This can be demonstrated effectively by appropriate roentgenologic studies with barium as a contrast medium. The bowel above this abnormally functioning segment shows changes commonly found above an area of obstruction; the bowel is dilated and hypertrophied.

Removal of the functionally abnormal bowel (rectum or rectosigmoid) has relieved the patients' symptoms. A new operative technique is utilized in performing this resection. The fact is stressed that these children will have to be followed for a period of several years before any claims of a permanent cure can be made.

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Spanish Abstract

Nuevos Conceptos de la Etiología, Diagnosis, y del Tratamiento del Megacolon Congénito (Enfermedad de Hirschsprung)

El megacolon congénito, enfermedad de Hirschsprung, se debe a un segmento funcionalmente anormal del intestino en el sigmoideo o rectosigmoideo. Este puede ser demostrado eficazmente mediante estudios roentgenológicos apropiados con bario como un medio de contraste. El intestino arriba de este segmento que funciona anormalmente revela cambios comunmente encontrados arriba de una area de obstrucción; el intestino está dilatado e hipertrofiado.

Los síntomas del paciente se han mejorado al remover el intestino (recto o rectosigmoideo) que funciona anormal. Se utiliza una nueva técnica operativa al hacer esta resección. El énfasis está en que hay que continuar observando a estos niños por un período de various años antes de poder darlos por curados.

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