PSEUDO-HIRSCHSPRUNG'S DISEASE*

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Largely thanks to the work of Swenson and Bill (1948), Whitehouse and Kernohan (1948), and Bodian, Stephens, and Ward (1949), the definition of Hirschsprung's disease and its differentiation from idiopathic and symptomatic megacolon has become a clear-cut diagnostic procedure which is now part of the daily routine in every paediatric surgical unit.

Occasionally a short-segment aganglionosis may present as an idiopathic megacolon, and complete aganglionosis of the colon may be difficult to distinguish from other types of low intestinal obstruction in the newborn. Rectal biopsy will always settle the diagnosis. Diagnostic difficulties in patients with a narrow segment of the usual length are rare. I have recently treated a child, presenting with an unusual type of megacolon which does not fit into the usual classification of this syndrome.

Case Report

A 10-month-old girl was admitted with a history of severe constipation and abdominal distension, starting at the age of 8 months, when she was weaned. Before this time she had emptied her bowels only once a week, without, however, showing evidence of discomfort or abdominal distension. The anus was normal and the rectum was empty. Radiological examination had been performed in another hospital (Fig. 1), showing a normal-sized rectum and distal sigmoid colon and marked distension and faecal impaction proximal to this. The radiographs were typical of Hirschsprung's disease. After an unusually long period of difficult bowel cleansing, laparotomy was performed. Inspection of the colon seemed to confirm the diagnosis of Hirschsprung's disease. A narrow segment extending to the apex of the sigmoid flexure, with a funnel-shaped zone of transition into the grossly dilated proximal sigmoid, was seen. Two atypical features were soon disclosed, however. There was no hypertrophy of the bowel wall, and the gaseous contents of the dilated bowel could be propelled down through the narrow segment without the typical resistance. No mechanical obstruction could be demonstrated. Proximally, the dilatation gradually decreased. A Duhamel type of recto-sigmoidectomy was performed.

FIG. 1.—Pre-operative barium enema suggesting typical Hirschsprung's disease.

Histological examination revealed no abnormality of the intramural plexuses or other abnormality. The post-operative course was uneventful. The child recovered but a slight constipation persisted, which has been easily managed with a mild aperient. At a recent follow-up, 7 months after operation, the child was in an excellent condition and had no abdominal distension. Rectal examination revealed an adequate Duhamel anastomosis without faecal retention in the rectal pouch. Barium enema showed a moderate persistence of colon dilatation.

This is obviously not a case of Hirschsprung's disease, nor does the distal narrow segment belong to the ordinary criteria for a diagnosis of idiopathic

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megacolon. There were no signs of gross mechanical obstruction nor of any other disease that might have caused a symptomatic megacolon. The intramural plexuses showed no degenerative changes, suggestive of the South American type of acquired megacolon. This seems to exhaust our diagnostic alternatives without, however, solving our diagnostic problem.

I would like to call the disease picture presented by this patient 'pseudo-Hirschsprung'. This term has been used as a synonym for idiopathic megacolon, especially by Ravitch (1958); idiopathic megacolon, however, as a rule has little in common with Hirschsprung's disease. Besides, idiopathic megacolon is a well-established term and a very good one, when properly used. It thus seems unwarranted to abandon an adequate name for one less appropriate. At the present time, 'pseudo-Hirschsprung' seems to be an adequate name for a chronic constipation with essentially the same clinical, radiological, and gross pathological changes as in Hirschsprung's disease but without the pathognomonic congenital aganglionosis.

Before entering into a discussion of the type of disturbance presented by our patient, a few reports of similar cases should be reviewed. Bill, Creighton, and Stevenson (1957) operated on 2 patients with constipation since birth, who fulfilled the clinical and radiological criteria for a diagnosis of Hirschsprung's disease. Both of the resected specimens showed normal ganglion cells in all areas. Both patients were relieved of obstruction following rectosigmoidectomy.

Swenson and Rathausser (1959) reported on 3 patients with a new disease entity entitled 'segmental dilatation of the colon'. All 3 patients had had constipation since birth. One child had a fusiform dilatation of the sigmoid colon which made it indistinguishable from aganglionic megacolon. In the second child, the lesion was in the proximal transverse colon and was thought to represent dilatation proximal to a long aganglionic segment extending from the anus to the mid-transverse colon. In the third patient the dilatation involved the entire sigmoid colon. At operation, the dilatation was found to start and end abruptly at its proximal and distal bowel attachments in all three patients. There was uniform hypertrophy of the muscle layer of the dilated segment. Normal ganglion cells and nerve fibres were found in the dilated segment as well as in the colon proximal and distal to it in all three patients.

Nixon (1961) described 2 patients with severe constipation and marked abdominal distension. Barium enema showed distension of the colon

proximally with an undilated distal segment. There was no hypertrophy of the bowel wall. In both cases a colostomy was performed. The first infant died with faecal masses persisting in the right colon. At necropsy, cerebral atrophy was found. In the second child the colostomy discharged excessive fluid. After treatment of the electrolyte disturbance the colostomy was closed and the child recovered. In both of these patients normal ganglion cells were found in the bowel wall.

The first question to be answered is whether discussion of these 8 patients (including our own patient) as a homogeneous group is justified or not. There are obvious similarities between all the case reports: the patients presented with symptoms and signs of partial intestinal obstruction; barium enema studies revealed proximal megacolon with an undilated distal segment; and finally and most remarkably, the histological findings were perfectly normal in all of the specimens examined.

In other respects, however, some more or less essential dissimilarities exist. The three patients reported by Swenson and Rathausser presented a gross pathology of a specific type, which is clearly divergent from that of the remaining cases, from the ordinary findings in Hirschsprung's disease, and from those of idiopathic and symptomatic megacolon. The suggestion of these authors that 'segmental dilatation of the colon' is a new entity seems justified, and these three patients should be excluded from further discussion on the 'pseudo-Hirschsprung' group.

One of Nixon's patients died. At necropsy, the brain showed cerebral atrophy. Dr. M. Bodian suggested the possibility of a central origin for the bowel abnormality. Megacolon with a distal narrow segment has been described in patients with myxoedema (Salmi and Lahesmaa, 1956; Ravitch, 1958). These cases are not included in this review, because they are considered to belong to the group of symptomatic megacolon. The same may be true with regard to Nixon's case.

In the remaining 4 patients, no obviously essential dissimilarities were found. Absence of hypertrophy of the wall of the dilated bowel was a striking feature in Nixon's remaining patient as well as in our own. Bill *et al.* do not furnish any evidence in this respect. This feature is possibly related to the lack of resistance to propulsion of intestinal contents through the narrow segment, noted in our patient but not mentioned by Bill *et al.* or by Nixon. Normal intramural plexuses were found throughout the entire segments resected in the present case and in the 2 patients of Bill *et al.*, which should eliminate the diagnosis of segmental aganglionosis.

It seems, therefore, that at least 4 of the original 8 patients belong to a homogeneous disease entity, tentatively called pseudo-Hirschsprung's disease. The pathology and the aetiology of this entity remain obscure. The possibility of a neurogenic disturbance of unknown origin, located more centrally than in the intramural plexuses of the colon may be postulated. This is, however, a purely theoretical and hypothetical suggestion. The course of the disease in Nixon's surviving patient is most interesting from the pathogenetic point of view: this patient recovered after a temporary colostomy and was progressing nicely after closure of the colostomy. But the length of the follow-up period is not stated. Nixon suggests that the obstruction was due to a temporary functional paralysis of the colon. It is regrettable that our patient, as well as the 2 patients of Bill *et al.*, were treated by recto-sigmoidectomy, thus depriving us of the chance to confirm or exclude the temporary nature of the disorder in our patients.

Summary

A case with many of the clinical features of Hirschsprung's disease, but without any abnormality

of the intramural plexuses, is described. Similar cases have been reported previously by other authors, and the term pseudo-Hirschsprung's disease is suggested to describe the entity, the aetiology of which is obscure.

REFERENCES

- Bill, A. H., Jr., Creighton, S. A., and Stevenson, J. K. (1957). The selection of infants and children for the surgical treatment of Hirschsprung's disease. *Surg. Gynec. Obstet.*, **104**, 151.
- Bodian, M., Stephens, F. D., and Ward, B. C. H. (1949). Hirschsprung's disease and idiopathic megacolon. *Lancet*, **1**, 6.
- Nixon, H. H. (1961). In Discussion on megacolon and megarectum with the emphasis on conditions other than Hirschsprung's disease. *Proc. roy. Soc. Med.*, **54**, 1037.
- Ravitch, M. M. (1958). Pseudo Hirschsprung's disease. *Ann. Surg.*, **147**, 781.
- Salmi, T., and Lahesmaa, P. (1956). Pseudo-Hirschsprung'sche Erkrankung bei einem Myxödem-Säugling. *Acta paediat. (Uppsala)*, **45**, 428.
- Swenson, O., and Bill, A. H., Jr. (1948). Resection of rectum and rectosigmoid with preservation of the sphincter for benign spastic lesions producing megacolon. *Surgery*, **24**, 212.
- , and Rathauer, F. (1959). Segmental dilatation of the colon. *Amer. J. Surg.*, **97**, 734.
- Whitehouse, F. R., and Kernohan, J. W. (1948). Myenteric plexus in congenital megacolon. *Arch. intern. Med.*, **82**, 75.