Hirschsprung's disease

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Introduction

The first description of this disease is usually credited to Frederick Ruysch, a seventeenth-century Dutch surgeon (Whitehouse, Bargen & Dixon, 1943). The first well-documented paper was published in 1888 by Hirschsprung, a Danish paediatrician.

Treves (1898) discussed two types of megacolon, one arising out of habitual constipation and a second appearing to have an obstructive origin. He described his treatment of a 5-year-old girl with a megacolon apparently of the obstructive variety whom he treated by operation. This patient was seen 57 years later at the Royal Free Hospital, London (Johnson, Davis & Evans, 1957).

After the beginning of this century, many workers reported absent or reduced and abnormal ganglion cells in the narrowed segment (Tittel, 1901; Hawkins, 1907; Dalla Valle, 1924; Cameron, 1928). The significance of this was not fully appreciated until the reports of Robertson & Kernohan (1938) and Tiffin, Chandler & Favour (1940), who suggested that the primary pathology was a lack of peristaltic waves in the narrowed segment due to an absence of the ganglion cells, and that the megacolon occurred secondarily to this physiological obstruction. However, it was not until after World War II that the abnormal histology and the abnormal physiology of the narrowed segment were correlated and the disease established as a separate entity (Zuelzer & Wilson, 1948; Whitehouse & Kernohan, 1948; Swenson, Neuhausen & Pickett, 1949; Bodian, Stephens & Ward, 1949).

Diagnosis

In children, chronic constipation (often punctuated with bouts of obstipation or intestinal obstruction) and a failure to thrive are the classical symptoms of this disease. These symptoms begin in the first year of life, and the diagnosis is not usually difficult if there is a high index of suspicion. The child may, however, present during an attack of enterocolitis (Forshall, 1964) with a grossly distended abdomen and give a history of diarrhoea and sometimes also of vomiting (Nixon, 1964). The majority of these children will also give a previous history of bowel dysfunction starting from birth or within the first few months of life.

Following the work of Ehrenpreis (1946), who gave a classical clinical and radiological study of the disease in infancy, the diagnosis has been made with increasing frequency in the infant. In a series of 110 patients, the diagnosis was made in sixty-six before the age of 4 weeks (Forshall, 1964). Of these, forty-four were admitted to hospital during the first 3 days of life, mainly with vomiting, reluctance to feed and some variation in the passage of meconium or faeces accompanied by some degree of abdominal distension. Vomiting usually started on the 2nd or 3rd day and if it persisted the vomitus became bile-stained. In nearly all the patients in this group it was possible to produce meconium either by the passage of a thermometer or finger into the rectum or as a result of colonic irrigation.

X-ray examination is of value in confirming the diagnosis at all ages. In the child, a barium enema will often show the megacolon with a classical narrowed segment in the distal colon or in the rectum. In infants, in the first few days of life, plain films of the abdomen are taken in the erect and supine positions to determine the spread of air to the distal colon. Differentiation of the large from the small bowel may be extremely difficult, but a lateral X-ray will usually show the sweep of the descending colon to help in the differentiation (Berdon & Baker, 1965).

If there is still doubt, further help may be obtained from a plain film with the infant in the inverted position (Berman, 1956). However, adequate radiological diagnosis of Hirschsprung's disease usually requires contrast enema studies. The infant needs no previous preparation and is screened while the contrast medium is being run in. Frontal and lateral spot films are taken, and when the colon has been filled no effort is made to produce evacuation.

A further X-ray taken the following day will often show the narrow segment, although in the infant this is not well developed and may be very difficult to demonstrate (Forshall, 1964). Even if this narrow segment is not demonstrated, X-rays
taken at 24 hr, and occasionally at 48 and 72 hr, will show persistent distension of the colon with air fluid levels and retained contrast medium (Berdon & Baker, 1965). Histological diagnosis may then be obtained from a rectal biopsy.

In 1956, Clatworthy, Howard & Lloyd described 'the meconium plug syndrome' and since that time errors in differentiating between this condition and neonatal Hirschprung's disease have occurred. Gillis & Grantmyrie (1965) reported the fatal termination of a case of neonatal Hirschprung's disease in which such an error was made. Ellis & Clatworthy (1966), in a recent review of 'the meconium plug syndrome', mentioned two such patients and suggested a careful follow-up, using further barium enema studies and even rectal biopsy, in any infant who does not rapidly return to normal bowel habits after apparent dislodgement of the plug.

Rectal biopsy was introduced by Swenson, Fisher & MacMahon (1955), who suggested that the specimen should contain an adequate amount of rectal muscle for examination for ganglion cells. Subsequently, rectal biopsy of a large piece of mucosa and submucosa was introduced by Bodian (1960), but the histological interpretation of this specimen requires considerable experience. Because of the difficulties encountered with the Swenson technique of rectal biopsy, Hiatt (1958) introduced a posterior approach to the rectum, incising midway between the coccyx and the anus and removing a specimen of rectal muscle without opening the rectal mucosa. Because all of these methods at all ages require a general anaesthetic, Shandling (1961) introduced a new method of rectal biopsy. Through a sigmoidoscope, using a side biting biopsy forceps, he removed four small pieces from the valves of Houston which contain circular muscle fibres (Hughes, 1957). This procedure is simple, is not attended by any serious complications and can be carried out without an anaesthetic in the infant and under sedation only in the child.

In older infants and children, the diagnosis can be made on the typical clinical features and the radiological findings. However, there are several reports of patients with the diagnosis of Hirschsprung's disease made on clinical grounds with normal ganglion cells demonstrated on rectal biopsy (Ehrenpreis, 1965; Shandling, 1961). For this reason, rectal biopsy, probably by the method recommended by Shandling (1961), is mandatory.

Management

Because the pathology was ill understood, the management of these patients was at one time a haphazard affair. Swenson & Bill (1948) first described a sphincter-preserving recto-sigmoidectomy which was performed successfully in the laboratory animal and in three children. The following year Bodian et al. (1949) classified the types of megacolon and established the treatment of megacolon, including the congenital variety, on a rational basis. However, the mobilization of the distal rectum, required as part of the Swenson procedure, needs considerable operating skill and is time consuming, and will therefore cause profound shock in the infant (Forshall, 1964). Wyllie (1957) reported a 9% mortality in 152 patients, while Hiatt (1958) had no mortality but some 16% of 150 patients had some remaining colo-rectal dysfunction. Many surgeons have become dissatisfied with the Swenson procedure because of a mortality rate often higher than Wyllie's and a cure rate often lower than Hiatt's (Forshall, 1964).

State (1952) reported sixteen cases of Hirschsprung's disease treated by a new operation, based on his opinion that the rectum was normal and that the entire left colon was abnormal. He divided the rectum 6-10 cm above the anus, excised the entire left colon and anastomosed the right colon to the rectal stump. This is a much simpler procedure than the Swenson operation and in State's hands has given good results (State, 1963). Rehbein & von Zimmerman (1960) performed a similar operation on sixty-seven infants of whom six died. In about one-third of the surviving sixty-one patients there was stenosis of the anastomosis or constipation which was treated by repeated dilations. Follow-up barium enemas also showed the remaining colon to be more dilated than the pre-operative examinations.

Duhamel (1956) described a modification of the Swenson procedure. The aganglionic segment is excised and the rectum is closed on a level with the peritoneal reflexion. The normal colon is then brought through the posterior wall of the closed rectal pouch and the opposing anterior wall of the colon and the posterior wall of the rectum are crushed between Kocher clamps resulting in a triangular defect. The resulting large capacity rectum has rectal wall in its anterior half to provide sensation and normally innervated colon in the posterior half to provide propulsive power. This pelvic dissection is only carried out behind the rectum and there is no interference with the pelvic nerves. It carries a low operative risk and so, unlike the Swenson procedure, permits a definitive procedure even in the very young infant (Ehrenpreis, Livaditis & Okmian, 1966; Forshall, 1964).

Medical management is seldom justified (Forshall, 1964). Richards & Hiatt (1953) and Ziskind & Gellis (1958) have reported the danger
of repeated colonic irrigations in the infant. Also, while the infant or child is undergoing medical management, it is susceptible to the very lethal necrotizing enterocolitis. At present, patients with Hirschsprung's disease are usually treated either by the Swenson or the Duhamel procedure. Forshall (1964) believes that an early colostomy prevents the attacks of intestinal obstruction to which these infants are prone and will guard against developing enterocolitis, but as the Duhamel procedure permits early definitive surgery, preliminary colostomy is now rarely necessary (Ehrenpreis et al., 1966; Kostia, 1962).

**Review of cases**

In the 4 years between the beginning of January 1962 and the end of December 1965, ten patients were treated at the University College Hospital, Jamaica, for Hirschsprung's disease. Seven of these patients were males showing the usual preponderance of males seen in this disease (Pilling & Cresson, 1962). Only three patients were seen during their first year of life. Altogether eight of the ten children were under 3 years of age, the other two being over 3 years and having had operative procedures elsewhere. In five patients there was intestinal obstruction or alteration in the bowel habits before the child was 1 year old (Table 1). Two of these five patients presented and were diagnosed in the neonatal period. The other three did not attend this hospital until they were all over 2 years of age and had been treated by purgatives only or by purgatives and evacuations under anaesthetic. Of the remaining five patients, one developed constipation at 3 months of age and was given purgatives with the occasional colonic washout until it attended this hospital at 3 years of age. In two other patients, symptoms started at the ages of 3 and 6 months respectively. They were treated by repeated colonic irrigation until the ages of 8 and 16 months respectively. Of the last two patients, one attended at another hospital at the age of 3 years with symptoms of 2 years duration. A colostomy was performed and was subsequently closed without the correct diagnosis being made. The last child is unusual in that bowel symptoms developed later than usual. She had been treated for malnutrition as an inpatient in the University College Hospital at the age of 16 months. Bowel habits were normal at that time and during subsequent attendance at the Paediatric Clinic until the child was 2 years old.

The extent of involvement may be divided into those in which the involvement extends to the junction of the descending and sigmoid colon, which will be classified as 'short' or those in which the colon above this level is also involved, which are classified as 'long' (Forshall, 1964). Among our patients there were six of the short segment type and three of the long segment type. In one patient the extent of the disease was not known. Of the three patients with long segment involvement, two were very ill on admission at the age of 5 days and 8 months respectively. In the third patient with a long aganglionic segment, symptoms started at the age of 2 years. The six patients with short segment involvement did not attend at this hospital until they were over 1 year of age. They had been controlled until this time by purgatives and colonic irrigation.

The main presenting symptom was chronic constipation. Two infants had intestinal obstruction, while a third gave a history of repeated attacks of abdominal distension. On examination all patients were malnourished and all showed some degree of abdominal distension. In one child aged 10½ years distension was gross. In all six patients over 1 year of age there was a palpable faecal mass occupying the descending and sigmoid colon. In two of these patients the rectum also was dilated and packed with hard faeces down to the anal canal. One of these children seen at 16 months of age was misdiagnosed as having an acquired megacolon because of this finding and an equivocal barium enema. He did not at that time have a rectal biopsy.

**Investigation**

Contrast studies using barium were carried out in eight patients. In three patients a megacolon with retention of barium for more than 24 hr was found but no narrow segment could be demonstrated. In four patients a narrow segment was demonstrated in addition to the megacolon. In one patient a barium enema examination could not be interpreted because of a previous operation elsewhere in which the colon had been excluded and the ileum anastomosed to the rectum; no adequate description of the operative procedure was available. In two infants plain frontal films of the abdomen only were taken. In both, air was seen throughout the small and large bowel and

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<th>Age when symptoms began</th>
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<td>Less than 7 days</td>
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<td>7 days to 3 months</td>
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there was moderate distension with fluid levels. Intravenous pyelography was carried out in six patients and in four the urinary tract was normal. In one patient there was slight enlargement of both ureters in the pre-operative pyelogram, and in another patient there was definite dilatation of both ureters and a large, relatively atonic, bladder. Unfortunately this latter patient defaulted from follow-up and a repeat of this examination following his definitive treatment was not obtained.

**Treatment**

Colostomy was performed in six patients. In two patients a blind transverse colostomy was performed and the colonic muscle at the site of the colostomy biopsied. In both of these patients there were ganglion cells present and the transverse colostomy functioned satisfactorily. In four patients sigmoid colostomy was performed. In two of these a frozen section examination confirmed the presence of ganglion cells. One patient had a sigmoid colostomy at another hospital. Biopsy was taken from the non-functioning sigmoid colostomy and this showed ganglion cells. In the last patient a blind sigmoid colostomy was performed. This did not function. Ganglion cells were absent in the biopsy of the colostomy. This patient had a long aganglionic segment. Eventually a right transverse colostomy was performed, siting being determined after repeated frozen section biopsy carried out at a second operation.

No definitive treatment was possible in one patient who died from enterocolitis. In one a colostomy has been performed with a view to resection later. In the other eight patients the definitive treatment consisted of a rectosigmoidectomy as described by Duhamel (1956) or the modification of the Duhamel procedure with preservation of the internal sphincter, as described by Grob (1960). There were no deaths and only minor complications. Patients have been followed up for from 4 months to 3 years.

**Results**

Soiling persisted for over 3 months after the operation in two patients. In one patient it had cleared completely by 1 year and in the other there was very occasional soiling only at the last follow-up 5 months after operation. This patient had considerable soiling in the immediate post-operative period, only slightly improved on the administration of kaolin and linctus codein, but markedly improved on arrow-root.

In two patients a post-operative examination suggested that the rectal pouch might be too long but both of these patients have since defaulted from follow-up. In one patient the pouch is definitely too long and he has had two attacks of faecal retention in the pouch over the past 3 years. It is planned to readmit him for enlargement of the colo-rectal opening. One child has continued to have five bowel actions per day on the last follow-up 5 months after operation; he has since defaulted from follow-up. None of the other patients has more than four bowel actions per day.

**Mortality**

There was only one death. This infant presented at 4 weeks of age with abdominal distension, was correctly diagnosed and was started on medical management. He was satisfactorily controlled while in hospital and was discharged home 4 weeks later having gained nearly 2 lb in weight. He was readmitted a few days after discharge with a severe enterocolitis and was treated by intravenous fluids only. The importance of decompressing the bowel by passing a rectal tube and by saline colonic irrigations was not appreciated, and the infant died shortly after readmission.

**Discussion**

Hirschsprung's disease usually leads to symptoms early in life, but Kottmeier & Clatworthy (1965) have found that bowel dysfunction due to functional megacolon may also present in infancy. This is a further reason for the use of low rectal biopsy in all patients prior to any form of resection. So-called 'skip areas', if they occur, must be of very rare occurrence (Forshall, 1964; Kottmeier & Clatworthy, 1965).

In retrospect, the only death was probably preventable. A definitive procedure should have been carried out during the first admission. Our experience agrees with other authors (Ehrenpreis et al., 1966; Kostia, 1962) who advocate a Duhamel resection in one stage. However, colostomy is a useful preliminary in those patients who have had Hirschsprung's disease for a long time and in whom the colon is large and very hypertrophied. Such a colostomy was performed in two children and the resection delayed for over 9 months. At the end of this period the size of the bowel had returned to manageable proportions.

The diagnosis of Hirschsprung's disease can be made in infancy. In Jamaica, where gastroenteritis is a common cause of death in infants, it is possible that some of the deaths attributed to gastroenteritis are in fact due to enterocolitis secondary to Hirschsprung's disease.

Genito-urinary complications do not appear to be serious. Swenson et al. (1952) found that of twenty-two patients with Hirschsprung's disease, twelve had an increased bladder capacity and decreased detrusor activity of the bladder muscle.
In addition, four of seventy-six patients operated on for Hirschsprung's disease showed dilatation of the upper urinary tract. He suggested that these abnormalities may be due to a diminution of the number of parasympathetic ganglion cells in the bladder. However, Langer & Thomson (1959) found only one abnormal pyelogram in fourteen cases and this one returned to the normal pattern post-operatively and was attributed to chronic lower tract obstruction secondary to the pressure of a grossly dilated rectum. Furthermore, Leibowitz & Bodian (1963) performed ganglion cell counts in two children with Hirschsprung's disease and found them to be normal both in number and morphology. We were unable to carry out a follow-up pyelogram on one patient who appeared to have a large atonic bladder and dilated ureters.

The Duhamel procedure is technically an easy operation with a low operative risk (Ehrenpreis et al., 1966). The Swenson procedure is difficult, has a higher operative risk and post-operatively is more likely to develop severe complications usually due to leakage at the anastomosis (Kostia, 1962; Forshall, 1964). The results from the State procedure (1952) and its modification described by Rehbein et al. (1960) are unsatisfactory.

The increasing necessity for performing the definitive procedure in infancy favours the easier and safer Duhamel procedure although even this procedure is not entirely free of complications. Leakage from the rectal stump has been reported (Ehrenpreis et al., 1966). Soiling or anal incontinence has also been a significant complication in all reports. Only one of our patients had significant soiling and this responded to linctus codein and arrow-root. Faecal impaction of the rectal stump may also occur but it is amenable to treatment and should not be a permanent disability.

Summary
Ten cases of Hirschsprung's disease treated at the University College Hospital, Jamaica, are reported. The diagnosis and surgical treatment are discussed. Early diagnosis based on symptomatology, radiography and rectal biopsy is stressed. Early treatment by the Duhamel resection is advocated as a safe procedure.

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