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The Early Diagnosis of Hirschsprung's Disease

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Abstract. The majority of cases of aganglionosis present and can be diagnosed in the neonatal period. Enterocolitis is a common and fatal complication so that early diagnosis is important. The plain films and enema features of 47 cases diagnosed in the neonatal period were examined. Except when a bowel wash out had been given, all these cases showed obstructive fluid levels in the plain films and the common type of short segment aganglionosis gave an "egg on end" appearance of low small bowel obstruction in the inverted lateral view. The classical enema features take time to develop and the narrowed aganglio wic area is not always obvious but can be shown by a careful technique of slow filling with delayed films. A high degree of accuracy was obtained except in the cases of preliminary lavage, enterocolitis and meconium plug. To make an early correct diagnosis, it is important to have close correlation between clinicians, radiologists, and pathologists.

Key words: Newborn, intestinal obstruction, barium enema technique, aganglionosis, Hirschsprung's disease.

Introduction

Currently the diagnosis of neonatal aganglionosis can be established by (i) a perceptive clinical assessment, (ii) a careful evaluation of plain radiographic appearances together with a reliable technique at barium enema, (iii) rectal pressure studies, and, (iv) histological studies.

Eighty per cent of all cases of aganglionosis may present in the neonatal period and a high degree of accuracy in radiological diagnosis can be achieved [23]. At Red Cross War Memorial Children's Hospital aganglionosis is the commonest form of intestinal obstruction. The "classical" appearance of the child with a distended abdomen, wasted limbs, stunted growth, and severe constipation is now much rarer: such children were probably the few survivors among many patients born with aganglionosis. Barium enema can be diagnostic in the neonate [10] as well as in the older patient.

In this paper the findings in 85 cases of aganglionosis who were studied radiologically in the 8 year period ending in 1972 are reviewed.

Case Material

Among 85 patients with Hirschsprung's disease there were 47 (55%) in whom the diagnosis was made in the neonatal period. No patient was a premature infant and the sex ratio was 3 male to 1 female (19).

Eighty per cent of patients had a short aganglionic segment involving the rectum or recto-sigmoid colon [25]. Total colonic aganglionosis occurred in only 3 patients and among the remainder most patients' aganglionic segment extended only into the descending colon. 8 cases (17%) presented as meconium plugs on barium enema.

A. Clinical Features

Clinical evaluation initially indicated the correct diagnosis. Important features in the neonatal period suggesting obstruction, were vomiting and abdominal distension but there was almost invariably a concomitant diarrhoea [2]. Commonly these infants were well at birth but they developed the clinical features during the first few days of life. Obstruction was not always progressive for it was sometimes temporarily relieved by spontaneous evacuation of the large bowel and this had sometimes been initiated by rectal examination.

Enterocolitis was a common complication and it was responsible for the death of 7 patients, 6 of whom were neonates (12.7 per cent of all neonates).

B. Radiological Features

1. Plain Radiographs

Supine, erect and inverted lateral projections were used.

Aganglionosis was suspected in all infants with multiple fluid levels. In the common short segment type of disease the gas filled dilated bowel often formed an "egg on end" appearance in the lateral inverted film with the gas shadow tapering at the point where gas entered the narrow transitional zone and aganglionic segment. Frequently there was an absence of rectal gas.

2. Barium Enema

Technique. A 30 per cent W/V barium was injected into the rectum from a 50 ml syringe via a simple rubber catheter. (An inflatable balloon catheter was never used for these may damage or even perforate the bowel as has been seen in patients referred from other centres). The catheter was not

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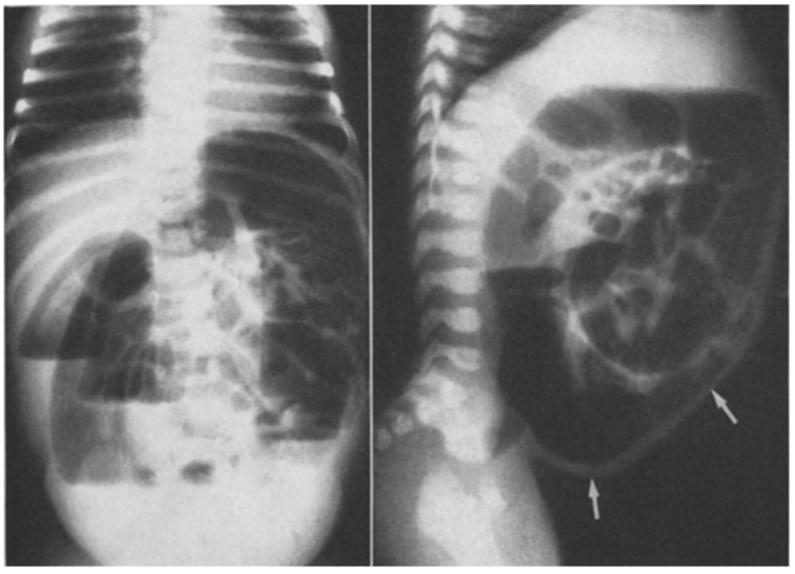
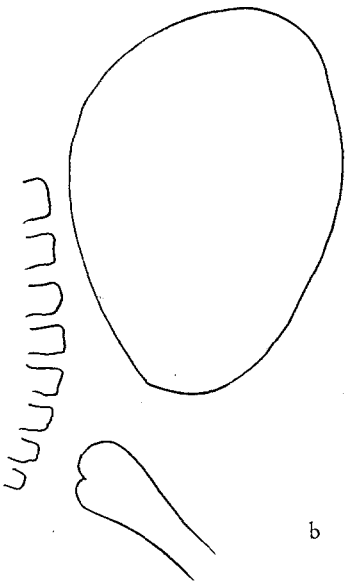


a



b

Fig. 1a. Plain films of short segment aganglionosis. The erect A. P. and inverted lateral view. The white arrows point to lower portions of the 'egg on end' appearance in the inverted lateral view. — Fig. 1b. Diagrammatic representation frequently the 'egg' is tilted on its end, by the distended posterior colon

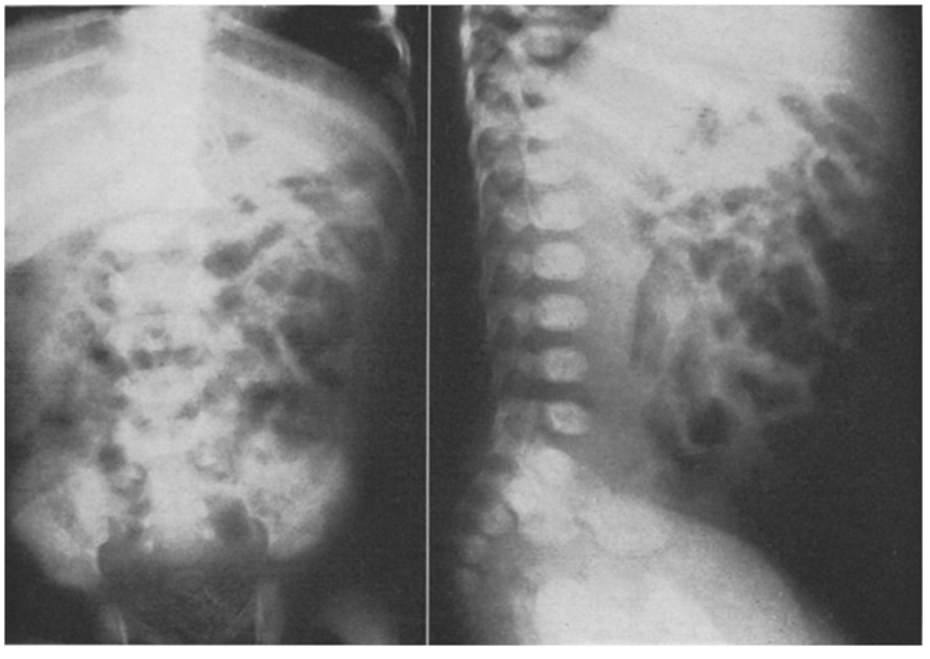


Fig. 2. Same case as Fig. 1a. The X-ray appearances immediately after saline cleansing washouts had been given

fixed by tape but it was held in place by digital compression across the buttocks with the infant in either the lateral or prone position. This technique necessitated either protection of the radiologist's hand by a lead shield or careful collimation of the X-Ray beam. Only 0.006 rads were recorded on the radiologist's hand by monitoring during a 3 minute fluoroscopy period [10].

Interpretation of enema findings. Accurate diagnosis was not possible in the first examination when (i) a preliminary washout had been given (Fig. 2), (ii) enterocolitis was present, producing spiculation and spastic areas or a cobble-stone appearance due to mucosal oedema (Fig. 3, 4, and 5), and, (iii) aganglionosis was present in conjunction with meconium plugs (17 per cent of the neonates).

Fig. 3

Fig. 5

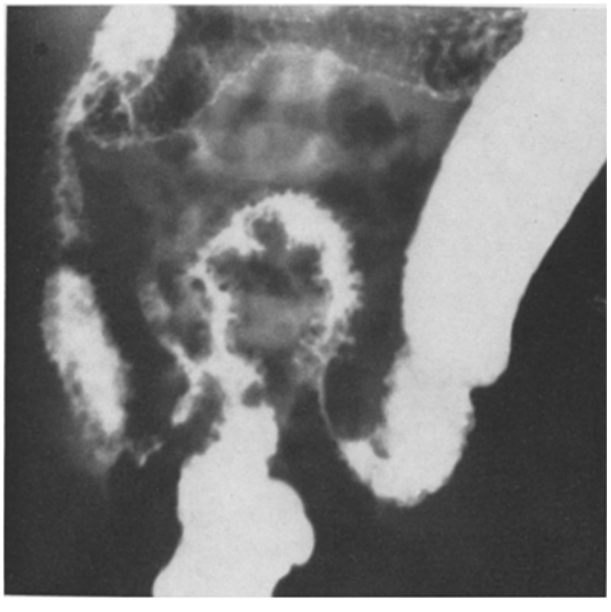


Fig. 4

Figs. 3, 4, 5. A proven case of short segment aganglionosis with a complicating enterocolitis. Note the lack of haustal pattern, the spiculation, mucosal oedema and the varying appearance and spasticity of the rectosigmoid and descending colon in films taken during the same examination

The narrow segment with the more proximal megacolon could be easy to miss at the time of injection but delayed films proved most important. Some minutes after completing the injection of barium the *early delayed film* showed the spastic narrow aganglionic segment more clearly. Abnormal contractions and writhing movements of an incoordinated type were visible at fluoroscopy in the narrow segment and these findings, recorded on videotape or film over the last 3 years, have been reviewed. The *late delayed films* taken 12 to 24 hours after the injection of barium showed retention of barium with the aganglionic segment often being best seen on the lateral projection (Fig. 6): this film was of limited value when enterocolitis was present.

In the long segment type with aganglionosis extending into the descending colon, the same diagnostic principles were used as in the commoner short segment cases: but the diagnosis was often more difficult. The lateral delayed film proved very useful in assessing the extent of the lesion. In two patients with total aganglionosis in whom barium enema had been carried out at the referring hospital retention of barium was still evident some days later (Fig. 7, 8).

In one infant who, in addition to aganglionosis, had both pyloric stenosis and a malrotation the diagnosis of aganglionosis was not suggested by radiological studies.

C. Manometric Study

This technique is a relatively recent development [18, 24] which is likely to be of increasing importance [7]. In the normal rectum distension by balloon results in relaxation of the internal sphincter and contraction of the external sphincter. When aganglionosis is present rectal distension does not produce relaxation of the internal sphincter. In eight cases of proven aganglionosis such a failure of inhibition of

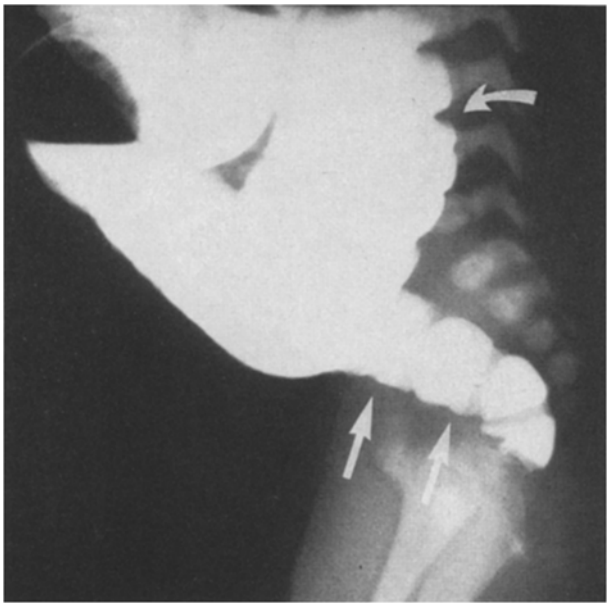


Fig. 6. Late delayed film in a short segment aganglionosis. This film was chosen to demonstrate that the relative narrowing of the aganglionic area (straight arrow) is often difficult to evaluate and may sometimes only be appreciated when compared to the dilated ganglionic area (curved arrow)

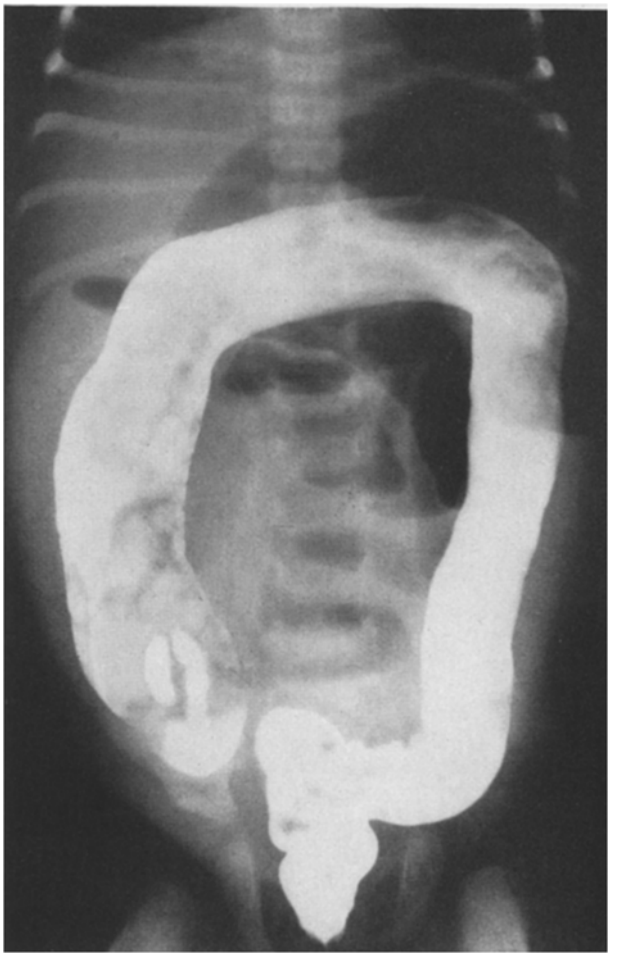


Fig. 8. Total aganglionosis. The enema appearances were not appreciated before the infant was referred to hospital. This film was taken six days after the initial examination, it shows small bowel fluid levels and retention of barium. The colon is not 'shortened' though the normal sigmoid redundancy is not visible. The child subsequently died from enterocolitis and perforation

the internal sphincter was recorded in only five instances: however, this represents a limited experience, but already further use of the technique seems likely to increase its value.

D. Histological Studies

These provide the definitive method of diagnosis. The material for study may be either a full thickness muscle wall specimen or a suction biopsy. In all cases criteria for diagnosis was complete absence of ganglion cells, the situation regarding immature cells still being subjudice [5].

←
Fig. 7. Total aganglionosis. Obstructed fluid levels are present in the small intestine. There is retention of barium in a 'shortened' colon in this film taken three days after the initial examination

Such material for histological study was obtained at least 2 cm above the mucocutaneous junction for distal to this level ganglion cells are normally absent or scanty. Material for histological study was often taken from near the site of colostomy and initial frozen sections were studied during the operation.

Discussion

In the patient, especially the infant, in whom a diagnosis of aganglionosis is suspected certain other possible diagnoses require consideration and exclusion. Among such conditions are anorectal malformations [8, 9], ileal atresia, volvulus, meconium ileus, inspissated milk syndrome [11, 12], ileus with septicaemia and severe gastroenteritis, and, necrotising enterocolitis [1, 2]. Important features in these considerations have recently been presented [10].

A meconium plug type of obstruction is commonly associated with aganglionosis, so that when this condition is diagnosed a repeat enema examination will often be required. Other causes for meconium plugs during this period were meningitis and hypothyroidism and idiopathic.

Careful clinical assessment of the patient leads to the radiological investigation. Plain film appearances of obstruction may be found and the dilated intestine proximal to the aganglionic segment may suggest the diagnosis.

A reliable barium enema technique is imperative, but there is no reason why such diagnostic studies should not be made in general hospitals where a radiologist trained in paediatric radiology can perform the study. By defining the transition between dilated proximal bowel and narrow (aganglionic) distal bowel the diagnosis can be established and the possible site for the commonly performed colostomy can be indicated.

The initial filling with barium must not be too rapid for the narrow segment may be dilated by this, and, the diagnostic evidence may then disappear. The transitional zone may be quite short and show a finely notched rippled contour [16]. The proximal dilatation takes time to develop and the intestine may be no longer dilated if evacuation has occurred for any reason (spontaneous, rectal examination or colonic lavage). Generally about 20 ml of barium have been required to define the aganglionic segment, transitional zone and the dilated segment proximally. Overfilling of the gut must always be avoided and especially in total aganglionosis of the large intestine for complete reflux through the small intestine has been recorded [6].

It may not be possible to wait for the delayed films, particularly in patients with complicating enterocolitis, because of the urgency of the clinical situation. Abnormal churning and writhing contractions may be seen at fluoroscopy in the aganglionic segment.

After studying the videotape and kinescope recordings of enemas carried out over the last 3 years of this 8 year period it is clear that there are limitations concerning the question of the narrowed aganglionic segment, transitional zone, the dilated segment and the barium retention in delayed films [2, 4, 10, 14, 16, 17, 20, 22]. Furthermore overall shortening in patients with total aganglionosis is a debatable feature [3, 13, 15] for it may not be apparent or it may be seen only in the late delayed films (Figs. 7, 8).

Detailed histological discussion is beyond the scope of this paper, but we wish to make two statements. We do not believe in either skip areas or the concept of an ultra short segment. As radiological diagnoses, skip areas are not acceptable as a developmental possibility and hypoganglionosis is normal in the region of the internal anal sphincter. Radiographically it is not possible to make either of these diagnoses in the neonate.

Manometric studies are a relatively recent development and at the time of completing this series our own experience was limited. However, such studies will find an increasingly important place in the future evaluation of patients [18, 24, 7]. Manometric studies, suction biopsy studies and radiological studies are becoming essentially complementary techniques for establishing the diagnosis of aganglionosis. However, radiological methods provide evidence which can indicate not only the diagnosis but also the level to which normal innervation of the gut has occurred.

In the successful management of the patient who is suspected of having aganglionosis the coordination of the efforts of clinicians, radiologists and pathologists is of paramount importance. The increasingly early age at which the condition is being recognised serves to emphasise this.

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