Case 60 A very constipated small boy

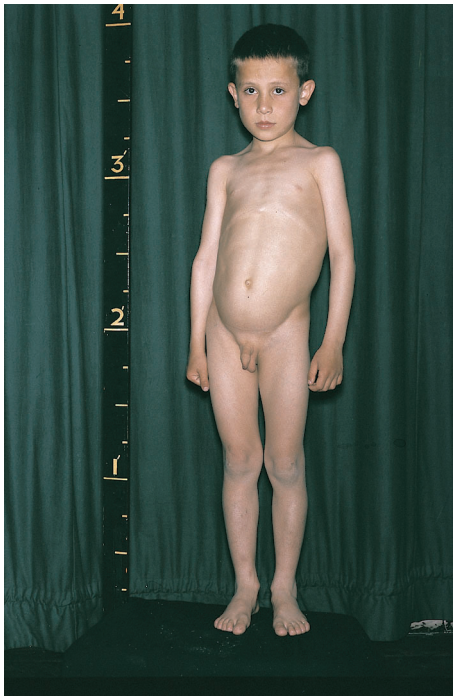

Figure 60.1

This is a boy of 15 years, who looks much younger, and is much smaller than his chronological age would suggest. He was brought to this country from overseas for treatment. His parents gave a history that his abdomen had appeared distended ever since he was a baby. He has been constipated all his life; he might go days or even a week without a bowel movement. His parents had tried a whole range of aperients without success and now his aunt, who was a trained nurse, was giving him an enema once a week. This usually produced a constipated stool. There was no blood seen in the motions at any time.

On examination he was a very bright, intelligent boy, who attended school with children of his own age and easily kept level with them in his school subjects. The abdomen, as can be seen in Fig. 60.1, is grossly distended but not tender. On rectal examination, the anal canal was normal and empty, but a mass of hard faeces could be felt at the tip of the examining finger. The testes were in the normal scrotal position, but he was pre-pubertal, with no genital, facial or axillary hair.

What is your clinical diagnosis? Give both the scientific name and the eponym by which it is commonly called.

Congenital megacolon or Hirschsprung's disease.*

What is the pathological basis of this condition and how is this pathology demonstrated in the laboratory?

There is faulty development of the parasympathetic innervation of the distal bowel in that there is an absence of the ganglion cells of the plexuses of Auerbach† and Meissner‡ in the rectal wall. This defect sometimes extends into the distal colon and may rarely affect the whole of the large bowel. The involved segment of intestine is spastic, with gross proximal distension of the large intestine above the narrowed segment. The diagnostic test is to take a biopsy of the rectal wall, which must include muscle; complete absence of the ganglion cells is confirmed.

*Harald Hirschprung (1830–1916), Professor of paediatrics, Queen Louisa Hospial, Copenhagen.

†Leopold Auerbach (1821–1897), Professor of pathology, Breslau.

‡George Meissner (1829–1905), Professor of anatomy, successively in Basle, Freiberg and Gottingen.

What is the sex distribution of this disease?

For some unexplained reason, 80% of these patients are male.

What important differential diagnosis must be made in these cases?

Acquired megacolon. This is a condition of severe constipation which usually commences when the child is 1 or 2 years old; many are mentally subnormal. Rectal examination in these patients is typical – impacted faeces are present right up to the anal verge. If necessary, a biopsy is performed to clinch the diagnosis; normal ganglion cells are present. This condition responds to treatment with regular enemas and aperients.

What are the radiological findings in patients with congenital megacolon?

A plain X-ray of the abdomen shows dilated loops of large bowel in which faecal masses may be seen. A barium enema demonstrates the characteristic narrow rectal segment, above which the colon is dilated and full of faeces.

Outline the treatment of this condition

If a baby is obstructed in the neonatal period, a colostomy may be necessary. Elective surgery should usually be carried out when the infant is 6–9 months old or until 3 months or more have elapsed from establishment of a colostomy. The aganglionic segment is resected and an abdomino-perineal pull-through anastomosis established between normally innervated colon and the anal canal. It is very important at the time of surgery to ensure, by frozen section histological examination, that ganglion cells are present in the remaining colon.

This procedure was indeed carried out successfully in this young patient, but earlier surgery would have saved him years of misery and would probably have allowed normal development to have taken place.