21.1 Introduction

Disorders which mimic the distal colonic obstruction typical of Hirschsprung’s disease are described under a number of terms including adynamic bowel, pseudo-Hirschsprung’s disease, megacystis-microcolon hypoperistalsis syndrome, and visceral neuropathies and myopathies. They may be caused by a wide variety of different disorders including conditions which are intrinsic to the enteric neuromusculature as well as disorders where the environment in which the neuromusculature operates is abnormal such as in inflammatory conditions. Most of these diseases have their effect by disrupting the normal control mechanisms of the motor apparatus of the gut. The normal patterns of contraction of the muscle coats of the gut are dependent upon the control mechanisms which act upon the smooth muscle cells of the muscularis propria at many different levels to produce the required effects. The control mechanisms consist of properties of the smooth muscle cells themselves, an intrinsic network of nerves, the enteric nervous system with multiple neurotransmitters modulated by extrinsic nerves, paracrine and endocrine hormones, and other neuromuscular active compounds.

If a clinical condition which mimics Hirschsprung’s disease is considered using this mechanistic framework then the cause of the patient’s symptoms may often be readily understood. In some there may, however, be obstruction of the anus or rectum due to congenital anorectal anomalies, tumors of the anus and rectum, e.g., leiomyoma, hemangioma or external compression of the anorectal area.
21.2 Clinical Presentation

The disorders may present with either the primary or the secondary effects of the underlying condition. These may result in either functional obstruction or severely delayed transit of intestinal contents. A patient may thus complain of severe constipation, acute or chronic urinary retention, a distended painful abdomen or even vomiting. In addition, some further effects of these episodes might include fecal impaction, adhesional obstruction associated with previous surgery, and episodes of bacterial overgrowth. In those conditions in which there is an underlying neuropathy of the gut, the consequences of denervation may not only be on motor activity but also upon intestinal secretion and sensation. Visceral hyperalgesia may be one consequence of a denervation hypersensitivity produced following severe drop-out of enteric neurons. In those conditions in which there are developmental defects of the enteric nerves or muscle layers, children present most commonly either in the neonatal period or under the age of 1 year, whereas children with acquired disease present in later childhood [1, 2].

21.2.1 Antenatal and Neonatal Period

Some may be recognized before birth with dilated loops of bowel or a distended bladder or both on sonography, particularly where the mother suffers from polyhydramnios. In the neonatal period the commonest presentation is failure to pass meconium with constipation dating from within a few days of birth. In others, the abdominal symptoms may be more generalized with, in addition to the failure to pass meconium, abdominal distension and bilious vomiting.

In our experience all those who, in addition to gastrointestinal symptoms, fail to pass urine or have recurrent urinary tract infection secondary to poor bladder contractility, hydroureter or hydronephrosis have disease of both the enteric and urinary musculature [2, 3].

21.2.2 Infancy and Later Childhood

The majority of children presenting in later childhood present because of acquired disease. Some patients with congenital defects of the enteric neuromusculature may present outside the neonatal period but mostly do so with similar symptoms to those presenting earlier [2]. Hypothyroidism may first have its effects during the first year of life, but it is unusual for other acquired disorders to be seen at this time. After the first year of life and in later childhood, whilst children may present as they do in infancy or in the neonatal period, often the initial presentation may simply be for constipation. In some, particularly those who have an inflammatory disorder affecting the myenteric plexus, severe abdominal pain may occur as visceral hyperalgesia consequent upon denervation hypersensitivity responses. In others, symptoms may mimic an acute abdomen. Bowel sounds may be totally absent or markedly reduced, but others will have high-pitched bowel sounds more likely to be due to a mechanical rather than a functional obstruction and clear differentiation is required.

21.3 Disorders Causing Pseudo-Hirschsprung’s Disease

Disorders which mimic Hirschsprung’s disease cause disturbance of the control mechanisms of the smooth muscle coats. Thus the disorders and disease may be primarily of the intrinsic enteric nerves with or without involvement of the extrinsic autonomic nerves or central nervous system, the smooth muscle cells themselves, and of the hormonal and endocrine environment. Primary diseases of the gut motor apparatus are considered under two headings, Enteric Nervous System Disease (Section 21.4) and Disorders Affecting Intestinal and Urinary Smooth Muscle (Section 21.5). Secondary causes of pseudo-Hirschsprung’s disease consist of a variety of diseases and drugs, and these are listed in Table 21.1.

21.4 Enteric Nervous System Disease

Disease of the enteric nervous system may be familial and limited to the colon or be part of a more diffuse disorder affecting the whole gut or as part of a familial peripheral and autonomic neuropathy such as familial visceral neuropathy. The commonest disorders mimicking Hirschsprung’s disease are those in which there are malformations of intestinal neurons as in intestinal neural dysplasia, intestinal ganglioneuromatosis, MEN 2a and 2b, and hypoganglionosis. All of these conditions are considered in Chapters 8, 9 and 10 and are not considered further here.

21.4.1 Intestinal Ganglionitis

21.4.1.1 Idiopathic Lymphocytic Intestinal Ganglionitis

This condition may present with the sudden onset of acute severe constipation usually in late childhood. By the time that investigation takes place the appearance is often that of aganglionosis in the rectum. In two patients studied by the author the process seemed to start in the rectum and gradually ascend the gut [4]. In one patient in whom full-thickness biopsies over a number of years were available for study the condition could be shown to