lack of peristalsis, there is a loss of the rectosphincteric reflex. Normally, when a stool bolus enters the rectum, the internal sphincter relaxes and the stool is evacuated. In Hirschsprung disease, the internal sphincter does not relax. In most cases, the aganglionic segment includes the rectum and some portion of the distal colon. However, the entire colon or part of the small intestine may be involved; this is considered long-segment Hirschsprung disease. Occasionally, skip segments or total intestinal aganglionosis may occur. Rarely, total colonic aganglionosis, in which there is no innervation of the large and small intestine from the anus to the ileocecal valve, will occur in 2% to 13% of cases with Hirschsprung disease (Moore, 2012).

Diagnostic Evaluation

Most children with Hirschsprung disease are diagnosed in the first few months of life. Clinical manifestations vary according to the age when symptoms are recognized and the presence of complications, such as enterocolitis (Box 22-1). A neonate usually is seen with distended abdomen, feeding intolerance with bilious vomiting, and delay in the passage of meconium. Typically, 99% of normal term infants pass meconium in the first 48 hours of life, but fewer than 10% of infants with Hirschsprung disease do so (Gourlay, 2013).

Box 22-1

Clinical Manifestations of Hirschsprung Disease

Newborn Period

Failure to pass meconium within 24 to 48 hours after birth

Refusal to feed

Bilious vomiting

Abdominal distention

Infancy