Chapter 2
Functional Disorders of Elimination

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**Introduction**

Disorders of Defecation include inadequate evacuation of stool and/or age-inappropriate fecal soiling. Children with these symptoms may have abnormalities in the anatomy or physiology of the ano-rectum and/or psychological or developmental impediments to normal toileting.

About 25% of patients referred to pediatric gastroenterologists suffer from disorders of defecation [1–3]. Functional disorders are 50–100 times more prevalent than organic disorders of defecation [4]. About 2% of all children are troubled with fecal soiling at some time during primary school.

The following is a review of the physiological anatomy of the apparatus of defecation and fecal continence, the developmental process by which toileting skills are acquired, problems of disordered pelvic floor motility, followed by a review of functional disorders of defecation, their differential diagnoses, and their management.

**Physiologic Anatomy of the Apparatus of Defecation and Fecal Continence [5–7]**

The physiological apparatus that permits sensing, discrimination, withholding, and controlled evacuation is comprised of the anus, pelvic floor, rectum, colon, abdominal muscles, diaphragm, and glottis. It can be understood in terms of sensory and motor functions. (Figure 2.1)
Sensory Aspects

The urge to defecate has visceral sensory and somatic sensory components. The **visceral sensory component** is mediated by tension receptors within the colo-rectal wall and within the surrounding pubo-rectalis portion of the levator ani muscle at the level of the ano-rectal junction. Stool or gas entering the rectum distends it, thereby increasing the mechanical tension within the rectal wall and the surrounding pubo-rectalis muscle fibers [8] where it is appreciated as the urge to stool. A sense of pelvic fullness progressing to colicky pain is felt when a progressive stretch stimulus is applied 15 cm or more above the anus [9]. A feeling of pressure on the perineum and a sense of impending defecation are felt when distention is applied closer to the anus [10]. Mechanical tension (and therefore afferent sensory excitation) is heightened when stretch is applied rapidly rather than gradually [11]. Therefore, the sensation of fullness may progress to intense pain when the colo-rectum is rapidly filled with air or fluid, as during an enema or acute diarrhea. By contrast, the gradual rectal distention caused by chronic stool withholding is associated with comparatively less rectal pain or discomfort, although acute colicky pain can occur during bouts of active gut wall contractions in a patient with a partially obstructive intra-rectal fecal mass [12].
The **somatic sensory component** of the urge to defecate involves receptors for touch, pain, temperature, pressure, and friction located within the anoderm [5, 13]. When stool or flatus comes into contact with the upper end of the anoderm, nerve impulses traverse the somatic sensory fibers within the pudendal nerves en route to the CNS where imminent incontinence is appreciated. Anal sensory receptors allow the discrimination of flatus vs. solid stool [14, 15].

**Motor Aspects**

Fecal continence results from resistance to outflow created by two muscle groups: the smooth muscle of the *internal anal sphincter (IAS)* and the striated muscles of the pelvic floor and *external anal sphincter (EAS)*. The IAS is the thickened terminal portion of the smooth muscle of the rectal wall. It extends part way down the anal canal and is surrounded by the EAS. The relation of the two sphincters can be likened to a funnel within a funnel. The caudal limit of the IAS can be felt by digital palpation of the *inter-sphincteric line* part way up the anal canal. The IAS is innervated by the enteric nervous system and is not under voluntary control. Its principal reflex activity is relaxation. It contributes 5% of the occlusive pressure of the anal canal at rest [16, 17]. The striated muscle of the EAS, levator ani, and the rest of the pelvic floor is innervated by the pudendal, third and fourth sacral nerves. They are under voluntary control, tend to act in unison, and provide the principle barrier during voluntary withholding when continence is threatened.

The rectum exits the pelvis via the anal canal which passes thru an elliptical opening in the mid-sagittal plane of the levator ani muscles at the level of the recto-anal junction [5]. Simply stated, the pubo-rectalis portion of the levator ani and upper part of the EAS are a unit, anatomically and functionally [18]. The pubo-rectalis is a U-shaped sling that originates from the posterior surface of the pubic bone and extends along the sides and back of the ano-rectal junction (Fig. 2.2). This sling pulls the ano-rectal junction forward and upward creating the *ano-rectal angle* which keeps stool from entering the anal canal. When imminent but unwanted incontinence of stool or flatus occurs, the sling contracts above the level of its resting tension (“squeeze”) thereby pulling the upper end of the anal canal further.

![Fig. 2.2 Mid-sagittal diagram of the pubic bone, ano-rectum, and ano-rectal angle with the pelvic floor: at rest; during maximum contraction to prevent unwanted defecation (“squeeze”); and during defecation](image-url)
forward and upward, making the ano-rectal angle more acute, elongating the anal canal, and occluding its lumen. During normal defecation, the sling relaxes below its level of resting tension allowing the ano-rectal angle to become less acute, the anal canal to shorten and its lumen to more easily open, thereby lessening the mechanical resistance to passage of stool or flatus [5, 18, 19].

**Reflexology of the Colo-Rectum, Anal Sphincters, and Pelvic Floor**

Although the pelvic floor and EAS muscles can be contracted and relaxed voluntarily, the motor activities that permit continence and defecation also involve reflex activity of the colon, rectum, anal sphincters, and pelvic floor [14, 15]. The passage of chyme from stomach to duodenum induces mass movement within the colon which propels stool into the rectum. Accumulation of withheld stool in the rectum slows propulsive motility in the right colon and sigmoid [20] and, in at least one instance, has been shown to result in retrograde movement of recto-sigmoid contents back up to the distal transverse colon [21]. In addition to its effects on colon motility, rectal distention slows gastric emptying and small bowel transit [22]. The appetitesuppressive effect of delayed gastric emptying and the hardening of stool that results from slow transit and rectal stasis may contribute to the anorexia in children with fecal retention and the return of hunger following evacuation of a retained fecal mass.

**Recto-Sphincteric Reflexes**

The *sampling reflex* [15], also known as the recto-anal inhibitory reflex, is activated when stool or flatus enters the rectum in the amounts sufficient to stretch (i.e. increase mechanical tension within) the rectal wall. This causes a reflex relaxation of the internal anal sphincter lasting 8–20 s [23]. It permits stool to descend into contact with the upper anoderm, stimulating its tactile receptors, causing conscious awareness of imminent incontinence and permitting discrimination between stool and flatus [15]. This reflex is intrinsic to the ano-rectum and is not dependent on an intact spinal cord or extrinsic innervation.

**The Rectal Inhibitory Reflex** [9]

The *rectal inhibitory reflex* is felt as an uncontrollable urge to stool caused by massive rectal filling, such as occurs with high volume diarrhea or rapid administration of an enema. It can be understood as a culmination of progressive increases in the sampling reflex. Each time a substantial amount of stool enters the rectum causing the IAS to relax, continence is preserved by contraction of the pelvic floor and EAS. Continued rectal loading causes more prolonged, deeper relaxation of the IAS and
contraction of the pelvic floor [16, 24]. Rectal loading can reach a point at which the IAS relaxation persists and the pelvic floor and EAS reverse their usual response; instead of tightening, they relax and their ability to preserve continence lapses [25]. The rectal inhibitory reflex loop is viscero-spinal, demonstrable in normal and paraplegic subjects. It probably is a major factor in “retentive posturing” behavior that is a feature of the functional fecal retention syndrome [26].

**Pelvic Floor Reflexes**

The muscles of the pelvic floor differ from most other skeletal muscles in that they contain not only “fast twitch” fibers which produce rapid, phasic contractions characteristic of skeletal muscles, but also “slow twitch” fibers which produce sustained contraction over relatively long periods [27]. Pelvic floor muscles remain partially contacted at rest and during sleep. They contract above their level of resting tension during continence-preserving actions and relax below resting tension during defecation [9].

**The Postural Reflex [9]**

The tonus of pelvic floor muscles results from a spinal reflex involving spinal segments below L-2. Waxing and waning contractility occurs during sleep and wakefulness. For example, entry of stool into the rectum may trigger IAS relaxation (the sampling reflex) that escapes conscious awareness, but nevertheless, causes contraction of the pubo-rectalis, which may also go unnoticed [28]. Indeed, except for the intention to defecate, micturate, or give birth, all activities that increase intra-abdominal pressure are accompanied by reflex contractions of the pelvic floor above the level of resting tension; talking, coughing, lifting—all elicit this unnoticed reflex.

**The Continence-Preserving Reflex [9]**

Events that create a sudden awareness of imminent incontinence trigger the *continence-preserving reflex* [9]. A sudden urge to defecate or urinate causes contraction of the pelvic floor. However, the levator ani fatigues after about a minute [5] and returns to its level of resting tension. When this occurs, a fecal bolus may either descend and remain in contact with the anoderm or it may remain higher in the rectum, out of contact with the anoderm. When the fecal bolus remains out of contact with the anoderm after levator relaxation, the anus’ somato-sensory component of the defecatory urge ceases (and, by the young child’s magical thinking, “it goes away”).

Whether or not this response to a sense of imminent incontinence is an inborn reflex or voluntary act is controversial [29]. Before toilet training, it is presumed that infants do not inhibit defecation or urination [30]. Toilet training induces a conditioned response to eliminative urges for the purpose of maintaining continence.
until the child can get to a toilet. Therefore, it seems reasonable to assume that the usual response to the urge to eliminate is a pelvic floor, continence-preserving reflex acquired during toilet training. It is also subject to voluntary control during defecation or micturition [31].

The Closing Reflex

The closing reflex is another stimulus-induced contraction of the pelvic floor above its level of resting tension [32]. It expels stool passing through the anal canal during defecation. Like continence-preserving reflexes, it is “automatic,” but also subject to voluntary control. Sensory input for the closing reflex originates in the somatosensory receptors of the anoderm. It is likely that visceral afferents from the rectum are also involved because the pubo-rectalis and EAS immediately contract when an inflated balloon within the rectum is suddenly deflated, suggesting that the closing reflex can occur when contents within the rectal lumen suddenly shrink.

The Cutaneo-Anal Contractile Reflex (“Anal Wink”)

Reflex contraction of the pelvic floor and EAS is triggered when a tactile or painful stimulus, e.g., an examining finger or pin prick, is applied to or near the perianal skin or anoderm [9, 23]. This motor response is the same as that of the continence-preserving reflex. Although the reflex action elicited in both cases is the same, the two reflexes serve different functions: the stimulus for the continence-preserving reflex is the threat of incontinence caused by increased intra-rectal pressure and stimulation of the anoderm; by contrast, the cutaneo-anal contractile reflex is nociceptive, a protective response to actual or anticipated anal penetration or trauma. Both reflexes can be elicited in patients with spinal cord transections above the sacral level.

The Development of Toileting Skills

In order to understand aberrant toileting, it helps to appreciate the skills inherent in normal control and the processes by which they are achieved [33].

A normal infant has no difficulty passing stool or urine because he or she is naïve of any need to exercise control. By contrast, a five-year-old child is able to perceive the urge to eliminate, suppress the impulse to immediately void, disengage from play, find a bathroom, insure privacy, unfasten clothes, climb up onto the toilet, initiate passage of stool or urine, recognize when it is done, dismount, clean up, refasten clothes, unbolt the door, and emerge to successfully resume play [34]. This ordinary, taken-for-granted behavior is a result of perceptual-motor developmental skills that take months or years to develop [35, 36]. They are acquired by two simultaneous processes: toilet training, what parents do to help their children towards
socially appropriate, self-sufficient toileting; and *toilet learning*, what children think and do while learning the mores of eliminative behavior, recognition of sensory signals, and achieving control that enables them to choose to either void or retain urine or stool [37].

Beginning toilet training prior to 27 months of age does not benefit the child or the parent because it does not lead to earlier mastery of toileting skills [38]. Age-appropriate toileting is acquired through a process analogous to learning how to ride a two-wheeled bicycle. The parent can teach or “train” the child to ride by giving advice, assistance, and encouragement, but the parent cannot implant the skills of balance and locomotion in the child and the child cannot acquire the skills by passively receiving them. He or she has to learn them by repeated attempts at “getting it to work.” The attempts require a desire and ability to learn, courage, and the absence of emotional or cognitive problems severe enough to hinder the learning process.

Babies are born with instinctual sequences of behavior that are evoked by specific stimuli from their outer or inner environments, such as the neonate’s sucking response to an insertion of a finger into its mouth or the defecation response caused by contact of stool or a foreign body into the anus. These activities require no learning; they are present at birth.

With maturation and experience, motor activities become more and more purposeful. The newborn exists without a sense of separateness of its surroundings [39]. Development within the nurturing environment leads to the emergence of a sense of self and an awareness of the existence of others as separate centers of initiative. The older toddler and preschool aged child are inquisitive, but not able to think rationally. The psychiatrist and student of early childhood, David A. Freedman, wrote, “Typically, toddlers are not competent to understand why particular expectations or prohibitions are being imposed upon them… Most often a toddler can only know whether he or she ‘got it right’ after the fact, as a consequence of the effects of the behavior. For the toddler, who is just beginning to find his way as a separate center of initiative, this means learning to behave ‘correctly’ as defined by the strictures of parental authority. Often, ‘correctness’ is only known from the parents’ response to the child’s behavior. The youngster must, in this regard, always be playing catch-up” [40].

During the second and third years, parents place expectations on their child in activities such as toileting, self-feeding, and the ability to wait for help and attention. Normal parenting enables the child to strive to accomplish these goals. Each success entails the child’s pride of accomplishment. The infant’s tolerance and enjoyment of messiness is superseded by an increasing desire for order and cleanliness reinforced by admiration and approval from its parents. However, progress isn’t always easy. Incorrect behaviors that evoke parents’ disapproval threaten the child with perceived loss of the parent’s love, presence, or trust. Parents, for their part, assume responsibility for the success of their child in accomplishing self-control and they are vulnerable to feelings of anger, self-reproach, and guilt when their child-rearing efforts seem to fail. First-time parents may be more susceptible to self-doubt.
Parents’ regulation of their child’s eating, toileting, and sleeping is prone to difficulty. In contrast to the parents’ ability to foster social skills (e.g., consideration of playmates’ desires and rights) and safety practices (e.g., not running into traffic), attempts to regulate the child’s chewing and swallowing of food, the child sensing the urge to urinate or stool and exercising sphincter control, and getting a child to fall asleep—these are bodily functions which ultimately are entirely within the control of the child. For example, parents can oblige their child to sit at the table, but they cannot force their child eat; attempts to do so may result in oral defensive behaviors that may be too intense for the child to prevent, such as gagging or vomiting. Parents can oblige their child to sit on the toilet or force him to undergo an enema, but they cannot operate the child’s apparatus of retention and voiding. Noxious measures may succeed for the moment but they may increase the child’s fear of the defecatory urge and may intensify the withholding response to it. Parents can oblige their child to stay in his or her crib at bed time, but they cannot make the agitated toddler feel sleepy and lie down rather than standing up, holding on to the top of the side rail, fussing, or fighting sleep in some other way.

If conflict develops over eating, toileting or falling asleep, and if the preschooler who is in the process of acquiring autonomy and control resists parental guidance, and if his resistance brings punishment, he may respond with deepening obstinacy. A self-perpetuating impasse may develop with intensifying parental frustration and an increasingly anxious, noncompliant child.

Therefore, advising parents to somehow “make” their child eat, stool, or fall asleep may burden them with an impossible task, one likely to increase conflict between them and their child. No physician, parent, diet, or medication “cures” a functional disorder of defecation. Only the child can learn to overcome it. Optimal management consists of sensitive, empathic use of measures that facilitate the child’s efforts, such as orally administered stool softeners for children who have painful constipation, clarity regarding society’s expectations of individuals’ toileting practices, and avoidance of measures that heighten the child’s fears and stubbornness, such as coercive rectal interventions or attempts at imposing the parents’ dominance and their child’s submission regarding control over eliminative functions.

Clinical experience suggests that functional disorders of defecation are not so much a result of parents’ training methods, which are culturally diverse and seldom seem strange or abusive, but rather the result of difficulties the child experiences during toilet learning, such as how the child thinks about and copes with the threat of a painful bowel movement and the shame resulting from fecal soiling [37, 41].

Anxiety and Toilet Learning

Anxiety is an important factor in the pathogenesis of functional disorders of elimination, especially when it affects preschoolers. Three sources of anxiety are common: (1) fear of the dangerous stool within and the anticipation of pain should it come out (discussed below); (2) threats of coercion or humiliation and the retaliatory fantasies they engender; (3) environmental instability, especially perceived
disturbances in parents’ wellbeing, such as a difficult pregnancy, post-partum depression, marital discord, moving to another house. An atmosphere of family disharmony results in tendencies for young children to be less concerned for others, more self-centered, less compliant, and more defiant [42–44]. It requires mental and emotional effort for a child to overcome a functional disorder of defecation and that effort can be disrupted by, for example, autism, attention deficit disorder, anxiety disorders, depression, or oppositional defiant disorder [45]. In any particular child, the difficulty of the therapeutic challenge in functional fecal retention syndrome, for example, is not the size of the retained fecal mass or the diameter of the megarectum that contains it. Rather, it is the psychological comorbidities that may be present [46]. Five-year follow-up studies in children with functional fecal retention syndrome revealed persistence of the disorder in 25–52% of patients. The published rates of success or failure would be more meaningful were the factor of psychological comorbidity identified and elucidated [47–49].

**Piaget’s Childhood Animism**

The first of the above-mentioned three sources of anxiety (fear of the dangerous stool inside) merits further discussion. Piaget described the thinking of children under the age of 7 as animistic [39]. In other words, young children believe that inanimate objects are alive and capable of willful action. “Objects that provoke pain or fear are regarded as doing so from a conscious purpose because the self is still egocentric and in consequence is unable to give a disinterested or impersonal judgment.” As an example, a two-year-old might bump his head on a doorknob and then react by “spanking” the door as though the hurt he experienced was caused by the door’s willful, hostile act.

E.J. Anthony recognized the relevance of childhood animism to toilet learning and the potential for stool withholding [34]. Toddlers and preschool children view their stools animistically; it is common for two-year-olds to wave “bye–bye” before flushing their toilet. One 3–5/12-year-old girl told me that, “the pooh–pooh doesn’t want to come out because it’s cold outside.” A 3–8/12-year-old boy, whose father routinely wiped him after bowel movements, asked his father to leave a bit of stool on the skin near his anus “to help the new poop come out.”

If a young child in the process of toilet learning has never experienced anal pain or frightening events related to defecation, he and his bowel movements “get along.” He does not feel threatened and learns control easily. By contrast, if a child passes a hard bowel movement that causes a minor anal fissure, he suddenly feels pain in part of his body he cannot see, associated with a bodily function he feels is not completely under his control. He cannot control it because “pooh–poohs” have a will of their own and are capable of being nice or nasty. And when one willfully hurts him, he becomes frightened by the defecatory urge and may respond by keeping the stool inside at all costs.
What are some of the implications of childhood animism for toilet training and the treatments of disorders of elimination? First, the style and method of toilet training aren’t nearly as important as whether or not they frighten the child and add to the anxiety associated with mastering control of fecal expulsion. Second, almost all normal children experience rectal intrusions, such as digital examinations or enemas, as hurtful and coercive. Unless it is critically important to perform a digital exam of a child’s rectum (e.g., to diagnose organic anal stenosis or pelvic appendicitis), then it is important to avoid intrusion into the anus of a resistant child, especially a child with a disorder of defecation or a child too young to understand the reasons for the procedure and incapable of cooperation. Third, when a child is afraid to pass stool because “it hurts,” the meaning of “hurts” encompasses more than physical pain. It includes the emotional distress of dealing with something he feels is a threat. Fourth, many children with functional fecal retention syndrome may persist in withholding stool even though they haven’t experienced a painful bowel movement in several weeks because of effective ongoing laxative administration. They continue to respond to the urge by withholding until the entirely soft content of the rectum is so voluminous it can no longer be withheld. It is persistent fear, not physical pain, that perpetuates this retentive behavior. The fear aspect of the problem as well as the mechanical aspect of the problem must be overcome for the disorder of defecation to end.

Pelvic Floor Motility: Coordinated, Uncoordinated, and Dyssynergic

The rectum, uterus, and bladder are all hollow pelvic viscera that communicate with the outside via conduits that pass through the pelvic floor. Resistance to flow through these conduits (anal canal, vagina, and urethra) is controlled by the tonus of the pubo-rectalis portion of the levator ani muscles. When contracted above the level of resting tension, this muscle simultaneously presses these conduits closed [50].

Normal defecation, micturition, and intromission during coitus require the relaxation of the pelvic floor below its level of resting tension. Normally, this relaxation is coordinated with expulsive pressure within the rectum or bladder or, in the case of female sexual intercourse, a state of psychophysiological receptivity. The sense of bladder fullness which prompts a decision to urinate involves the cerebral cortex which, through a conscious act of will, releases inhibition of parasympathetic nerves to the detrusor muscle of the bladder causing it to contract, thereby increasing expulsive pressure. Normally, micturation results from detrusor contraction coordinated with adrenergically mediated relaxation of the smooth muscle of the internal urethral sphincter and somatically mediated relaxation of the striated muscle of the external urethral sphincter and pelvic floor [51].

Pelvic floor dyssynergia (PFD) [52–56] is defined as a pattern of persistent incoordination that interferes with normal emptying of the bladder or rectum but is not
caused by any disease or lesion. PFD is central to the pathogenesis of stool withholding by children with functional fecal retention syndrome and those who go on to develop secondary urinary tract disease.

How does PFD originate and become established? I had two experiences early in my career that prompted me to consider the effects of fear and anxiety on the motility of the pelvic floor. While interviewing the father of a six-year-old girl with massive fecal retention, I attempted to convey the idea that whenever his daughter felt the urge to stool, she anticipated anal pain and that this fear could prevent her from relaxing her bottom so that stool could pass. “Oh, you mean the pucker factor?” he asked. I asked him what that term meant. He explained that he had been a military pilot and that the “pucker factor” referred to a phenomenon that occurred en route home from combat missions. The moment the plane crossed into safe territory, crew members became aware of how tense their bottoms had become, which they then deliberately relaxed.

This prompted me to perform an experiment in which I placed a motion sensor on the anus of a volunteer subject without inserting anything into the anal canal, penetrating the skin, or causing physical discomfort of any kind. The sensor recorded the activity of the pelvic floor. The experimental subject was fully awake, comfortably supine and able to engage in conversation. The experiment revealed: (a) waxing and waning of pelvic floor tone at rest; [57, 58] (b) contraction of the pelvic floor above the level of resting tension whenever my tone of voice or behavior caused mild surprise or apprehension in the subject; and (c) these variations of pelvic floor activity occurred entirely out of the subject’s awareness. These findings supported the hypothesis that emotions influence pelvic floor motility and that this psychophysiologic effect occurred out of the subject’s awareness. Fear increases anal sphincter pressure [59]. PFD is important when managing children with fecal retention; treatment methods should not be frightening and therefore less likely to exacerbate PFD [56]. Biofeedback therapy for PFD aims at bringing dyssynergic reflex activity that is out of awareness into awareness to enable the patient to learn how to make dyssynergic muscles function synergistically [60–62].

To summarize: there are at least five ways in which appropriate, synergistic relaxation of the pelvic floor can be impaired: (1) persistently painful lesions of the anus or perineum causing excessive contraction of the pelvic floor as a result of persistent activation of the cutaneo-anal contractile reflex; (2) the anticipation of pain caused by lesions that become painful during pelvic floor activity, such as an anal fissure that hurts during defecation; (3) anal, urethral or vaginal pain in the past that caused a learning experience that, in the present, inhibits synergistic relaxation of the pelvic floor during defecation, urination or coitus; for example, a child recovering from fecal retention syndrome who suffered severe anal pain during the passage of megastools in the recent past, may be frightened and have great difficulty relaxing his pelvic floor during defecation, even though currently effective stool softening would prevent physical discomfort; (4) fear and anxiety heighten skeletal muscle tone [63], including the pelvic floor, and fear of intrusion into one’s privacy during toileting may impair coordinated pelvic floor motility; (5) past experiences of combined physical and emotional trauma, such as rape involving vaginal or anal penetration,
may result in a persistently dyssynergic pelvic floor [64, 65]. Vaginismus is functional spasm of the muscles surrounding the vagina resulting from a conditioned response to a previous real or imagined frightening sexual experience. It may be analogous to refractory fecal retention in children who have been sexually abused.

**Hinman’s Syndrome**

Hinman described a syndromic form of PFD in which functional fecal retention is accompanied by urinary retention and obstructive uropathy [52–54, 56, 66–68]. Prior to Hinman’s publication in 1973, it was generally thought that obstructive deformities of the urinary tracts in children with fecal retention were caused by crowding of the bladder within the pelvis by the impacted mega-rectum [69]. Therefore, if rectal constipation could seriously damage the urinary system, then serial enemas to remove the fecal mass seemed warranted.

Hinman conceived of a different pathogenesis and a different approach to management. He proposed that the voiding dysfunction was caused by dyssynergia of the bladder’s detrusor muscle and external urinary sphincter (i.e. pelvic floor). In retrospect, the abnormal radiologic findings described by Shopfner [69] (fecal impaction, anterior displacement of the urethra, hydronephrosis, vesico-ureteral reflux) are compatible with simultaneous obstruction of both bladder and rectum. Thus, fecal retention was not the cause of the obstructive changes in the urinary tract; rather, incomplete evacuation of stool accompanied by incomplete voiding of urine had a common cause, namely, the inappropriate anxiety- or fear-induced “uptight” pelvic floor [70–76].

Therefore, if it were important that treatments for functional fecal retention avoid exacerbating the child’s fear and anxiety, then it would be especially important to avoid frightening measures, e.g., serial enemas, in functional fecal retention complicated by obstructive uropathy.

Hinman’s Syndrome should be suspected in any child with functional fecal retention, particularly one who also wets during the day or night. The disorder can have its onset from early childhood to adolescence. Typical ultrasonographic findings are those of chronic proximal urethral obstruction: incomplete emptying of the bladder, bladder trabeculations, vesico-ureteral reflux, and dilatation of renal calyces in the absence of organic lesions that obstruct bladder outflow or neurologic disease. Incomplete emptying of the bladder predisposes to urinary tract infections [77], cysto-ureteral reflux and damaged renal function. As important as it is to keep the rectum clear of fecal impactions, it does not relieve incomplete bladder emptying; and as important as ureteral re-implantation may be for severe cysto-ureteral reflux in Hinman’s syndrome, such surgery won’t cure PFD or prevent the ureterocystic valves from becoming incompetent again [78].

Children with Hinman’s Syndrome are chronically anxious [79]. The condition improves with effective psychotherapeutic and pharmacologic measures that lessen anxiety and decrease resistance to bladder emptying. Table 2.1 summarizes management of Hinman’s Syndrome.
Philosophical Context of Clinical Management of Disorders of Elimination

Before proceeding to a descriptive classification of functional disorders of defecation, it is important to recognize that sincere clinicians have widely differing attitudes as to their role in these disorders. The various attitudes can be viewed on a spectrum, one end of which conforms to the biomedical model of practice which focuses on determining the presence or absence of organic disease, and the other end which conforms to the biopsychosocial model of practice which focuses on illness and all factors which may contribute to its development [85].

The following illustration contrasts the biomedical verses the biopsychosocial approaches using the commonest functional disorder of elimination, Functional Fecal Retention Syndrome (FFRS).

The purpose of the consultation in the biomedical model is to diagnose FFRS by recognizing its clinical features and excluding organic causes. A physician employing the biopsychosocial model is certainly responsible for not missing organic diseases, but goes further in attempting to elucidate the pathogenesis of each child’s stool withholding: how it came to be, as well as the strengths and weaknesses that the individual child and family bring to the task of recovery.

A biomedical view of FFRS is that it is disease-like, with the fecal mass equivalent to a foreign body that deforms the rectum, impairs rectal sensation, and promotes further retention. A biopsychosocial view is that the disorder is caused by a dysfunction having mechanical and emotional factors, i.e., the fecal mass and the child’s fear that it will cause pain and harm if it comes out.

If FFRS is analogous to an organic disease, then the key component of management in the biomedical view, is removal of the foreign body-like fecal mass.
This is typically done with enemas or intestinal lavage. Because the preferred clinical challenge is organic disease, and since most children with FFRS have none, the plethora of such patients in pediatric gastroenterology clinics is commonly viewed as clinical clutter. Following the initial consultation, management may be turned over to a nurse. A doctor isn’t necessary for treatment of an uncomplicated nondisease and his/her time is better utilized for patients with “real,” diseases [33].

By contrast, a biopsychosocial view is that the child has a functional disorder that, by definition, is not analogous to organic disease. Removal of the fecal mass is certainly important, but is best done by helping the child accomplish it for himself. Explaining the mechanics of fecal retention and bypass soiling helps the child comprehend what he needs to do. Avoidance of coercive measures that frighten him can be achieved with effective stool softeners to liquefy the fecal mass to the extent that it cannot be withheld. The physician develops parallel relationships with the child and the parents. The child is listened to. The essential mechanics of fecal soiling and laxative treatment are reiterated in terms the child can understand. The doctor empathizes with the child’s worries about passage of a bowel movement and attempts to reassure the child that, even though it might cause discomfort, it will not damage his/her body. The parents’ fears about physical harm must be elicited and relieved; they need to be reassured that their child is in no danger, even during “retentive crises” (see below). The physician continues to help parents avoid resorting to measures that might stress the child even further and impair his trust in them and his doctor. The knowledge that they can call the doctor during what feels like a crisis is enormously supportive.

With respect to functional disorders of elimination, the type of doctor–patient relationship that is typically utilized in the biomedical model is “guidance-cooperation,” to use the classification described by Szasz and Hollender [86, 87]. The type most useful in the biopsychosocial model is “mutual participation.”

Pediatric gastroenterologists have engaged in heated discussions over the necessity for digital examination of the rectum as part of the evaluation of patients with FFRS [88]. As stated by Doctors Ann Buchanan and Graham Clayden, “In children beyond infancy, it may be very distressing, especially when the constipation is likely to be related to anal pain. Little can be learned from a writhing child who is clenching every muscle to protect him/herself” [89]. “Children who have experienced coercive physical treatment for their constipation such as enemas or suppositories can produce symbols in drawings that are highly suggestive of violation or sexual assault” [89]. Inspection and palpation of the abdomen and inspection of the anus [90, 91] can obviate the need for a rectal examination in most cases.

In the biomedical model, the physician’s first concern is to rule out Hirschsprung’s disease and other pathologies; accomplishing that is more important than rapport with the child, especially if further treatment will be carried out by someone else. By contrast, in the biopsychosocial model, the physician’s first concern is to develop a trusting relationship with the child. The doctor tolerates the initial diagnostic uncertainty caused by not having done a rectal exam so that the child can feel that the doctor is his protective ally and can be trusted during the therapeutic process by which the child overcomes the disorder.

To sum up, until the day arrives when there are data that clearly support the superiority of either the biomedical or biopsychosocial models with respect to functional disorders of defecation, it behooves us to acknowledge that these differences exist.
Diagnostic Techniques in the Diagnosis of Disorders of Defecation (Historical, Physical, and Radiologic)

Diagnosis of the various kinds of disorders of defecation is based upon the presence or absence of three phenomena: Soiling, fecal retention, and organic disease. Historical, physical, and radiologic findings can be misleading. Therefore, each diagnostic tool warrants more precise definition.

Objective evidences of soiling consist of deposits of stool in underwear greater than a smudge (that could be caused by passage of flatus or incomplete wiping after a normal bowel movement) and persistent fecal odor. Because its receptors for touch, temperature, and friction may fail to signal presence of liquids at body temperature, the anoderm can be “fooled” by a small amount of liquid stool, mucus, or oil. Imminent leakage isn’t sensed quickly enough and soiling occurs largely out of the individual’s control. Such soilage is not necessarily abnormal, indicative of diarrhea, “bypass” soiling around the fecal mass, or sensory deficits of the anoderm [91].

Persistent stool withholding results in a mid-line mass palpable trans-abdominally behind and above the public symphysis. After weeks or months of retention, the mass grows upward to the level of the umbilicus and, with further enlargement, can extend to the costal margin. The rectal accumulation causes a mega-rectum which fills the true pelvis and enlarges upward [25], but progressive accumulation of stool does not fill and distend the more proximal colon in patients with FFRS (Fig. 2.3).

The presence of fecal retention can be assessed by abdominal palpation, digital rectal exam, or radiography [92]. The extent of rectal enlargement can be determined by a radiograph of the pelvis. The recto-pelvic ratio (RPR) is the diameter of the rectum divided by the maximum transverse diameter of the pelvic outlet. A RPR above 0.61 is characteristic of a mega-rectum [92].

Fig. 2.3 An enormous megarectum in a 13-year-old boy with FFRS who could not recall defecating during the preceding year. Notice the scybalous stools in the nondilated colon orad to the recto-sigmoid
Abdominal palpation is least intrusive and does not expose the child to radiation. It is best done with the patient supine, knees drawn up, and abdominal muscles relaxed (Fig. 2.4).

It is difficult to palpate abdominal masses through the rectus abdominis muscles. Therefore, place the finger tips of one hand just to the left, and the fingertips of the other hand to the right of the rectus muscles. The finger tips on one hand are the “pusher” and those of the other are the “sensor.” A fecal accumulation can thus be balloted as a firm, movable, nontender mass in the midline, extending upward from deep within the pelvis anterior to the sacrum. (Some children have a prominent fifth lumbar vertebra that might be mistaken for a midline mass, but it is not movable and does not extend down into the pelvis.)

In contrast to the midline mass of intra-rectal stool that accumulates as a result of functional or organic rectal outlet obstruction, the typical finding in Irritable Bowel Syndrome constipation consists of lumps within the distal descending and sigmoid colon palpable in the left iliac fossa. This results from heightened segmenting motility of the sigmoid colon, not from fecal retention within the rectum (Fig. 2.5). Of course, both midline and left iliac fossa findings may coexist. The stools of IBS constipation may scratch or fissure the anoderm during defecation and thereby predispose to functional stool withholding in vulnerable children. However, blurring the distinction between “pellet stool constipation” and “stool withholding constipation” is not helpful, if for no other reason than increased fiber intake helps the former but does nothing for the latter.

It may be difficult to be sure of the presence of a fecal mass that has been recently passed or softened to the extent that it is no longer palpable. Or, the child’s soiling may be of the nonretentive type [93]. In such cases, the response to a few days of osmotic laxative in doses sufficient to liquefy a hard fecal mass will aid diagnosis.
If massive amounts of stool are passed, followed by improved mood and appetite and cessation of soiling, then it is reasonable to assume that the child’s soiling was due to functional fecal retention and management can proceed expectantly based on that assumption.

Digital examination is warranted when Hirschsprung’s Disease is a clear possibility. A narrow anal canal and an empty lower rectum in a child with a palpable fecal mass in the abdomen and/or chronic abdominal distention suggests Hirschsprung’s disease or other organic motility disorder. A limited contrast enema, without prior “clean out,” using a water-soluble contrast medium is warranted if Hirschsprung’s disease is a clear possibility.

Abdominal radiographs should not be used routinely to assess fecal retention because of radiation exposure and the efficacy of the interval history, abdominal palpation, and inspection of the anus for soilage. However, accurate palpation may not be possible through the panniculus of an obese child. In such cases, one radiograph that includes the perineum and entire abdomen is useful to document the diameter of the rectum, whether or not a mega-rectum fills the true pelvis and extends all the way down to the pelvic floor, the extent to which it occupies the abdominal cavity [25], and whether or not there are defects in the sacrum suggestive of neuropathic fecal incontinence.

**Functional Disorders of Defecation Syndromes**

A retrospective review of 395 randomly selected charts of children seen by me with problems of defecation revealed seven syndromic patterns (Fig. 2.6): **Functional Fecal Retention Syndrome (FFRS)**, which I have sub-categorized into established and emerging types; **Functional Nonretentive Fecal Soiling (FNRFS)** [93]; children who had features of both functional retentive and functional non-retentive fecal soiling at various times in their history; children 3–6 years of age...
with age-inappropriate use of diapers, “diaper dependency” (a.k.a. “stool toileting refusal”), which can be sub-grouped as essentially “permitted” vs. “contentious”; and seven patients in the “miscellaneous” group, which included one child with the Infant Dyschezia Syndrome; the remaining six presented with complaints that I could not categorize as conforming to a recognizable pattern or syndrome. What follows is the discussion of the features of each category and concepts of management.

**The Functional Fecal Retention Syndromes (FFRS)**

Painful bowel movements due to, for example, an anal fissure, may cause a child to feel alarm when he or she experiences the urge to defecate. He responds by tightening his pelvic floor to prevent passage of stool until the urge subsides (Fig. 2.7). He has lost, or perhaps never fully mastered, the ability to easily relieve himself of the defecatory urge. Feces accumulate and harden within the rectum and may either be passed with great distress or retained for weeks or months at a time [95, 96].
Withholding in response to the defecatory urge may begin in infancy or later childhood. In its early phase, the predominant behavior may consist of straining at stool and may result in passages of stool as large as a tennis ball or a soda can. However, as the intra-rectal fecal mass enlarges, it becomes virtually impassable. Memorable experiences of anal pain and the normal animistic view of the fecal lump that hurts results in fewer efforts at evacuation. Retentive posturing becomes more apparent. It consists of tightening, rather than relaxing, the pelvic floor and tightening of the gluteal muscles and thighs (Fig. 2.8). These actions

Fig. 2.7  Summary of the sequence of events during the defecation, fecal continence, and the functional fecal retention syndrome [50, 94]
reinforce the withholding action of the fatiguing pelvic floor. A young child may seclude himself behind furniture or in his room as he is fighting the urge to stool. This behavior may be a desperate attempt to secure privacy and avoid the attention of his parents who may become alarmed, intrude and give advice he doesn’t want. Retentive posturing may consist of “heel-sitting,” by which a child may stop to “tie his shoes” and, in the process, occlude his anus by sitting on his heel. Or, the child may stiffen and cross his legs while standing, holding onto furniture to steady himself, or lying prone with his lower limbs held straight and stiff [97].

Increasing retention causes the full clinical picture to emerge [9, 98] (Table 2.2). Fecal masses become palpable in the abdomen. The patient experiences abdominal discomfort and becomes less interested in eating. He is irritable and less playful. “Bypass” or “overflow” soiling may be mistaken for diarrhea [99]. Retentive posturing

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**Fig. 2.8** Three examples of retentive posturing: tightening of the buttocks to reinforce maximal contraction of the pelvic floor in response to the urge to stool

**Table 2.2** Clinical features of the functional fecal retention syndrome in children

- Passage of enormous stools at intervals of one or more weeks
- Obstruction of the toilet by such stools
- Indications of increasing fecal accumulation:
  - Retentive posturing
  - Soiling
  - Irritability and decreased playfulness
  - Abdominal discomfort or pain
  - Decreased appetite
- Dramatic disappearance of these symptoms after evacuation of enormous stools
- Behaviors indicative of patients’ irrational efforts at coping with soiling:
  - Apparent nonchalant attitude regarding soiling
  - Alleged lack of awareness of soilage
  - Hiding of soiled underclothes

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recurs several times a day. After days or weeks, a *retentive crisis* may occur during which the urge to stool becomes overwhelming, causing not only physical distress, but panicky, pain-like behaviors. Typically, parents become alarmed and may rush the child to an emergency room fearing an imminent intestinal catastrophe. A gigantic stool that may be too large to flush is finally passed. Success in passing the *mega-stool* is promptly followed by a dramatic change in behavior. What appeared to be an acute emergency just minutes before changes to relief and composure followed by the return of the child’s appetite and playfulness.

The child probably experienced the retentive crisis as something horrible. Nevertheless, children’s nonrational thinking prompts a withholding response the next time they feel the urge to stool and the cycle typically repeats itself over the course of a week or more. In time, the child becomes overwhelmed by a loss of control. He may attempt to cope with the problem by making believe it doesn’t exist. He may seem unaware or unconcerned about soiling, which frustrates his parents and causes them to suspect a neuropathic loss of the child’s ability to feel the urge to stool.

The child’s apparent nonchalance contrasts with his desperate attempts to conceal his problem. He may clumsily hide his soiled underwear in inappropriate places, such as the floor of his closet, in his underwear drawer, behind his clean clothes or under his bed. (One child put them in his sister’s guitar. Another hid them in the living room under the lid of the piano stool.) The child’s soiling, refusal to use the toilet for bowel movements, and his seeming lack of concern vex his family. Of all the accidents that might trouble the child, fecal soiling evokes the most intense deprecation from peers and parents and damage to self-esteem [34, 100].

**Emerging Functional Fecal Retention Syndrome**

Emerging FFRS is presented to emphasize the importance of prompt and effective treatment of constipation in infants and toddlers, before their initial responses to the urge to stool change from predominantly expulsive to predominantly retentive.

**Case Vignettes**

Case 1: A first-born five-month-old boy passed meconium within the first 24 h of life. He was breastfed for 3 months, during which he passed one mushy stool per day. At 3 months, the patient and his parents visited relatives in a foreign country. The patient did not defecate for 4 days and became irritable during the third week of their trip. A teaspoon of mineral oil was followed, an hour later, by the distressful passage of a firm, wide, blood-streaked stool. He was then given prune juice every day, but again failed to stool for 4 days. He then passed a soft-formed, nonbloody stool with much effort. At 4 months of age the family again traveled out of the country to visit relatives. They returned home 9 days later. Prune juice was discontinued out of concern that the infant might become “dependent” on it. The patient continued to stool every 4 or 5 days and did so lying on his back with knees extended, buttocks tense, back arched (a retentive posture), and face
flushed). Physical exam revealed a robust, socially responsive infant. His abdomen was soft; there was no palpable mass of retained stool. The anus and spine were normal to inspection. The physical exam was otherwise negative. Management included avoidance of rectal intervention and prompt, gentle wiping with a lotion that would not sting if applied to fissured skin, using a soft tissue or cotton ball. The perianal skin was protected with heavy applications of a bland barrier ointment. Stools were kept soft, but not runny, with orally administered lactulose and mineral oil in increasing doses, depending on the number of days he had gone without a bowel movement.

Case 2: A first-born, three-month-old girl was the product of a normal pregnancy and delivery. She was fed cow’s milk formula and passed unformed stool 6–7 times a day. Her perianal skin became irritated. At 1 month, bowel movements were preceded by 20–30 min of inconsolable crying during which she stiffened her body with her legs extended, arms flexed, and “face beet red.” This culminated in a passage of a pasty or loose stool after which her distress resolved within a few minutes. Blood was present on cleansing tissues after some of her bowel movements. Applications of an anti-mycotic steroid cream and digital exams of the rectum failed to relieve her symptoms. Physical examination revealed a healthy-appearing infant. Abdominal palpation was negative, as was the rest of the examination, except for the anus, which had a small tag with an adjacent fissure which oozed blood. Management included avoidance of rectal intervention, maintenance stool softeners, comfortable wiping after bowel movements and ongoing protection of the perianal skin with a bland barrier ointment.

Comment

Both of these infants’ symptoms were prodromal to FFRS. Why should infants only a few months old develop symptoms of emergent functional fecal retention? If a month-old baby experiences anal or perianal pain associated with defecation, the ease with which the infant defecates may be lost. Put in terms of classical conditioning theory, the defecatory urge is a neutral stimulus which induces a reflex response, i.e., passage of a comfortable bowel movement. If anal pain accompanies reflex defecation, the defecatory urge becomes a conditioned stimulus which triggers the conditioned response of emotional distress plus nociceptive reflex contraction of the pelvic floor and other muscular activities that constitute a withholding response. Management of fecal retention, in terms of behavioral psychology, amounts to extinction of the fear/withholding response. This is accomplished over time during which the causes of anal pain are removed by making stools consistently soft and allowing the abraded or fissured anoderm and perianal skin to heal. In addition, noxious stimuli, such as anal dilatation, suppositories, enemas, or thermometers are avoided. The conditioned stimulus (i.e., the defecatory urge) in time reverts to a neutral stimulus as reflex bowel movements are invariably pain free and the conditioned response (distress and withholding) is extinguished.
Management of Functional Fecal Retention Syndrome

What Do Parents Need?

After discovering what the parents have been previously told and their concerns about their child’s soiling and inability to defecate comfortably, I begin with “de-mystification” to address their cognitive needs [101]. This consists of a didactic description of the functional anatomy of the apparatus of defecation and fecal continence. For the purposes of orientation, I begin with a diagram of the GI tract, describe its basic functions, and then focus on the colo-rectum and pelvic floor. I describe the normal sequence of activities as stool enters the rectum, causes the urge to defecate, is withheld, and passed. Using a diagram, I describe how a mega-stool develops and how it effaces the anal canal, thereby preventing the pelvic floor muscles from operating as a sphincter. I explain how their child’s report of “not being able to feel it come out” can be attributed to the inability of the anoderm to sense contact by fluids at body temperature, a normal rather than a pathologic phenomenon.

The focus then shifts to unrealistic worries about fecal retention. Parents often worry unnecessarily that retained stool may accumulate throughout the length of the intestine (instead of mostly in the enlarging rectum [25]) and that it poses the threat of toxic contamination to the entire intestinal tract. Other common worries are that the colon might burst or that the mega-rectum is permanently damaged and may never return to its normal size and function. Parents are usually amazed to hear that children with FFRS have safely gone for months without bowel movements during which nothing passes except seepage. Intestinal perforation rarely, if ever, occurs in otherwise healthy children who withhold stool (even those who have retained stool for so long that their parents cannot remember when their child last passed a bowel movement) and a fecal mass that is palpable up to the costal margin. An analogy with the uterus, another smooth muscle viscus, may be useful for purposes of reassurance; the uterus expands to accommodate its contents and returns over time to normal size after its contents have been delivered. Clinically significant small bowel bacterial overgrowth is not a feature of chronic stool withholding in otherwise healthy children. As uncomfortable as fecal retention may be, it poses little danger, except for those children with pelvic floor dyssynergia (discussed above) and girls with recurrent urinary tract infections due to the ascent of bacteria from a chronically soiled perineum.

Parents are also burdened by the notion that their child’s toileting skills are mostly created by their efforts at toilet training, with little consideration given to the child’s toilet learning process. They may doubt their competence as parents. They also suffer embarrassment caused by their child’s soiling in school and in public. Their embarrassment is reality-based. The emotional strain on the child and his family may result in angry and perhaps abusive interactions. Nevertheless, a punitive reaction by parents may lessen the child’s willingness to do the many little acts of courage necessary for recovery, such as taking medication or making efforts to relax his pelvic floor during defecation or doing so on the toilet rather than in his underwear while hiding.
Since laxatives are a mainstay of management, parents need some understanding of how they work. The safety of long-term administration of osmotic laxative needs to be contrasted with the routine use of detergent or stimulant cathartics [102]. Whereas long term daily use of stimulant laxatives may induce tolerance (which can be misinterpreted as unwanted habituation and dependency), osmotic laxatives such as polyethylene glycol do not [103]. They may prefer feeding their child a high fiber diet as a healthier means of achieving laxation. Many dietary measures are harmless [104], provided they don’t cause conflict with their child around eating. However, they are ineffective in melting-down an established hard fecal mass within the rectum; water is required for that and the easiest way to bring water into the rectum is with an orally administered osmotic laxative, such as polyethylene glycol.

Parents are helped to react to their child’s toileting problems with less frustration if they see his difficulty in terms of his sense of desperation and loss of control. Lecturing a child with fecal retention on the reasonableness of his passing of bowel movement will not relieve his fear. Helping parents empathize with their child helps them become less anger-prone and better able to facilitate progress [105].

Parents may harbor fears that have little or no scientific basis and should be assuaged. Although bouts of abdominal cramping may become severe, even to the extent of causing reflex vomiting on rare occasions, colonic obstruction of a surgical nature almost never occurs, regardless of how much stool has accumulated, provided that there is no organic disorder such as aganglionosis or intestinal pseudo-obstruction. A dilated rectum does not prevent effective defecation, provided the contained stool is liquefied or at least softened to a pasty consistency. The main impediment to the passage of soft stool is inappropriate contraction of the pelvic floor, not failure of sensation or motility of the stretched rectal wall.

How much laxative is needed depends on how soft stools have become. The best indication of stool softness is its diameter when passed. Spherical or columnar stools wider than about an inch are too firm. If stool is soft enough, through and through, it will be extruded with a diameter that will not stretch the anus to a painful extent. Regardless of how soft stools appear on their surfaces, stools wider than normal may cause anal pain when passed [106].

What Does the Child Need?

Let it be said that no doctor, medication, or parent “cures” FFRS. The cure can be accomplished only by the child, the only person in control of his or her body. The physician, parents, school teacher, and other caregivers function as helpers to the child [107]. They cannot “fix” the problem without the child’s motivation and initiative. Although behavioristic concepts of positive and negative reinforcements have value, it should be born in mind that overcoming constipation and soiling is its own best reward. Bribes or punishments may not produce sustained improvement. The offer of a bribe changes the incentive to stay clean from something done to get rid of a problem that damages self-esteem, to something done to acquire a reward. Sooner or later, there may be demands for bigger rewards.
The “positive reinforcement” may stop working as the parents become resentful and feel that they are being taken advantage of.

Parents’ concern that their child will suffer humiliation may prompt extraordinary measures to shield their child from such painful experiences. To what extent should their child be protected from the humiliation caused by soiling? The danger is reality-based. Deprecation by peers and others who do not owe the child parental love is the consequence of the child’s actions and, as such, may help motivate the child to try to get rid of the problem. An important difference between suffering the consequences of one’s behavior and being punished for the behavior is that the former may be painful, but it does not engender a desire to retaliate the way punishment does. Too often, the effect of punishment is to lessen the child’s motivation to work on his problem; his motivation shifts from feeling bad about his problem to feeling angry towards those who punish him. It is more helpful for the child’s caregivers to sympathetically validate the social dangers of soiling and the inescapable fact that family members and friends tend to react to someone who soils with rejection and avoidance. It should be made clear to the child that, in contrast to his attempts to solve the problem of embarrassment by making believe it doesn’t exist, others cannot go along with this “solution.” Instead, they react as though fecal soiling is an offensive act. Caregivers’ sympathetic but forthright tone serves to get their point across without sounding punitive, thereby maintaining their role as concerned helpers. They should not attempt to ease their child’s embarrassment and self-reproach by minimizing how offensive fecal soiling actually is.

Because, ultimately, the child is the agent of recovery, success in management of FFRS requires an understanding of the child’s point of view. The child’s animistic concept of feces and defecation are of central importance [34]. Children typically fear the “dangerous” stool inside of them. For example, one patient told me that his bowel movement is “a poisonous snake that bites me when it comes out.” Another said, “It’s an alligator.” Another described his stools as “a house” (with sharp gables and steeples). The magical thinking used to cope with such threats is exemplified by the following: “If I can keep it from coming out, then it won’t hurt me.” Another said that he holds it back “until it goes away,” i.e., until the urge subsides. One seven-year-old girl, the child of a socially prominent family, was remarkably articulate, pretty, poised, and tastefully dressed, but she had a fecal mass that extended above her umbilicus as well as an inescapable malodor. In our private chat, I asked her when she last passed a bowel movement. “Oh doctor!” she replied, “I don’t do that any more!” To her way of thinking, her problem had been solved.

Management is based upon gaining an understanding of the pathogenic factors in each case. Although childhood animism is a key factor in the pathogenesis of FFRS, there are other factors to be sought and considered. One of them is the child’s constitutional stool pattern. Some individuals defecate at regular intervals or times of day; others are “irregular” and defecate sporadically [108]. The notion that “irregularity” is not as healthy as a regular pattern of bowel movements is untrue and may cause unnecessary pressure on the child to defecate at times she does not feel the urge to stool because there is no stool in the lower rectum. This may cause performance anxiety or conflict that gets in the way of easy defecation.
A child with heightened segmenting motility in the sigmoid colon may produce firm, lumpy stools at irregular intervals, as is typical of constipation-predominant IBS. Scyballous stools, singularly or in agglutinated lumps, may abrade or fissure the anoderm. The resultant dyschezia may prompt a withholding response caused by anticipated pain. This is common in infants and young children with emerging FFRS.

Temperament refers to stylistic characteristics which are evident in infancy and later on. Infants’ temperaments differ. Some children are characterized by regularity, positive approaches and responses to new stimuli, high adaptability to change, and mild or moderately intense mood which is predominantly positive. At the other end of the spectrum are infants characterized by irregularity in biological functions, negative withdrawal responses to new stimuli, poor adaptability to change, and intense mood expressions which are frequently negative [109]. Parents may interpret fussiness of a temperamentally more irritable infant as physical discomfort caused by the need for a bowel movement. They may then resort to a suppository or enema. Any subsequent quieting may be taken to mean that the infant needed such intervention which, therefore, is more readily administered the next time the baby fusses. A vicious cycle may become established leading to the perpetuation of stooling difficulties in infants and toddlers.

Children with neurodevelopmental problems may have more difficulty mastering toileting skills. Any condition that interferes with the child’s ability to focus attention and effort may impair recovery. Functional disorders of elimination may be comorbidities of attention deficit disorder or autism [110, 111]. Spastic cerebral palsy may interfere with coordination of abdominal and pelvic floor actions that facilitate defecation. Anticholinergic medications harden stools.

Excitement, vacations, or emotional distress within the family are often important when progress is interrupted or regression occurs. The dilution of parental attention when a new baby is brought home may cause a regression in a young child’s toileting. A not infrequent problem arises when the child of separated parents spends part of the week with one parent, the rest of the week with the other and the parents have conflicting attitudes about their child’s disorder and the prescribed treatment regimen.

An empathy gap may interfere with parents’ ability to understand the challenge faced by the child. Parents cannot remember their own experience of toilet learning. For as long as they can remember, socially appropriate toileting has been “automatic” and “natural” and they are vexed by their child’s failure to “just do it!” Toileting is hampered when children lack the sense of comfort and security necessary for them to communicate with their parents, thereby enabling their parents to appreciate their thoughts and fears and respond in helpful ways.

Another source of trouble may be emotionally traumatic experiences with toileting. Parents may try various measures aimed at getting their child to function acceptably. These may range from patient, loving indulgence to severe punishments. Parents’ tirades, use of force, enemas, or other noxious measures are stressful. The more anxiety-prone and the younger the child, the more likely these experiences will be traumatic and impair his ability to manage his own processes of elimination [112, 113].
It should be born in mind that “soiling by its nature is a relapsing condition,” regardless of the treatment applied to it. Buchanan found only about one-fifth of children became permanently clean following the initial course of treatment. Relapses occurred in the majority of patients [89]. Yet, each relapse is an opportunity for the child, the parents, and the clinician to discover why and how it recurred and to practice the skills leading to recovery.

One of the problems in management of functional disorders of elimination is caregivers’ impatience, which is understandable in light of the offensiveness and complexity of fecal soiling. However, patience is rewarded, sooner or later, by the satisfaction of having helped a child and family recover from the misery of uncontrollable fecal soiling.

The Mechanism of Soiling in Functional Fecal Retention Syndrome

Perhaps the commonest reason parents seek medical help for their child with FFRS is uncontrollable soiling [114]. Meunier et al. [115] postulated that soiling is caused by fecal impaction which causes an acquired sensory deficit in the chronically stretched rectal wall. In other words, the child is unable to respond to the urge because he is unable to fully experience the urge as the rectal wall becomes increasingly compliant.

An alternative hypothesis for soiling involves the rectal inhibitory reflex which is triggered by rectal loading: as the urge intensifies, the internal anal sphincter remains open, the pelvic floor relaxes and incontinence is virtually unavoidable. Ordinarily, activation of these reflexes results in uncontrollable passage of stool. But in the case of fecal impaction, the hardened mega-stool cannot pass, but at the same time, any soft or liquid stool cannot be withheld and “overflow” soiling occurs.

In addition to the effects of the rectal inhibitory reflex, the presence of the fecal mass mechanically interferes with the action of the pubo-rectalis muscle which normally prevents stool from entering the anal canal; however, in the case of fecal impaction, it cannot keep the mega-stool from breaching the relative seal at the upper end of the anal canal. In addition, partial relaxation of the pelvic floor during obligatory passage of flatus worsens soiling even further.

The hypothesis that retentive fecal soiling is perpetuated by the insensitivity of the stretched rectum is attractive because of its simplicity. However, it does not hold up in the face of experimental evidence or clinical outcomes [116, 117]. If it were valid, then keeping the rectum free of impaction (using, if necessary, serial enemas) should lead to shrinking of the mega-rectum to a more normal diameter, the return of the normal call to stool, and resolution of soiling with restoration of normal stooling.

Outcome studies have not documented the superiority of such a sensory-mechanical process [95, 116, 118–120]. Van den Berg’s prospective longitudinal study of rectal compliance over the course of a year was done in 101 children with functional constipation [121]. Rectal compliance measured by barostat was
determined at the time of entry to the study. Then, half of the children were treated noninvasively (demystification, bowel diary, toilet training, and daily polyethylene glycol at 0.5 g/kg by mouth); the other half of the cohort were treated with the same regimen plus enemas (120 ml of sodium dioctyl sulfosuccinate and sorbitol) 3 times a week for 3 months, then twice a week for 3 months, then once a week for 3 months. Success was defined as three or more spontaneous bowel movements per week and fecal soiling less than once a week. Success, by this definition, occurred in 41% of patients in both the noninvasive and invasive sub-groups. Follow-up barostat studies were done in 80 of the cohort after 1 year; about half of them had received the non invasive treatment and the other half the same plus enemas. One would expect that patients with the most severe symptoms of FFRS would have abnormal increased rectal compliance [115, 122]. However, no significant change in rectal compliance was found in either of van den Berg’s sub-groups and rectal compliance did not improve in successfully treated patients. Infrequent defecation and large diameter stools at intake were found not to be related to higher rectal compliance. She and her colleagues concluded that increased rectal compliance was not of central importance in the pathogenesis of FFRS, and that enemas did not improve rectal compliance.

Sensory threshold to a distending stimulus was also measured by barostat during the compliance studies. Rectal sensitivity was statistically the same for patients who responded to noninvasive and to the enema treatments, both at the start of the observation and after a year of treatment.

Dr. van den Berg and her colleagues concluded, “a clear relation between achieving successful outcomes of childhood constipation and normalization of disturbed ano-rectal parameters has never been identified… our data show that rectal enemas are ineffective in improving [i.e. decreasing] rectal compliance” [121].

Fully understanding functional disorders of defecation requires more than an understanding of ano-rectal physiology. It requires an understanding of the psychology and psychophysiology of children with these disorders [123]. Arriving at objective truth requires attempt to understand functional disorders of elimination must not let “the more measureable drive out the more important” [124] and not forget that “as nuances of patients’ experiences are compressed into standardized responses, statistical power is achieved, but depth is lost” [125].

**The Nature and Pace of Recovery**

The following general concepts of management are applicable to most children with FFRS. FFRS has two components: the mechanical component and the fear component. The mechanical part of the problem (the retained fecal mass) is relatively easy to overcome, provided the child is emotionally capable of taking laxatives and trying to cooperate. By contrast, children who have been abused or have lived in unstable, environments may not be able to work on their problem. When a child is in an embattled relationship with his parents and parents are entrapped by punitive
responses to their child’s soiling, the child’s desire to oppose and defy his parents may supersede the desire to recover.

Many experts recommend initial “clean-out enemas” [126]. Others have found that such intervention is unnecessary and have advocated a “no-enema approach” to rectal dis-impaction [127, 128]. The use of enemas does not improve rates of recovery. My experience in treating more than 2,500 children with FFRS has been that every fecal accumulation, no matter how firm or large, can be softened to the extent that it becomes impossible to withhold by using enough orally administered osmotic laxative long enough. When the fecal mass becomes very soft or loose, it cannot be withheld because of the rectal inhibitory reflex, the same mechanism that causes incontinence in patients with large volume diarrhea when the rectum becomes overloaded with liquid stool.

Prior to the 1990s, it was difficult to get some children to take mineral oil or magnesium salts because of their taste or texture. However, the introduction of polyethylene glycol powder (PEG) provided a tasteless osmotic laxative that could be dissolved in a child’s favorite beverage [129]. Thus, struggles at the oral end of the GI tract aimed at solving problems at the rectal end could be avoided. Any conflict or coercion during treatment of FFRS tends to impede cooperation between child, parent, and clinician. Therefore, this easier way of administering a tasteless osmotic laxative has been very helpful.

Solving “the mechanical” part of the problem can be accomplished with a “campaign” and a “routine” of laxative administration. The campaign, in children with massive retention, consists of high doses of polyethylene glycol, e.g., 17 g per dose, two or three doses per day [128, 129]. The child and parents must be forewarned that soiling will temporarily increase because the water that has been brought into the rectum by the laxative will soften the fecal mass from its surface towards its center. Therefore, soiling will increase while the lump is softening, before it is soft enough, through and through, to be passed comfortably. A “campaign” should not be attempted on school days. It might begin after school on Friday with another dose of PEG and one more at bedtime. Then, three doses may be given on Saturday and two more, if necessary, on Sunday morning and noon. If massive amounts of stool have not been evacuated by Sunday, stop high doses of laxative and revert to one dose a day after school. If the child hasn’t evacuated by the next Friday, repeat the “campaign” on the next weekend and for as many weekends as necessary for the child to spontaneously empty his rectum. Follow-up visits to the doctor are necessary to assess progress by abdominal palpation and inspection of his bottom for soilage and to encourage their efforts. Failure to make progress may be due to the child’s refusal to drink the beverage that contains the PEG because it isn’t completely dissolved and has a gritty texture. PEG may require considerable stirring before it dissolves completely. A rare child may refuse PEG solution because he or she can taste it. In such cases, older methods of laxation involving flavored milk of magnesia and/or mineral oil should be considered. Laxative failure may also be caused by parents’ fear that the laxative will harm their child or create laxative dependence. Parents need to be reassured that PEG does no harm even when taken for months or years, and that it never results in physical dependence or loss of efficacy.
Parents’ concern and attempts to exercise control over their child’s bowel function can lead to behavioral enmeshment in a process that should instead become the child’s area of control and responsibility. The principles of an agreement between the child and parents might be as follows: The child is “the boss of his own behind” and his own toileting. The parents will not attempt to intrude or exercise control. This does not mean that parents must force themselves to be silent when they see retentive posturing or hear complaints of tummy aches. They can express their concern to the child about his suffering. And they can make suggestions, e.g., “You look so uncomfortable. If I were you, I’d try to make a bowel movement. I bet that will make you feel better.” That kind of statement acknowledges the child’s misery, but is not coercive and it reinforces the reality that the child is the one (the only one) capable of exercising control over his eliminative functions.

If a child seeks his parent’s constant involvement, their response may be that toileting is private. A child does not keep track of his parent’s bowel movements or violate their privacy, and they are not going to invade his. If the child needs help, the parents should offer it willingly, but sparingly—always assisting the child to perform whatever clean up he is having trouble with, but never “taking over.” If the child is anxious when he sits on the toilet by himself, the parent can sit by him to provide calm reassurance and emotional support as long as he seems to need it, but not longer. It’s worth repeating to the child that, even though he has pain or is afraid it might hurt, there is no danger. He or she should take more laxative and keep trying; sooner or later he or she will succeed and then feel so much better! Paul Hyman and colleagues have recently shown that children with fecal retention misleadingly use the term abdominal pain to describe the sensation of intense defecatory urgency; the “abdominal pain” ceases immediately upon passage of accumulated stool [117].

But, what about the soiling? How should the child cope with it especially during the campaign to soften an impassable fecal mass? How can the parents cope with such an obnoxious imposition on their lives and, at the same time, avoid the impulse to punish their child?

The child must be given the means to stay clean. His bathroom should be well stocked with clean underwear within his reach, a stack of wipes and a covered bucket containing a disinfectant solution. When the child has soiled, he is to go to the bathroom, remove his soiled undershorts, drop them into the bucket to soak, replace the cover, clean himself, put on clean underwear, get dressed, wash his hands, and be welcomed back into the presence of the family. There could be a dedicated box of disposable gloves parked under the bathroom sink, to be used by the parent when the liquid waste in the bucket is flushed down the toilet and the underwear is put into the washer. (No other clothes are included with that laundry and liberal amounts of bleach are added to the wash.) The practical and the symbolic purposes of the gloves are to protect the parent from the unpleasantness of contact with soilage. The child should be told that handling waste is disgusting and gloves are worn to avoid the anger that would normally be felt otherwise. It is commonly held that making the child clean up his soiled underwear will “teach him a lesson.” Unfortunately, the clean-up often turns out to cause an even bigger mess which, in turn, causes more anger. Any measure that avoids anger helps focus efforts on achieving the goal of recovery.
Then “a deal” is made with the child. The parents will not be involved or intrusively interested in the child’s bowel function, provided they are not forced to be aware of it. The child can participate normally in family life, but the moment that anyone perceives a persistent malodor, the child is immediately obliged, in a firm, but nonpunitive tone, to remove himself from their presence and stay away until he cleans himself up. His not being permitted to be near others when he soiled must be an “iron rule.” Parents can explain their insistence by saying they don’t want to feel angry towards their child; they love their child. However, it is normal for anyone to become angry when someone in their environment soils and ignores it. Therefore, the child’s denials notwithstanding, he must remove himself and clean himself up; that is his obligation.

Although laxatives can solve “the mechanical part” of the problem, they don’t cure “the fear part” of the problem. The child who has successfully evacuated the “big lump” and regains physical comfort is still afraid of the defecatory urge. And the next time he feels it he will withhold his stool. The cycle of withholding will continue.

The child will get over the withholding response by repeatedly experiencing all good and no bad experiences over time. Re-accumulations must be kept from hardening. This can be done using a “routine” of lower dose laxative, not enough to cause diarrhea, but enough to keep re-accumulations from hardening. If the rectal contents are kept soft enough, they can be extruded with a diameter of about an inch or less. As the rectum fills with soft stool, the urge to defecate will intensify and will reach a point at which he can no longer withhold it. He may then pass a very long stool, but because it is not uncomfortably wide or abrasive, he may look back and realize that the feared bowel movement wasn’t so bad after all.

Children overcome their fear by going through repeated cycles of retention and evacuation. Each time defecation turns out to be easier than expected, the child becomes less afraid of it [130]. Fear perpetuates retentive cycles. Fear can be overcome, provided, (a) there are no other circumstances in the child’s life, such as generalized anxiety, that interfere with his ability to master this or any other challenge; and (b) he has no “bad experiences” with his bowel movements. It is a common error for parents, relieved by their child’s success during the “clean out,” to decide to stop the osmotic laxatives because they worry about the long-term use of any medication. They, like their child, want to believe that the problem is over. But, it is not. It usually requires repeated retentive cycles, each invariably culminating in comfortable success, for the child to lose his fear of the urge to stool [131].

A typical “routine” might involve two teaspoons of PEG a day, increasing to a tablespoon or a capful (17 g) per day by the third day without a bowel movement. The dose could be increased to 17 g twice a day if the child hadn’t stools in a week and that dose might be continued until the child empties his rectum again. Then, the routine dosage is resumed. The above doses are arbitrary. The dose should be adjusted according to the child’s response, aiming for stools that are neither too wide nor uncontrollably loose. Either extreme is felt by the child as a loss of control and should be avoided.
Recovery usually requires routine administration of laxatives for months or more [132]. When the child recovers completely, even an uncomfortable bowel movement will not trigger renewed worry or a recurrence of withholding.

When should laxatives be discontinued? If a child is of school age and able to think rationally, ask him. He knows that the laxatives keep stools soft and comfortable. Does he want to try to stop or does he not want to take the chance at this time? If he chooses to stop and then has a recurrence of discomfort, the “campaign” (if it is even necessary) and the “routine” may be resumed. A relapse poses no danger of bodily harm. It is important to remember that he has choices, after all. As the child grows older, he or she is able to make rational choices about management of the disorder.

Why is it that the reported failure rates of treatment regimens for FFRS range from approximately 25–70%? [116, 118–120, 131, 132] These relatively poor outcome statistics might be due to our habitual tendency to view constipation in biomedical terms, as though it were an organic disease with a specific cure, analogous to pneumococcal pneumonia, in which the patient takes the prescribed medication and passively waits for the disease to end. But, FFRS is a functional disorder that can result from countless contributory factors, not merely the parameters of defecatory function. The challenge for the caring clinician, motivated by an attitude of concerned inquisitiveness, is to search for and ameliorate the contributing factors that lie outside the limits of what is conventionally considered in the assessment of each patient with a disorder of elimination.

The “Retentive Crisis” and Its Importance

The retentive crisis is that feature of the cycle of retention and evacuation that occurs when the stool-withholding child reaches the point at which he or she feels no longer able to keep the bowel movement from coming out. The child behaves in a desperate, agitated, panicky manner that frightens parents and may prompt a rush to an Emergency Room. Actually, the retentive crisis is a unique opportunity for progress, provided its significance and safety are understood. The child may complain of pain and not be able to stay still. Although the child experiencing a retentive crisis does have physical discomfort, the parents must be confident in the fact that most of the distress is panic rather than physical pain. If the parents are frightened, they may resort to an enema, which the child will resist. The productive expulsion of an enema may bring an end to the crisis. The child’s frightening behavior is promptly replaced by calm. However, the problem with this management is that forced enemas compound the distress of the resistant child. Such experiences may worsen, rather than lessen the anticipatory anxiety for future bowel movements.

Enemas are not necessary [127, 129, 130]. In fact, the retained stool may be soft or even liquid. Caregivers help most when their efforts work through, rather than work on the child. The parent needs to be comforting and supportive to their child and reassure him that his stomachache can be relieved if he passes a bowel movement and that no harm will be caused by its passage, even if it hurts his bottom.
Taking more laxative will help. Sooner or later, massive defecation will occur by virtue of the rectal inhibitory reflex, and the crisis will be over. This time, however, it would have been the child who accomplished it, not an externally applied, scary agency. Successful evacuation is then an act of bravery, rather than an experience of submission and humiliation. The retentive crisis thus managed would have a positive effect on overall recovery by making it easier for the child to comply with suggestions for use of laxatives and toilet, rather than reinforcing the fear and self-defeating stubbornness that promotes withholding.

Parents cannot easily provide support to their child during a retentive crisis if they themselves are frightened. They need immediate access to their physician when they begin to have doubts. The physician may need to promptly examine the child to reassure himself, the parents, and the child as well. The only way the child can be brave enough to persevere is if he senses that his parents aren’t scared. And it is the clinician’s job to promptly make sure that there is nothing to be frightened about.

**Problems in the Differential Diagnosis of Functional Fecal Retention Syndrome [26]**

**Hirschsprung’s Disease**

The possibility of Hirschsprung’s disease should be considered in any child of any age who presents with established or emerging functional fecal retention syndrome. Although the diagnosis of Hirschsprung’s is relatively easy in patients who have unequivocal signs and symptoms (see Table 2.3), the diagnosis may be elusive in patients with atypical presentations.

Eighty to ninety percent of Hirschsprung’s disease is diagnosed in infancy [133]. The typical presentation in young infants is that of distal intestinal obstruction with tense abdominal distension, sporadic explosive diarrhea, vomiting, and failure to

<table>
<thead>
<tr>
<th>Table 2.3</th>
<th>A comparison of typical features of FFRS and Hirschsprung’s disease</th>
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<tbody>
<tr>
<td>Typical features</td>
<td>FFRS</td>
</tr>
<tr>
<td>History of delayed passage of meconium</td>
<td>No</td>
</tr>
<tr>
<td>Fecal soiling</td>
<td>Yes</td>
</tr>
<tr>
<td>Abdominal distention</td>
<td>Some</td>
</tr>
<tr>
<td>Spontaneous passage of megastools</td>
<td>Yes</td>
</tr>
<tr>
<td>Retentive posturing</td>
<td>Yes</td>
</tr>
<tr>
<td>Palpable abdominal fecal mass</td>
<td>Yes</td>
</tr>
<tr>
<td>Narrow, empty rectum</td>
<td>No</td>
</tr>
<tr>
<td>Growth lag</td>
<td>No</td>
</tr>
<tr>
<td>Vulnerability to enterocolitis</td>
<td>No</td>
</tr>
<tr>
<td>Transition zone on limited, unprepared contrast enema</td>
<td>No</td>
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gain weight. Withdrawal of the examining finger on digital exam of the rectum evokes a torrent of liquid stool as the seal caused by the contracted aganglionic segment is breached. An infant with this presentation is in imminent peril from enterocolitis, sepsis, and death. Hirschsprung’s Disease can also present in older children and adults [134, 135] and can mimic FFRS.

Is a diagnostic work-up for Hirschsprung’s Disease indicated in all patients with fecal retention? Imposing a rectal biopsy or recto-anal manometry to rule out Hirschsprung’s disease in every patient with apparent FFRS would be onerous and expensive [136]. If there are features suggestive of Hirschsprung’s disease, consultation with a pediatric gastroenterologist or pediatric surgeon is indicated. If symptoms in an older child are equivocal, the problem with differential diagnosis might also be approached with a brief therapeutic trial of laxative therapy, provided there is good rapport and good communication between the physician, parent, and patient. In functional fecal retention syndrome, several days of high dose osmotic laxatives should result in passage of massive amounts of stool with resolution of the palpable fecal mass and a softer, flatter abdomen. By contrast, osmotic laxatives in a typical Hirschsprung’s patient cause increased abdominal distention, but little evacuation of stool.

Hirschsprung’s Disease and hypothyroidism are more prevalent in Down Syndrome patients; these diseases should be considered in any Down Syndrome patient with chronic constipation. If the diagnosis still remains unclear, motility studies and rectal mucosal biopsy are warranted [137]. Intestinal neuronal dysplasia and other forms of intestinal pseudo-obstruction may mimic Hirschsprung’s disease. If constipation persists and is poorly responsive to laxative therapy, evaluation in a center capable of studying these motility disorders is warranted [137].

**Multiple Endocrine Neoplasia Type 2B [138–140]**

**MEN type 2b** is a rare but ominous form of intestinal neuronal dysplasia that should be considered in every child or adolescent that presents with a history suggestive of FFRS or Hirschsprung’s disease. Every child with this autosomal dominant disease will, sooner or later, develop multifocal, metastatic medullary thyroid carcinoma, and may eventually develop pheochromocytoma. Fortunately, there are pathognomonic physical features that are less apparent in infancy but emerge during childhood. These include neuromas of the lips (causing their characteristic “lumpy” or “blubbery” appearance), tongue, eyelids, oral and intestinal mucosae, thickened corneal nerves visible by slit lamp examination, high arched palate, “marfanoid” habitus, and a long face with a prominent, narrow chin.

The lethality of thyroid cancer, which has been reported in children younger than 5 years, warrants consideration of early prophylactic total thyroidectomy and ongoing surveillance for pheochromocytoma [139, 140]. Figure 2.9 and the accompanying case synopsis is that of a girl with MEN type 2b.
Pelvic Tumor

Rhabdomyosarcoma, germ cell tumors, or other pelvic neoplasms may mimic FFRS [141].

Case Vignette

A two-year-old boy had been laboring to pass stool for some weeks and presented with abdominal pain, scant fecal soiling, and a midline pelvic mass typical of fecal impaction. His symptoms were attributed to fecal retention. In-patient administration of high dose mineral oil for 1-½ days resulted in leakage of mineral oil.
per anus, but no passage of stool or lessening of his severe abdominal pains. Imaging revealed a pelvic tumor diagnosed at surgery as a rhabdomyosarcoma.

The possibility of a pelvic tumor should come to mind whenever a presumed fecal mass is palpable in a child whose difficult defecation is actually caused by organic obstruction.

**Stooling Hiatuses in Normal Breastfed Infants**

There is another clinical picture that should not be mistaken either for an organic motility disorder or functional constipation. Most breastfed infants stool several times a day [142]. However, there are a few breastfed infants who may not stool for a week or two, but nevertheless, feel well, gain normally, and remain free of unusual distress [143]. Each hiatus ends with the passage of voluminous soft or loose stool.

This unusual stool pattern is not constipation and should not be treated as such. It is worrisome, but not pathological and it resolves spontaneously within weeks or months. Optimal management consists of close monitoring of the baby for symptoms and signs of intestinal obstruction, such as marked abdominal distention, poor feeding, vomiting, (bilious or otherwise), and increased fussiness. Examine the infant frequently and remain accessible should their infants’ behavior cause alarm, until a more common stool pattern emerges. In my experience, the longest interval without stooling was 20 days in such an infant.

**Anal Ectopy, Anal Stenosis**

Congenital malformations involving dysgenesis of the embryonic cloaca are many and complex [144]. In infants with low imperforate anus, the rectum may end in a fistula to the perineum which may be mistaken for a normal anus on newborn physical examination. However, the opening is not surrounded by the external and internal anal sphincters and is not located where the true anal opening should be. The fistula is narrower and less distensible than a normal anal canal. As such, it may impede normal evacuation of stool and may require dilation or surgical intervention.

The recto-perineal fistula should not be confused with an anteriorly displaced anus [145] which, by definition, is located closer than normal to the posterior fourchette or scrotum, but is surrounded by the external anal sphincter and has the structure, distensibility, sensitivity, and function of an otherwise normal anal canal; it is less likely to cause constipation.

Two kinds of potentially serious comorbidities associated with congenital anomalies of the anus bear emphasizing: (1) the VACTERL association (Vertebral anomalies, Anorectal anomalies, Cardiovascular anomalies, Tracheo-Esophageal fistula, Renal anomalies, Limb anomalies); [146] and (2) tethered spinal cord [147].
**Neuropathic Fecal and/or Urinary Soiling**

Unsuspected damage to sensory and motor nerves of the pelvic floor and its sphincters mimics functional retentive fecal soiling, nonretentive fecal soiling, daytime urinary wetting and/or nocturnal enuresis. Although neuropathic incontinence may occur in a child without any obvious abnormality on physical examination or in radiographs of the lumbosacral spine, tell-tale cutaneous lesions in the lumbosacral area accompany occult spinal dysraphism in 90% of cases. Such lesions include a soft tumor, a hairy patch, vascular marks, a sinus pore superior to the inter-gluteal cleft, or a short gluteal cleft with flat buttocks. Underlying lesions of the spine include lipomeningocele, intradural lipoma, tight filum terminale, neurodermal sinus, tumor of the cauda equina, anterior sacral meningocele, ano-rectal anomalies, and diastematomyelia [148–152]. Rosen et al. [153] obtained MRI’s of the spine in 88 children with intractable constipation. Spinal cord lesions were found in eight. Six had tethered cords, one had a sacral teratoma and one had a syrinx of the thoracic spinal cord. Only one child had a skin lesion suggestive of an underlying spinal abnormality. None had numbness or pain or other abnormalities on neurologic exam of the lower extremities, although two had abnormal anal wink reflexes and one an absent cremasteric reflex. Timely diagnosis permits timely surgery in patients whose lesions involve tethering of the conus medullaris, cauda equina, or diastematomyelia. Unrelieved tethering of the spinal cord results in ischemic damage during the otherwise normal ascent of the conus medullaris from the L-2 level during infancy to the T-12 in adulthood.

A child’s incontinence tends to create frustration in his or her parents. When an organic etiology of soiling exists but is not suspected, the behaviors of the family members toward each other may resemble the distress experienced by families of children with untreated functional soiling and this may add to the mistaken presumption of a functional diagnosis.

**Fecal Retention Due to Anal Trauma or Perianal Dermatoses**

Anal trauma caused by accident or sexual abuse is likely to impede defecation because pain triggers the nociceptive reflex which increases pelvic floor contractility and impairs pelvic floor relaxation [154, 155].

Eczema, seborrheic dermatitis, and diaper rash are examples of perianal dermatoses that may cause anal discomfort and stool withholding. However, there are two perianal dermatoses that cause pain severe enough to result in massive fecal retention. I refer to them as “the red” and “the white” (for mnemonic purposes). The red is perianal streptococcal dermatitis; [156] the white is lichen sclerosis et atrophicus (LSA) [157]. If they are not recognized and successfully treated, management of the secondary fecal retention is likely to fail (Fig. 2.10).
Beta streptococcal perianal dermatitis causes a persistent, bright red, tender eruption confined to the perianal skin. There may be no physically evident infectious lesions in the throat or elsewhere on the skin. However, even though their throats felt and appeared normal, about half of the children in one series had positive throat culture for beta streptococcus [158]. I have treated this skin infection with oral penicillin for 10–14 days and the use of anti-bacterial cleansing agents for bathing. Recurrences are possible.

LSA is a chronic eruption of unknown pathogenesis involving the epidermis and dermis. It can occur anywhere on the skin, but has a predilection for the ano-genital area in girls. It may surround the anus, vulva, or both, resulting in a “figure 8” pattern [159]. It begins as white macules and papules which coalesce into a homogeneous hypo-pigmented area [160]. Eighty-five to ninety percent of cases occur in girls. Symptoms commonly begin between 3 and 7 years of age and may consists of pruritis vulvae, dysuria, and fecal retention. The perianal-perivulvar lesions may become injected and excoriated causing their white color to become reddened [160]. The prognosis of childhood onset LSA is good for improvement or subsidence by or before puberty. (By contrast, adult onset LSA is more resistant to treatment and can lead to atrophy and stricture of the vulva.) Treatment includes topical application of potent corticosteroids. The risk in causing atrophy secondary to long-term use of topical corticosteroids may warrant involvement of a pediatric dermatologist in management of these patients.
**Masturbatory Posturing Mimicking Dyschezia and/or Fecal Retention**

Infant masturbation presents as recurrent self-limited bouts of stereotypic posturing in infants and preschool-aged girls [161–163]. The posturing can be mistaken for stool withholding, abdominal pain, seizures, or dystonia. It typically consists of stiffening of the thighs while standing, sitting, lying supine, prone, or on one side, while applying pressure to the supra-pubic area or above, either manually or by pressing it against a firm edge. (Figure 2.11)

The posturing may be accompanied by facial flushing, diaphoresis, irregular grunting, or moaning respirations and preoccupation without loss of consciousness or responsiveness—activities resembling attempts at passing a difficult stool. The duration of these bouts range from less than a minute to hours. Their masturbatory nature may not be recognized because there is no direct rubbing of the genitalia; stimulation is produced by tightening of the adducted thighs. When asked, the child may complain of pain or discomfort. Moreover, minor irritations in the perineal area, such as diaper rash or irritation of the vulva (with or without synechia of the labia minora) may intensify the child’s sensory experience and exacerbate the behavior. Parents become alarmed and seek medical consultation.

**Fig. 2.11** Masturbatory posturing mistaken for constipation and dyschezia in infant girls [162]
Although infant masturbatory posturing has been well described in recent decades, physicians not familiar with the self-stimulatory nature of these episodes have pursued invasive diagnostic testing and treatment with laxatives, anti-convulsants, and surgery.

Management aims at helping parents change from viewing their child’s behavior as ominous, to considering it to be a harmless, nonpainful habit. Prohibition or punishment of the behavior tends to reinforce it [164]. The term “masturbation” may offend parents who aren’t familiar with concepts of infantile sexuality and auto-eroticism. Referring to the episodes as a “habit” or “self-stimulation” may be more acceptable. Parents who are skeptical of the diagnosis may be convinced and reassured by viewing photographs and video recordings of other children’s posturing episodes [162, 163]. A more realistic understanding should enable them to respond with less alarm, less unintended reinforcement of the behavior, less insistence on stressful diagnostic procedures, and more willingness (with the support of the child’s doctor) to allow time to affect a “cure” [164, 165].

Functional, Nonretentive Fecal Soiling (FNRFS)

This disorder of defecation occurs in children who have no diarrheal disorder, no palpable mass of retained stool, and who pass stools of normal size, consistency, and frequency [91, 166]. They have no anatomic or physiological abnormalities of the colon, ano-rectum, or pelvic floor. Nevertheless, they defecate in their clothes or other inappropriate places, often in addition to their appropriate use of the toilet for eliminative functions. Nonretentive soilers may have less frequent soiling events than retentive soilers, probably because defecation without retention does not occur as often as the uncontrollable seepage of “overflow” soiling in FFRS. Bouts of abdominal pain are less frequent than in children with FFRS, probably because they have no low grade rectal obstruction caused by a fecal mass or are not struggling to resist the defecatory urge (which they call “stomachache” [117]) These children do not evince the decreased appetite typical of children with fecal retention. As might be expected, laxatives worsen nonretentive soiling [167, 168].

My retrospective chart review revealed 50 nonretentive soilers whose findings are summarized in Table 2.4.

Anger is a characteristic feature of most children with FNRFS. It may be expressed overtly, e.g., temper tantrums, or passively, e.g., dawdling, passive disinterest, selective ineptitude—behaviors that embarrass or irritate others. The soiling seems to be impulsive, not premeditated, as it might be if it were a consciously mischievous act. These children and their parents were not consciously aware of the psychological forces that perpetuated soiling. Explanations and warnings of the humiliations and the uncomfortable perianal skin irritations soiling might cause were worth reflecting upon, but seldom were of immediate benefit.

Ordinarily, individuals have no impulse to soil when they feel the urge to defecate and it requires no emotional effort to keep from soiling. By contrast, children
with FNRFS seem to respond to the urge to stool by passing stool in their underwear on an impulse. If the child is promised the new red bicycle he yearns for, provided he uses the toilet and stays clean for, say, 10 days, he may manage to do so. But, compliance requires great vigilance on his part. If he is able to resist the impulse to soil and receives the promised reward, he may immediately relax his efforts and soiling recurs. Bribes don’t work for long, if they work at all. Parents get angry. The hurt of rejection may provide the child with something to feel justifiably angry about and his anger seems to promote more soiling.

Attempts to abolish this vicious cycle by techniques of behavioral modification alone may be perceived by the child as manipulative and he may respond with active or passive stubbornness, adding to a futile control struggle over his toileting.

The biomedical approach may view the soiling as a motility disorder of some kind, something that needs pharmacologic treatment or a behavioristic training procedure. Marc Benninga and his group in Amsterdam have investigated colonic motility in children with FNRFS and found no abnormalities [166, 167]. Biofeedback training is useful in patients with soiling due to pelvic floor dyssynergia, provided there are no emotional impediments to its use and patients are able to cooperate. However, children with FNRFS are generally not enthusiastic or cooperative about getting rid of their symptoms, at least not initially. Management that addresses the underlying causes of the child’s unhappiness, such as family therapy that restores parents’ emotional wellbeing and their ability to understand and ameliorate their child’s emotional distress, is more likely to succeed [46]. Benninga’s group prospectively studied 96 boys and 23 girls with FNRFS for 9 years. The ages of the children ranged from

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**Table 2.4 Features of functional nonretentive fecal soiling**

- Ninety-four percent were boys. A similar male dominance was found by Van Ginkel et al. [167]. This is in marked contrast to the nearly equal sex distribution of children with FFRS.
- The average age at the time of consultation was 7.8 years (range: 3–12 years; median: 7 ¼ years).
- The average age of onset of FNRFS was 4.6 years (range: 2–10 ¾ years; median: 4.5 years).
- The usual estimated volume of soilage consisted of full bowel movements in 64% and smaller amounts in 22% of patients. (It was not clearly recorded in the remaining 16% of patients).
- Forty percent had nocturnal enuresis. Twenty-five percent had daytime wetting (a figure similar to findings of Van Ginkel et al. [167]).
- Half of the daytime fecal and urinary soilers were dry during the night.
- Sixty-four percent were reported to have never soiled at school. (Typically, most children with FFRS do soil at school and it is the school’s intolerance of their soiling that impels the family to seek consultation for the problem).
- When parents are asked to describe their child’s behavior, 81% of their descriptions included one or more of these seven characteristics: irritating, stubborn, willful, negativistic, hostile and defiant, volatile temper, and poor peer relationships. More often than not, the child’s behavior during the course of the consultation allowed the physician and parent to notice and reflect upon the possibility that the soiling was one of several recurrent annoying behaviors, making the importance of emotional factors more plausible.
- Almost all families were in distress. The most frequent phenomena described during semi-structured clinical interviews were: divorce or severe marital discord; the physical and/or emotional absence of a parent; psychiatric illness in one or both parents; a health crisis; financial insecurity; and the death or loss of a family member with whom the child had had an emotional attachment. A history of sexual abuse was obtained in 2 of the 50...
7.9 to 11.6 years and the median duration of soiling prior to entry into their study was 4.4 years. Two years of intensive treatment succeeded in only 29% of patients. By 18 years of age, 15% of the cohort still had FNRFS [169]. It may be almost impossible to distinguish occult spinal dysraphism from functional soiling. When in doubt, an MRI of the lumbo-sacral spine and cauda equina is necessary for diagnostic clarity.

The following three case vignettes are typical of children with FNRFS.

**Case 1**

An 8–10/12-year-old boy, the fourth of four siblings, was seen in consultation for fecal soiling. During her pregnancy with the patient, his mother was preoccupied with her seriously ill mother. Her involvement continued after the patient’s birth and maternal care was largely provided by the patient’s 11-year-old sister. Control of urination and defecation was achieved between 2 and 3 years of age. At four years of age, the patient’s maternal grandmother died. The patient became more aware of his parents’ marital discord. He began to soil and soiled almost daily, except for occasional intervals of continence which never lasted longer than 2 weeks. At five years of age, the patient’s father was hospitalized several times for fractured pelvis and spine suffered in a motor vehicle accident. The patient soiled at home, not at school. He often defecated on the toilet after he soiled his pants. He hid his soiled underwear in the house. When he was obliged to wash them out, he did so, but created a mess. At seven years of age, a psychiatrist diagnosed the patient as a “tense child” and recommended marriage counseling.

Follow-up obtained by a mailed questionnaire seven years later when he was 15 years old, indicated that fecal soiling ceased a few months after the consultation during the course of ongoing family therapy. The patient received 6 months of individual psychotherapy at 14 years of age.

What had helped? The mother answered that the consultation was “…instrumental in helping our child and our family at a very excruciating time. At least we understood it wasn’t organic; it was about control and anger. Therapy alone would not have helped us without your practical advice to let him be in charge of his own toileting and that his bottom wasn’t anyone else’s business. That was hard for us to let go of. But it worked… he learned to separate his feelings from his parents’ problems more effectively.”

**Case 2**

A 7–3/4-year-old boy, the second of three siblings, was delivered by cesarean section which the mother perceived as difficult, mismanaged, and a source of anger. The patient didn’t breastfeed well. She “resented him” until he began to smile at 3 months. The mother returned to her stressful practice of corporate law at about that time. The father continued his work as a commodity trader. The patient was cared for by a housekeeper who placed the infant in front of the television set for long periods. His father was laid off when the patient was 15 months of age. The housekeeper was discharged and the patient was placed into daycare where most of his toilet training was accomplished. Urinary control was achieved at 3 years of age. He began wearing underwear but had inconsistent control of defecation. He began hiding soiled underwear in his room.
He soiled at school and was ridiculed by his classmates. He engaged in anal self-stimulation behaviors. A sister was born when he was 4 years old. The patient was expelled from nursery school because of rudeness and foul language. At six years of age, the patient’s beloved paternal grandfather died. Thereafter, the patient grew closer to his maternal grandfather, an elderly alcoholic described as “emotionally distant.” At seven years of age, the family was obliged to move from Connecticut to Arizona for the mother’s work. A marital crisis occurred which was eventually resolved. The mother continued as breadwinner and the father assumed the role of homemaker. The patient continued to soil at home and at school. His parents argued about his discipline. His mother felt guilty about the quality of the child care he received when he was an infant. During a private discussion, they described their son as a bright, good student but disruptive in class and “forgetful,” “klutzy,” and not always truthful.

In a private chat with the child, he said, “I just don’t feel it coming. I don’t realize I have to go to the bathroom sometimes.” At the end of our talk I asked him to fetch his parents who were in the waiting room so that they would be present during his physical examination. He refused to have his parents with him. I insisted, and we went into the waiting room where he immediately kicked apart the elaborate constructions with blocks he had made.

Follow-up questionnaire 8 years later (when the patient was 15 years of age) revealed that soiling ceased about 9 months after the consultation.

What had helped? “Understanding the problem and a course in biofeedback” was the mothers answer.

**Case 3**

A 10–3/4-year-old boy, the youngest of three siblings, was brought for evaluation of fecal soiling. At 18 months of age, his mother separated from his father, her second husband. Thereafter, his father largely withdrew from contact with the patient. Mother continued to work full time as a business executive. The patient suffered repeated middle-ear infections during his first four years. Between three and four years of age, he engaged in acts of cruelty to the family dog. Nighttime diapers were discontinued, although he was still enuretic. Between five and six years of age, stashes of soiled underpants were found in his room. The volume of fecal soilage approached that of full bowel movements. When he was 6 years old his mother met a man who became her third husband two years later. At 7 years of age the patient began psychotherapy. It took almost a year for the therapist to develop rapport with him. At 8-¼ years of age, the patient and his family moved to a new home, far from his therapist; treatment was discontinued. At about that time, the patient threw his pet hamster into the air and let it fall. The hamster had to be euthanized. At 8–11/12 years of age his mother married her third husband whom the patient admired. Fecal soiling ceased. At 9–10 years of age mother experienced severe financial insecurity because the company for which she worked went bankrupt. The patient resumed psychotherapy with a new therapist whom he liked. His enuresis ceased almost completely. He became able to express his anger more openly. At 10–7/12 years of age, nonretentive fecal soiling and
hiding of soilage recurred after a two year hiatus. His mother described him as perceptive, intuitive, and an excellent artist. But, getting him to do homework was “an ugly struggle—he lies about it.” His room was a “pig pen.” “He’s generally sloppy, although he becomes very meticulous when he decides to clean something up.” “His older siblings can reduce him to tears. He provokes them.”

Follow-up questionnaire 8½ years after the consultation, when the patient was 19 years of age, revealed that soiling ended at about 12 years of age.

What helped? The mother wrote that psychotherapy and “more one-on-one attention from me seemed most helpful.” He was generally happy, well adjusted, and worked full time as well as attending art school.

**Diagnosis and Management of Functional Nonretentive Soiling**

Management begins when the physician elicits the concepts and concerns harbored by the parents (best done with the child out of the room). This is followed by a careful—indeed, conspicuously careful—physical examination which is indispensable to not only the physician’s confidence in the diagnosis but also the parent’s willingness to trust the physician’s explanation of the diagnosis. I inspect the anus for soilage, anomalous location of the anal opening, and anal patulousness that might indicate weakness of the pelvic floor. It is especially important to search for evidence of occult spinal dysraphism and assess the sensory-motor innervation of the sacral segments and the pelvic floor. To do this, one must gain the cooperation of the child who may have previously been subjected, against his will, to digital examinations of his rectum. The method I use is as follows:

After securing the child’s trust, I ask him to help me do a test. I show him a wooden cotton-tipped applicator stick, during which I reassure him that I will not poke it into his bottom or hurt him in any way. I then ask him to watch as I draw a wisp of cotton out of the tip of the applicator. I show him how I touch the wisp to my skin and then to his skin. I explain that I want to test how well he can tell whether or not he can feel the cotton on the skin near the opening of his bottom. I then say that I will sometimes touch his skin and, at other times not do so, and that I will ask him each time whether he can feel it without looking. This allows for assessment of light touch. I thank him for his cooperation. Then I show him how I break the stick in half and demonstrate on my hand and his hand how I touch the skin with the sharp splintered end ever so gently so as not to cause pain. I attempt to elicit an “anal wink.” In repeating the touch-no-touch maneuver, I again asked him to report whether or not he feels the point as I look for this nociceptive reflex. Once accomplished, I thank the child for his cooperation during the test that was so important in helping to figure out the cause of his soiling.

There are two more elements of management: (1) measures aimed at ameliorating the anger caused by the child’s soiling; (2) approaching the mental health aspects that underlie the illness.

The parent’s reaction to each instance of soiling should be nonpunitive but unequivocally frank in saying that soiling is offensive and is felt by others to be a
hostile act. It is the child’s responsibility to keep from soiling or at least keep it from others’ awareness. The message should be verbal, reinforced by a “no-nonsense” tone of voice and facial expression. This temporary withdrawal of warmth and expressions of love conveys the message that the parents are displeased by the child’s offensive act. It does not involve the issue of love. Rather, it is a normal reaction to thoughtless, offensive behavior similar to any wanton act that causes physical hurt. Unlike punishment, which creates the impulse to retaliate, it is a temporary withdrawal of approval and feelings of love that the child feels as a loss and basically wants to regain. It is, after all, reality based. (Unfortunately, the temporary withdrawal of love and approval is ineffective with children without a secure attachment with parents who are unable to express love and approval to begin with.)

The child is told that his toileting is his private function and that, just as they would not welcome his intrusion into his parents’ toileting, so they expect that he would not want them to be involved in his. Of course, he might need help at times and help should be offered, if necessary, willingly but sparingly. It should never amount to taking over the clean-up; they should always assist him, not do it for him.

Next, in a talk with the parents alone, I discuss differences between functional fecal retention syndrome, a diagnosis for which the child may have previously been treated without success, and functional nonretentive fecal soiling, in which there is no “mechanical” cause for soiling. “The good news” is that their child has no organic disease and that his report of not being able to “feel it” is a perception/excuse that most children with FNFRFS offer. This finding is based upon the physiologic responses demonstrated during the physical examination. Most parents are relieved to learn that neurologic disease and surgical measures are out of the question.

But what is the cause of the soiling and what can be done to make it stop? The answers to these questions necessitate skillful communication during which the physician absolutely avoids saying anything that might sound judgmental. The attitude of the physician must come across as one of sympathetic concern, as an ally, not the judge of their parenting.

Is their child angry? I review the evidence revealed during the history of their child’s other irritating behaviors, correlate them with the history of emotionally stressful events, and propose that the soiling is, like his other behaviors, a passive rather than an overt expression of anger. His behavior certainly irritates those in his environment. In addition, I acknowledge how odious soiling is for parents and how difficult it is to avoid a vicious cycle of punishment and retaliation that serves to fuel the impulse to soil.

Some families welcome a discussion of emotional factors and are quite willing to pursue whatever individual and/or family therapy is suggested by the physician. Many other families, however, are unwilling to acknowledge the existence of emotional problems and are reluctant to assume the role of client in need of psychological help—as though it would indicate fault or weakness, rather than courage on their part. An authoritarian recommendation for psychological help to unwilling parents is seldom useful. The physician’s insistence, even as a condition of his or her continued care, is likely to damage rapport and may cause them to seek care from another clinician who “would take their child’s soiling seriously, rather than diagnosing it is as being ‘all in his head’—a doctor who would be willing to do more tests to find out what’s really wrong.”
A productive mental health referral that avoids such disruption is more likely if four prerequisites are met: [170] (1) parents are able to discern and feel troubled by signs of emotional difficulties in their child; (2) parents are able to view a mental health referral as potentially useful; (3) access to a competent therapist must be geographically and financially feasible; (4) care of the patient must not be entirely transferred over to the therapist because parents may worry that organic disease may have been missed or might have supervened. The therapist needs freedom to work on their emotional issues without having to feel primarily responsible for the child’s physical diagnosis or the type or amount of medication necessary. That is the physician’s responsibility.

The family therapist, Helen Reid [46], uses an interactive parent–child family therapy approach in which emotional factors within family life are identified and worked with. In her experience, one, or more of three clinical themes are frequently encountered in children with previously refractory fecal soiling: (1) parental conflict; (2) the repressed feelings of being displaced upon the arrival of a newborn sibling and the parent’s expectation that the child love the new baby; (3) tormenting by older siblings. The therapeutic event consists of bringing to light the emotions that perpetuate the patient’s soiling, leading to changes in attitudes and behaviors that contribute to the patient’s impulse to withhold and/or soil.

What is the role of the physician with patients who do not meet all of the prerequisites for mental health referral? The physician may act as a surrogate mental health professional guided by Hollis’s classification of therapeutic interventions and the biopsychosocial model of practice [85]. He or she can provide continuity of care, always alert for opportunities to validate their expressions of emotional pain and reflect upon possible approaches to a better understanding of it, until it either resolves or their resistance to a mental health evaluation is overcome [171].

Table 2.5 compares the typical features of retentive and nonretentive functional fecal soiling which may help in distinguishing them from each other.

<table>
<thead>
<tr>
<th></th>
<th>Retentive</th>
<th>Nonretentive</th>
</tr>
</thead>
<tbody>
<tr>
<td>Relative prevalence in practice</td>
<td>74 %</td>
<td>13 %</td>
</tr>
<tr>
<td>Nature of soilage</td>
<td>Frequent seepage</td>
<td>Sporadic full or partial BMs</td>
</tr>
<tr>
<td>Mid-line fecal mass</td>
<td>Present</td>
<td>Absent</td>
</tr>
<tr>
<td>History of constipation</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Passage of mega-stools</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>History of clogging the toilet</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Longest interval between BMs</td>
<td>Weeks or months</td>
<td>A few days or less</td>
</tr>
<tr>
<td>History of cyclic appetite or mood changes related to defecation</td>
<td>Typical</td>
<td>Not typical</td>
</tr>
<tr>
<td>History of anal pain and/or blood-streaked stools</td>
<td>Typical</td>
<td>Not typical</td>
</tr>
<tr>
<td>Radiologic megarectum</td>
<td>Present</td>
<td>Absent</td>
</tr>
<tr>
<td>Typical attitudes of the child</td>
<td>Anamistic fear of the emergence of stool</td>
<td>Passive or active aggressivity</td>
</tr>
<tr>
<td>Sex distribution</td>
<td>Nearly equal</td>
<td>Predominantly male</td>
</tr>
<tr>
<td>Indications for laxatives</td>
<td>Present</td>
<td>Absent</td>
</tr>
</tbody>
</table>
Children with Features of Both Retentive and Nonretentive Syndromes

Most children with functional fecal soiling are either retentive or nonretentive soilers. However, in 4% of the cohort of 395, it was not possible to categorize functional soiling as entirely retentive or nonretentive. They had features of both syndromes nearly simultaneously or at different times during their course. Management of such cases aims at the treatable aspects that are present at the moment of care. Sooner or later, the diagnosis will become apparent and/or the soiling will resolve. The family’s response to their child’s soiling should be similar in both situations requiring clarity of boundaries as to how offensive soiling really is, nonintervention in controlling the child’s toileting, clear expectations that the child confine eliminative functions to the bathroom, non-tolerance of his or her presence when soiling is perceived by others; and reasonable means of getting rid of soilage so that others in the child’s environment are not offended and the child can avoid embarrassment. When hard stools cause dyschezia or accumulate as a fecal mass, appropriate doses of osmotic laxatives should be administered, intermittently or continuously, in doses that will keep stools soft. Avoid high dose laxative in the absence of signs and symptoms of fecal retention so that soiling isn’t made worse during nonretentive intervals [167]. Emotional difficulties in the child and family life should always be addressed, to the extent practicable. The following case vignette exemplifies a child whose fecal soiling had both retentive and non-retentive features.

Case Vignette: Mixed Functional Fecal Soiling

A 6–3/12-year-old girl, the older of two siblings, toilet trained well between 2-½ and 3 years of age. At 3 years she accompanied her parents on an 8 week trip to another country. She did not defecate for two weeks during that time but did no soiling. After being giving a laxative, she passed a stool that was 3 in. in diameter followed by relief of abdominal discomfort. The patient had been cared for by a housekeeper to whom she was emotionally attached since two weeks of age. When she was 5 years old, the housekeeper left their employment, but continued to visit as a friend and kept in telephone contact with the patient. Her departure was followed by a succession of about ten housekeepers during the next 14 months. The patient entered kindergarten at 5-¼ years of age. Occasional fecal soiling was noticed 3 days later. Although she liked her teacher, soiling recurred intermittently, especially when the teacher was absent from class for a day. At 6 years of age the patient was enrolled in summer school. She was very competitive and successful but was made fun of by other children because of soiling. Her parents decided not to re-enroll her for the second summer session. At about that time, she passed an “orange-sized stool” during sleep. She soiled “all day long” at her entry into the First grade at 6 ¾ years. There was no enuresis or daytime wetting.

Social aspects: Mother remembered intense conflict with her own mother around toileting between 5 and 7 years of age and being given many laxatives and enemas.
The patient was described as bright, sweet, good-hearted, but stubborn and bold. The parents were openly affectionate towards each other, but also fought in the presence of their children. The patient’s father had never felt ready to be a parent. “He’s always cold and rejecting of her, never affectionate with her,” the mother said.

Physical examination was negative for sensory-motor defects in the sacral dermatomes and pelvic floor. Her abdomen was flat, its contents easy palpable; there was no fecal mass. The anus was normal to inspection, but soiled with about 5 cc’s of pasty stool.

The above patient’s history was that of functional fecal retention; however, her physical examination was typical of nonretentive fecal soiling. This case exemplifies functional fecal soiling with mixed features of retentive and nonretentive types, in a child of an emotionally troubled family.

**Diaper Dependency**

A child’s readiness for toilet learning/training is indicated, for example, by showing an interest in using the potty or indicating a need to void. Readiness skills usually appear between 24 and 29 months of age (although some children may not be fully ready until their fourth or fifth year.) [36] In the toddler age group, the interactive learning process may be accompanied by negative and oppositional responses to parents’ limits and requests [172, 173]. Failure to make the expected progress from diapers to underwear becomes a clinical problem when parents become concerned about the possibility of an organic etiology for prolonged use of diapers as well as the stress it causes in their relationship with their child [174–176].

Seven percent of our cohort of 395 children referred for evaluation for fecal soiling and other toileting difficulties were children 3–6 years of age who persisted in the use of diapers for urination and/or defecation. It turned out that the children who persisted in using diapers could be sub-categorized into two main groups: (1) Permitted diaper dependency; and (2) Contentious diaper dependency. The distinction between the two groups is important because their management differs; what might help a child with permitted diaper dependency may make contentious diaper dependency worse.

In the permitted group, parents were, on some level, accepting of their child’s toileting behavior and that sense of acceptance seemed to obscure the goal of mastering autonomous, age appropriate control [177]. By contrast, in the contentious group, parents were emphatically un-accepting of their child's immature toileting behavior, which most often consisted of use of the toilet for urination but refusal to defecate except in a diaper or pull up [178]. In contrast to the low level of parent–child conflict around toileting characteristic of permitted diaper dependency, alarming levels of parent–child conflict were typical in cases of contentious diaper dependency.
The management task in permitted diaper dependency is to help parents recognize the ambivalent quality of their reactions to their child’s refusal to use the toilet. This recognition enables parents to strengthen their expectations and demands on their child to stop using diapers and, instead, pass all stool and urine into the toilet like everyone else.

By contrast, such an approach in cases of contentious diaper dependency can worsen the problem [179]. The child isn’t confused and the parent’s expectations are quite clear. Placing more pressure on the child to change only intensifies the conflict. The management task in contentious diaper dependency is to help parents call off the battle by telling the child that toileting is his business, to be done by him in his own way, provided that he does not impose his soilage on others’ awareness. The hoped-for result of this change in attitude is for the child, sooner or later, to prefer using the toilet. This becomes possible for the child once doing so no longer feels like capitulation and that his parents no longer want to impose their will in his eliminative functions.

There were 27 diaper dependent children in the cohort of 395 patients. 19 (16 boys and 3 girls) were diagnosed as permitted diaper dependent; eight children (six boys and two girls) had contentious diaper dependency [180]. Their ages at the time of consultation ranged from 3–1/12 to 6–2/12 years. None of the permitted diaper dependent group had symptoms of fecal retention at any time. About half of the contentious group had, at some time in their course, experienced symptoms of fecal retention. However, relief of their dyschezia had not altered their refusal to use the toilet. The rest of the contentious group had no history of stool withholding (except during brief intervals in which parents refused to provide diapers or pull ups) and no memorable recurrent physical discomfort during defecation.

The following two case vignettes are of children with permitted diaper dependency.

**Permitted Diaper Dependency**

**Case 1**

A 3-½-year-old boy began toilet training at 24 months. He was given a potty and complied with requests that he sit on it, but he never passed stool or urine into it. One month later his parents adopted a 3 day old infant. Whenever his mother attempted to talk with him about his refusal to use the potty, “he changed the subject.” He nevertheless put his doll on the potty. He had been put into underewear at 3–2/12 years but continued to urinate and defecate into them, so his parents reverted into putting him into diapers. “We don’t force him. When I tell him I have to change him because I can’t bear to smell it anymore, he tells me, ‘I don’t want to change, Mom’” He was the only child in his nursery school class who wore diapers. His teachers changed him and didn’t seem to mind. “They say I’m over-reacting and that it was ridiculous to take him to a doctor for this. Everyone says he is a perfect child except for this one thing… When my husband
tells him to use the potty; he says he should, but he doesn’t want to. We haven’t exploded at him, it’s just not in our nature… My husband’s family are screamers, so he is not willing to be loud with him.”

Physical examination showed a healthy-appearing, articulate boy who hastened to assure me that he “goes” in his potty. He wore a wet diaper. The perianal skin was erythematous but not soiled. The remainder of the examination was negative. I discussed the importance of making it clear to her child that eliminating into diapers is offensive and that everyone expects children to use the toilet. I recommend that they get rid of diapers and offer him attractive underwear. I contacted his nursery school teacher and explained that their kindly acceptance of his immature toileting obscured the goal necessary for toilet learning. I recommended that they require him to wear underwear and make it clear that soiling was not acceptable because it was offensive. Six weeks later the patient wore underwear at school and at home and used the toilet for urination. He insisted on wearing a diaper at night and continued to defecate into it while in bed. (The patient was then lost to follow-up.)

Case 2

A 5–9/12-year-old boy had never used the toilet on his own initiative and continued to urinate and defecate into pull-ups. He was born prematurely weighing 1,847 g. He suffered necrotizing enterocolitis and underwent five abdominal surgeries during the 2–1/2 months he was in a neonatal intensive care unit. During his second year, he suffered repeated middle ear infections and Stevens-Johnson syndrome. Toilet training began at 18 months, but he didn’t seem interested and further efforts were suspended. At 2–6/12 years of age, he urinated into the toilet prior to his bath at the request of his parents. As he sat on the toilet, his older brother went up to him and said, “Just go pooh!” “It’s broken,” the patient replied. He continued urinating and stooling into diapers and occasionally stooled on the toilet at his mother’s request, provided she accompanied him. Otherwise, she did not insist that he use the toilet. If she noticed malodor she didn’t mention it, hoping he would ask to be changed. He never seemed bothered by the soilage, so she continued to change his diaper after every bowel movement. The patient entered preschool at 4–10/12 years of age. His parents warned him beforehand that soiling would not be acceptable there, but he did not seem to care. It turned out that his teachers were “very understanding” and quite willing to change his soiled pull-ups. They told his mother not to express displeasure at his infantile eliminative behavior. His mother had never been sure that there wasn’t a medical reason for it. When I asked the mother what it was that concerned her most about her son, she said, “We were told throughout the pregnancy that he could have deformities… I felt that it was my fault as a mother… I probably do dwell on the surgeries of the past.”

Physical examination showed a verbally articulate boy with a well-healed incision on his upper abdomen, mild perianal erythema, and no other abnormalities. I began by saying that, notwithstanding a series of intestinal catastrophies as a newborn, his GI function was now normal and there was no organic cause for his diaper dependency. I recommended that she tell him that he is the boss of his own
eliminative functions and that they stop reminding him because it made them responsible for getting him to use the toilet. At the same time, the parents were to make it clear that soiling was offensive. They were to place nonpunitive, but firm limits that would prevent him from imposing his soilage on others’ awareness. The use of pull-ups would cease. Instead, “big-boy” underwear was to be worn day and night, at home and at school. They were to expect him to do for himself with age-appropriate self-sufficiency, as though he were a perfectly healthy, intelligent 5-year-old—which indeed he was! Within 2 months of the consultation, the patient no longer soiled or wore pull-ups. He used the toilet appropriately. “I toughened-up,” his mother said. “If he made a mess, I told him to clean it up… I learned I could do a lot less for him and still be a good mother.”

Effective management of permitted diaper dependency involves helping parents discover and re-evaluate the nature and sources of the feelings and behaviors that cause their expectations regarding the mores of toileting to be unclear. In Case 1, the parents’ fear of conflict with their child prevented them from placing clear expectations for age-appropriate toileting. In Case 2, the child’s infantile toileting was perpetuated by his parents’ unrealistic fears that he was abnormally susceptible to illness or death. Such fears originated in previous experiences during which their child suffered life-threatening injuries, disease, or life-altering handicaps. This parenting dysfunction has been described as “the vulnerable child syndrome” [181].

Parents need to insist that their child stop using diapers and instead use the toilet at school and at home. The sanctions to be employed are nonpunitive, similar to those recommended for management for functional nonretentive fecal soiling. The clinician must remain accessible so that what was learned during the consultation can be “worked through” [182] until the problem has been overcome.

**Contentious Diaper Dependency**

In permitted diaper dependency, parents were, on some level, accepting of their child’s immature toileting behavior. As a result, children were unsure about what their parents and others expected. There was comparatively little conflict between the parents and their children, although the parents were concerned enough to seek medical consultation. By contrast, children with contentious diaper dependency were quite clear about what their parents expected of them because the parents were pointedly outspoken about the unacceptability of soiling and use of diapers or pull-ups. Although they insisted on diapers for defecation, these children used the toilet for urination. They wore underwear for most of the day and only asked for diapers when they felt the urge to stool. They all asked to have their soiled diapers removed when they were finished, as though they didn’t want anything to do with their feces. Anxiety, emotional regression and/or oppositional defiance were prominent features of their clinical presentation. Persistent parental pressure to use the toilet for defecation was met with increasingly obstinate refusal often leading to alarming levels of conflict with their parents.

The following case vignettes are children with contentious diaper dependency. The first had elements of fecal retention; the second child did not.
Contentious Diaper Dependency *with* Fecal Retention

**Case 3**

A 4–5/12-year-old boy toilet trained for urinary control between 2–9/12 and 3–2/12 years of age. Thereafter, he wore underwear and used the toilet autonomously for urination. He defecated on the toilet only twice, 3 weeks apart, at 4–4/12 years; otherwise, he only defecated in pull-ups. He was routinely provided a pull-up at bed time. He put it on, defecated, went to the toilet, removed his soiled pull-up, dumped its contents, flushed, wiped himself, put on a clean pull-up and went to bed. He withheld stool for 3 days, during which he displayed retentive posturing and soiled. He finally passed a large, blood-streaked stool.

At 3–4/12 years of age his maternal grandmother, who had been babysitting him and his younger brother, suddenly fell to the floor, paralyzed. She instructed the patient to telephone an uncle for help, which he did. During the ensuing 4 months, the patient’s mother was preoccupied with caring for her mother who then died at the age of 58.

The patient’s mother described him as happy, affectionate, but stubborn, especially when asked to give up his bottle or diapers. “He has a strong need to be in control, just like me.” In a separate chat with the patient he explained that he was “afraid of going to the potty.”

Contentious Diaper Dependency *Without a History of* Fecal Retention

**Case 4**

A 4–1/2-year-old boy was born soon after his parents emigrated from Croatia. His mother returned to work 6 weeks later and he was cared for at home by his maternal grandmother. At 12 months, his grandmother returned to Croatia and he was put into daycare where toilet training was begun. He was placed on a potty repeatedly during the day despite his resistance each time. He began urinating in the toilet standing up at 2 years and wore underwear during the day, but diapers at night. At 3 years, his parents realized that he mostly stooled in his diapers while in bed. “I asked him to sit on the toilet. He looked scared, his pupils dilated, and his heart raced… so, if he gets the urge, he just asks for a diaper and we give it to him and he goes.” Whenever they refused his requests for a diaper he withheld bowel movements. His parents never persisted longer than 3 days without acquiescing and he never developed symptoms of fecal retention. “Both my mother and I have anxiety and I don’t want my son to get that way,” his mother said. Past history included five hospitalizations for pneumonia and ENT procedures, including an emergency helicopter transport for acute laryngitis at 4 years of age. The patient’s mother suffered a “nervous breakdown” at 22 years of age and subsequent phobic anxiety which was incapacitating at times. The paternal grandmother had died of cancer when the father was 13 years old.
The patient was described as being fearful of new things of any kind, such as a new toy that he’d never seen before. He hated loud noises and insisted on being forewarned so he could leave the room before a vacuum cleaner or blender was turned on.

Physical examination revealed no palpable fecal mass or soilage and was otherwise negative. The salient feature of this boy’s diaper dependency seemed to be severe anxiety which interfered with his ability to try new things, including defecating on the toilet. The parents were advised to stop pressuring the child to give up diapers (without implying that his immature toileting was unimportant to him or them) and pursue treatment for his anxiety.

The first step in treating contentious diaper dependency is to call off the power struggle [178–180]. At the same time, the goals of cleanliness must be kept clear by reminding the child (without being irksome) that everyone expects everyone else to use the toilet appropriately and that the use of diapers is potentially embarrassing. Parents owe their child love, but peers and other adults do not, so if the child wants to be accepted socially, he must not soil or at least be vigilant and get the help he needs to keep it from becoming known. Cleaning up soilage for children who are old enough to do it for themselves is an odious burden for parents and they should make that clear in a nonpunitive, nonderisive manner, and should not allow their child to presume otherwise.

Regular administration of an acceptable osmotic laxative in doses that soften stools (without liquefying them) is necessary in those children with contentious diaper dependency who experience painful defecation.

**Infant Dyschezia [183, 184]**

Infant dyschezia occurs during the early months of life in some healthy infants. It consists of recurrent bouts of intense grunting and straining lasting many minutes or even the better part of an hour, culminating in the passage of a soft or liquid stool, followed by subsidence of the infant’s apparent distress. Stools are not hard or blood streaked. This condition was encountered once in our review of 395 patients. It is not associated with an organic disease and it resolves spontaneously before 6 months of age. However, it alarms parents.

The mechanism of this behavior is presumed to be dyssynergic. The infant responds to the defecatory urge with a valsala maneuver (grunting), but does not synergistically relax his pelvic floor. The passage of stool is impeded, the defecatory urge persists and unrelieved straining continues. Management consists of eliciting an unhurried history, performing a thorough exam including a gentle digital exam of the rectum to rule out anal stenosis or other blockage. This is followed by effective reassurance as to the mechanism, the safety and excellent prognosis of the condition. Recovery from infant dyschezia is accomplished by allowing the baby to struggle with repeated experiences of defecation during which synergy of abdominal and
pelvic floor muscles is finally mastered. “Helping” the infant defecate by inserting suppositories or other objects is not recommended because such help is not needed and such potentially noxious stimulation might interfere with the process by which the infant learns how to defecate with ease.

**Cases Vignette: Infant Dyschezia**

A 7-week-old girl, the only child of parents in their mid-20s, was the product of a normal pregnancy and delivery. There was no delay in passage of meconium. The patient was breastfed with occasional supplements of cow’s milk formula. She passed watery-to-pasty stools 2–3 times a day, except for 3 days at about a month of age during which she passed pellet stools.

She became quite fussy at 4–5 weeks of age and “…cried most of the time she wasn’t asleep or being fed.” The crying was “colicky,” i.e., she could be quieted by rocking, being placed in a swing or being carried about. By 5 weeks, the frequency of stooling decreased to once every other day. Nocturnal sleep increased to as long as 7–1/2 h at a time, but fussiness between 4 and 6 PM continued. At six weeks, there were 4 days during which she did not defecate. She did not seem unusually distressed. However, she began “grunting,” fussed more and slept no more than 4 h at a time.

Two days prior to the consultation, an abdominal radiograph was done because the patient hadn’t stooled in a week. No signs of obstruction and no abnormal accumulations of stool were found. The parents inserted a rectal thermometer that evening. This evoked copious liquid stool after which she was breastfed and fell asleep. The grunting recurred when the patient awoke the next morning.

Physical exam revealed an alert infant who was alternately socially responsive and intensely fussy. Her abdomen was soft. There was no palpable mass. The anus was normal to inspection. The physical exam was otherwise negative. During the physical exam, the patient spontaneously passed a large, liquid, brilliant yellow stool (negative for occult blood). Management consisted of effectively reassuring the parents of the nature and safety of their daughter’s behavior. I explained the importance of nonintervention so that the infant could learn how to coordinate her efforts to defecate. I offered unlimited access should they become concerned.

Follow-up 5 weeks later revealed that the infant’s fussiness subsided at 3 months of age. She stooled every day or two without distress, although she sometimes became cranky when she hadn’t stooled for more than 24 h. The mother felt that her daughter had greatly improved and they no longer harbored doubts about her health.

**References**


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