Normal bowel continence is a complex process that involves the coordinated interaction between multiple different neuronal pathways and the pelvic and perineal musculature. The importance of the anatomic relationships of the pelvic floor in maintaining normal continence has been suggested since the 1950s. Yet the complex series of neural and behavioral-mediated interactions, combined with a lack of an ideal study to take all elements into account, makes complete understanding of anorectal anatomy and physiology’s role in preserving continence difficult. Complicating this are multiple other factors that have a role in normal regulation such as systemic disease, emotional effects, bowel motility, stool consistency, evacuation efficiency, pelvic floor stability, and sphincter integrity.

Anorectal physiology testing allows evaluation of the patient with pelvic floor complaints using techniques such as manometry, endoanal ultrasound, electrophysiologic studies, and defecography, all of which help to elucidate anorectal structures and function. A physician with an in-depth knowledge of normal and abnormal anorectal physiology can apply the results in a meaningful way to diagnose and direct therapy. This chapter reviews the current knowledge regarding muscular, neurologic, and mechanical factors.

Pelvic Floor Muscles

The pelvic floor consists of a striated muscular sheet through which viscera pass. This striated muscle, the paired levator ani muscles, is actually subdivided into four muscles defined by the area of attachment on the pubic bone. The attachments span from the pubic bone, along the arcus tendineus (a condensation of the obturator fascia), to the ischial spine. The components of the levator ani are therefore named the pubococcygeus, ileococcygeus, and ischiococcygeus. The pubococcygeus is further subdivided to include the puborectalis. Between the urogenital viscera and the anal canal lies the perineal body. The perineal body consists of the superficial and deep transverse perinei muscles and the ventral extension of the external sphincter muscle to a tendinous intersection with the bulbocavernous muscle.

The fourth sacral nerve innervates the levator ani muscles. Controversy continues regarding the innervation and origin of the puborectalis muscle. Cadaver studies differ from in vivo stimulation studies as to whether the puborectalis muscle receives innervation only from the sacral nerve or also from the pudendal nerve. Comparative anatomy and histologic studies of fiber typing also support the inclusion of the puborectalis muscle with the sphincter complex and not as a pelvic floor muscle. In addition, electromyographic (EMG) studies of the external anal sphincter (EAS) and puborectalis muscle indicate that the muscles function together with cough and strain.

The rectal smooth muscle consists of an outer muscularis mucosa, inner circular muscle, and outer longitudinal layer. The inner circular muscle forms the valves of Houston proximally and distally extends down into the anal canal becoming the internal anal sphincter (IAS). This is not a simple extension of muscle because there are histologic differences between the upper circular muscle and the IAS. For instance, the IAS is thicker than the circular muscle because of an increased number of smaller muscle cells. The outer longitudinal layer surrounds the sigmoid colon coalescing proximally into thicker bands called taenia coli. This same layer continues down to the anorectal junction where it forms the conjoined longitudinal muscle along with fibers from the pubococcygeus muscle. Distally, this muscle lies in the intersphincteric plane and fibers may fan out and cross both the IAS and EAS muscles. In an ultrasound view of the anal canal, the longitudinal muscle is seen as a narrow hyperechoic line in the intersphincteric space. The terminal fibers extend to skin as the corrugation cutaneous ani muscles.

External Anal Sphincter

Anatomic and sonographic studies indicate that the EAS begins development, along with the puborectalis muscle, at 9–10 weeks’ gestation. At 28–30 weeks, it is mature and the
Internal Anal Sphincter

The IAS is an involuntary, smooth muscle. It is relatively hypoganglionic.\textsuperscript{11} There are nerve fibers expected in an autonomic muscle—cholinergic, adrenergic, and nonadrenergic, noncholinergic fibers. It receives sympathetic innervation via the hypogastric and pelvic plexus. Parasympathetic innervation is from S1, S2, and S3 via the pelvic plexus. There is considerable evidence that the sympathetic innervation can be seen in the exceptionally dissected specimen and, in most cases, the muscle is one continuous mass and should be considered as such.

The EAS is innervated bilaterally by the pudendal nerve arising from S2-S4. Motor neurons arise in the dorsomedial and ventromedial divisions of Onuf’s nucleus in the ventral horn of the spinal cord. Crossed of the pudendal innervation was first suggested in studies by Wunderlich and Swash\textsuperscript{9} on rhesus monkeys. Hamdy and associates\textsuperscript{10} evaluated corticoan stimulations of humans and found variable crossovers which were symmetric in some and either right- or left-sided dominant in others. This has been offered as one possible explanation for the inconsistent relationship between unilateral pudendal neuropathy and fecal incontinence.

Sensory

Anal canal sensation to touch, pinprick, heat, and cold are present from the anal verge to 2.5–15 mm above the anal valves. This sensitive area is thought to help discriminate between flatus and stool but local anesthesia does not obliterate that ability. The rectum is only sensitive to distention. Rectal sensation may be attributable to receptors in the rectal wall but also in the pelvic fascia or surrounding muscle. The sensory pathway for rectal distention is the parasympathetic system via the pelvic plexus to S2, S3, and S4. Below 15 cm, rectal distention is perceived as flatus, but above 15 cm, air distention causes a sensation of abdominal discomfort. Anal canal sensation is via the inferior rectal branch of the pudendal nerve that arises from S2, S3, and S4. This is the first branch of the pudendal nerve and along with the second branch, the perineal nerve, arises from the pudendal nerve in the pudendal canal (Alcock’s canal). The remainder of the pudendal nerve continues as the dorsal nerve of the penis or clitoris.\textsuperscript{14}

Reflexes

There are a great number of reflexes that end with the name “...anal reflex.” The reason for this is, in part, that the EAS is readily accessible and represents a convenient end point for recording during electrophysiologic study. Consequently, there are several ways that one can assess the integrity of neurologic connection through or around the spinal chord.\textsuperscript{15}

Cutaneous-anal Reflex

The cutaneous-anal reflex was first described by Rossolimo\textsuperscript{16} in 1891 as a brief contraction of the anal sphincter in response to pricking or scratching the perianal skin. This is a spinal reflex that requires intact S4 sensory and motor nerve roots. Both afferent and efferent pathways travel within the pudendal nerve.\textsuperscript{16} If a cauda equina lesion is present, this reflex will usually be absent. Henry et al.\textsuperscript{17} recorded the latency of the anal reflex in 22 continent patients as compared with 33 control subjects. The mean latency was 13.0 versus 8.3 ms, respectively. The mean latency was within normal range in only 3 (14%) of the continent patients.\textsuperscript{17} However, Bartolo et al.\textsuperscript{18} have suggested that latency measurement of the cutaneous-anal reflex may be an inadequate means of demonstrating nerve damage in patients with fecal incontinence. From a practical standpoint, this is a sacral reflex that can be interrogated during physical examination by simply scratching the perianal skin with visualization of contraction of the subcutaneous anal sphincter. The response to perianal scratch fatigues rapidly so it is important to test this as the first part of the sphincter examination.
Cough Reflex

Chan et al.,19 using intercostal, rectus abdominis, and EAS electrodes, studied the latencies in response to voluntary cough and sniff stimulation. When compared with latencies from transcranial magnetic stimulation, it appeared that the EAS response was consistent with a polysynaptic reflex pathway.19 The visible contraction of the subcutaneous EAS as a consequence to cough and sniff stimulation is a simple noninvasive validation of the pathways involved in the anal reflex. This response can also be displayed during anal sphincter manometry. Amarenco et al.20 demonstrated that the greater the intensity of the cough, the greater was the electromyographic response within the anal sphincter. The reflex is preserved in paraplegic patients with lesions above the lumbar spine but it is lost if the trauma involves the lumbar spine or with cauda equine lesions. The mechanism of the cough–anal reflex contributes to the maintenance of urinary and fecal continence during sudden increases in intraabdominal pressure as might also be seen with laughing, shouting, or heavy lifting.

Bulbocavernous Reflex

The bulbocavernous reflex was first described by Bors and Blinn21 in 1959. The bulbocavernous reflex is the sensation of pelvic floor contraction elicited by squeezing the glans penis or clitoris.22 The EAS is used as the end point because it is easily accessed either for visual assessment or by concentric needle EMG recording. The bulbocavernosus reflex latency will be prolonged by various disorders affecting the S2-S4 segments of the spinal chord.

Rectoanal Inhibitory Reflex

The rectoanal inhibitory reflex (RAIR) represents the relaxation of the IAS in response to distension of the rectum. This was first described by Gowers23 in 1877 and documented by Denny-Brown and Robertson24 in 1935. It is believed that this permits fecal material or flatus to come into contact with specialized sensory receptors in the upper anal canal.25 This sampling process, the sampling reflex, creates an awareness of the presence of stool and a sense of the nature of the material present. It is believed that this process of IAS relaxation with content sampling is instrumental in the discrimination of gas from stool and the ability to pass them independently.25 The degree to which IAS relaxation occurs seems to be related to the volume of rectal distension more so in incontinent patients than in constipated or healthy control patients.26 Lower thresholds for the RAIR have been found to be associated with favorable response to biofeedback therapy in patients with fecal incontinence for formed stool.27 The amplitude of sphincter inhibition is roughly proportional to the volume extent of rectal distension.

The RAIR is primarily dependent on intrinsic nerve innervation in that it is preserved even after the rectum has been isolated from extrinsic influences, following transaction of hypogastric nerves and the presence of spinal chord lesions. The inhibition response is in part controlled by nonadrenergic, noncholinergic mediators.28 The reflex matures quite early in that it is generally present at birth and has been detected in 81% of premature infants older than 26 weeks postmenstrual age.29 The reflex is destroyed in Hirschsprung’s disease when myenteric ganglion are absent. In addition, the reflex is lost after circumferential myotomy and after generous lateral internal anal sphincterotony.30 Saigusa et al.31 found that at an average of 23 months following closure of ileostomy after ileal pouch anal anastomosis, only 53% of patients maintained a positive RAIR as compared with 96% preoperatively. The incidence of nocturnal soiling was significantly greater: 72% in those who did not have preserved, or recovered RAIR as compared with 40% who had postoperative preserved RAIR.31

The RAIR seems to be nearly abolished in the early postoperative period after low anterior resection for cancer. In a study involving 46 patients, O’Riordain et al.32 found that the RAIR that had been present in 93% of patients preoperatively was only present in 18% 10 days after low anterior resection. However, at 6–12 months, the RAIR was intact in 21% of patients and this increased to 85% after 2 years.33 Similarly, van Duijvendijk et al.,25 in a study of 11 patients, found RAIR present in only 36% of patients after undergoing total mesorectal excision for carcinoma at 4 months after operation. However, 81% of patients had a detectible RAIR at 12 months after surgery. The degree to which IAS relaxation occurs appears to be related to the volume of rectal distension more so in incontinent patients than in constipated or healthy control patients.33

Loss of RAIR is often a consequence of restorative proctocolectomy. Saigusa et al.31 found that the RAIR was present in only 53% of double-stapled ileal pouch anal anastomosis patients at a mean of 23 months after closure of the ileostomy. Preservation of the RAIR correlated with less nocturnal soiling.

The RAIR in children can be elicited even when general anesthetic agents or neuromuscular blockers are used. Glycopyrrolate, an anticholinergic, seems to inhibit RAIR.34

Disturbances in the RAIR seem to be involved in the incontinence that is associated with systemic sclerosis. Heyt et al.35 found that 25 of 35 patients (71.4%) with systemic sclerosis demonstrated an impaired or absent RAIR compared with none of 45 controls. Impaired RAIR was closely correlated with fecal incontinence in that 11 of 13 (84%) of incontinent systemic sclerosis patients exhibited an impaired RAIR.

Rectoanal Excitatory Reflex

The rectoanal excitatory reflex (RAER), or inflation reflex, is the contraction of the EAS in response to rectal distension. Rectal distension sensation is likely transmitted along the S2, S3, and S4 parasympathetic fibers through the pelvic splanchnic nerves.36 However, on the motor side, a pudendal nerve block abolishes the excitatory reflex suggesting that pudendal neuropathy may interfere with the RAER. Common methodologies
for assessing the integrity of the pudendal nerve involve both single fiber density (SFD) of the EAS and pudendal nerve terminal motor latency (PNTML). However, derangement of the distal RAER was shown by Sangwan et al. to compare favorably with these more traditional and discomforting methodologies as an indicator of neuropathic injury to the EAS. It would seem that patients that have both an abnormal PNTML and an abnormal distal RAER do not require further study with SFD.

Mechanical Factors of Continence and Defecation

Anorectal Angle and Flap Valve

As a part of the pelvic floor musculature, the puborectalis arises from the pubic bone and passes horizontally and posteriorly around the rectum as the most medial portion of the levator ani muscle. This forms a U-shaped sling around the rectum near its anatomic junction with the anus, pulling the rectum anteriorly, and giving rise to the so-called anorectal angle. There are differences of opinion as to whether the puborectalis and anorectal angle are truly important in maintaining continence. Unlike the fine control of the external and internal sphincter muscles, the puborectalis sling is believed to be more involved with gross fecal continence. Parks postulated a mechanism by which this takes place. As intraabdominal pressure is increased—such as with sneezing, coughing, or straining—and the force is transmitted across the anterior wall of the rectum at the anorectal angle, the underlying mucosa is opposed against the upper anal canal, creating a flap-valve mechanism that prevents stool from passing to the lower anal canal and preserving continence. Yet other authors have disputed this flap-valve mechanism and downplayed the role and reliability of measuring the anorectal angle. Bannister et al., in a study of 29 patients including 14 patients with incontinence, found no evidence of a flap valve in the normal subjects by using manometric measurements during increasing intraabdominal pressures. However, in the incontinent patients, the manometric pressures were consistent with a flap valve. Yet, subjects still had leakage of stool, questioning the contribution to overall continence. Bartolo and colleagues also used manometric and EMG measurements in 13 subjects both at rest and during Valsalva, demonstrating a similar increase in rectal and sphincter pressures and puborectalis EMG recordings. Yet, with concomitant barium studies, the anterior rectal wall separated from the mucosa, allowing contrast to fill the rectum. The authors proposed that the puborectalis functions more like a sphincter rather than contributing to the flap-valve mechanism.

Furthermore, quantifying the anorectal angle and relating that to patient symptoms has resulted in mixed views. Jorgensen and colleagues noted significant interobserver variation in anorectal angle measurements among three interpreters but good intraobserver consistency, suggesting that variation in anorectal angle measurements may be attributable to subjective interpretation of the rectal axis along the curved rectal wall. The authors of another study, assessing the reproducibility of anorectal angle measurement in 43 defecating proctograms, found significant intra- and interobserver variations, and concluded that the anorectal angle is an inaccurate measurement. Jorge and associates measured the anorectal angle during rest, squeeze, and push in 104 consecutive patients and also found highly significant differences in each measurement category.

Reservoir

As an additional part of the continence mechanism, the rectum must be able to function as a temporary storage site for liquid and solid stool. With passage of the fecal stream into the rectum, the pliable rectal walls are able to distend and delay the defecation sequence until an appropriate time. This process relies both on rectal innervation to sense and tolerate the increasing volume of stool (capacity), as well as maintain a relatively low and constant pressure with increases in volume (compliance). Extremes of either of these components can lead to fecal incontinence through decreased accommodation or overflow states. Although decreased compliance has been demonstrated more often in patients with fecal incontinence, it has also been shown to occur as a normal consequence of aging. In addition, Bharucha and associates in a study of 52 women with fecal incontinence, demonstrated that the rectal capacity was reduced in 25% of women, and these lower volume and pressure thresholds were significantly associated with rectal hypersensitivity and urge fecal incontinence. Furthermore, after low anterior resection for cancer, those patients with resultant lower rectal compliance and lower rectal volume tolerability (capacity) have been associated with higher rates of fecal incontinence.

Normal Defecation

The awareness of the need to defecate occurs in the superior frontal gyrus and anterior cingulate gyrus. The process begins with movement of gas, liquid, or solid contents into the rectum. Distention of the rectum leads to stimulation of pressure receptors located on the puborectalis muscle and in the pelvic floor muscles, which in turn stimulate the RAIR. The IAS relaxes allowing sampling of contents. If defecation is to be deferred, voluntary contraction of the EAS and levator ani muscles occurs and the rectum accommodates with relaxation after an initial increase in pressure. When the anal canal is deemed to have solid contents and a decision to defecate is made, the glottis closes, pelvic floor muscles contract, and diaphragm and abdominal wall muscles contract, all increasing abdominal pressure. The puborectalis muscle relaxes, resulting in straightening of the anorectal angle, and the pelvic floor descends slightly. The EAS relaxes and anal canal
contents are evacuated. Upon normal complete evacuation, the pelvic floor rises and sphincters contract once more in a “closing reflex.”

Pathologic Conditions

Incontinence

Incontinence is the inability to defer the passage of gas, liquid, or solid stool until a desired time. Numerous alterations in anorectal physiology can lead to incontinence and many patients have more than one deficit. Structural defects in the IAS or EAS muscles occur because of obstetric injury, trauma, or anorectal surgery. The keyhole deformity is a groove in the anal canal allowing the seepage of stool or mucus. Originally described as a complication after the posterior midline fissurectomy or fistulotomy, it can also occur with lateral IAS defects. Intact sphincter muscles with impaired neurologic function, because of pudendal nerve damage or systemic disorders such as diabetes, can also result in incontinence, especially if the impaired sphincter is further stressed by diarrhea or irritable bowel syndrome.

Abnormal rectal sensation can lead to incontinence in two ways. Conditions such as proctitis caused by inflammation or radiation can result in hyperacute sensation. The rectum fails to accommodate and the reservoir function is impaired leading to urgency and frequency stooling. Fragmentation of stools is often described by patients after low anterior resection, particularly if the pelvis has been radiated as in the case of adjuvant therapy for the treatment of rectal cancer. In the case of blunted sensation, because of a large rectocele, megarectum, or neurogenic disorders, the rectum becomes overdistended and overflow incontinence occurs.

The majority of patients with rectal prolapse are incontinent. Chronic stretching of the anal sphincters from full-thickness prolapse leads to a patulous anus through which gas and liquid stool easily leak. A reflex relaxation of the IAS may also occur as the rectal wall descends toward the anal canal. Patients with mucosal prolapse may have seepage of mucus or small amounts of liquid stool. Correction of the prolapse can resolve the incontinence if anal sphincter tone sufficiently returns. Age and duration of prolapse can affect this.

Obstructed Defecation

Suspected Enterocele or Rectocele (Obstructed Defecation)

Patients with symptoms of enterocele or rectocele describe prolonged straining at defecation, with a sensation of partial or complete blockage (frequently a “closed trap door” preventing passage of stool). Defecography can demonstrate the presence of a rectocele or enterocele, suggest the presence of a peritoneocele, and clarify contributing disorders such as a nonrelaxing pelvic floor, rectal intussusception or prolapse, and potentially uterovaginal prolapse.

Rectocele

A rectocele is defined as greater than 2 cm of rectal wall outpouching or bowing while straining, and can precede or accompany rectal intussusception. The rectocele can prevent passage of stool both by obstructing the anal orifice and by acting as a diverticulum to sequester stool. Patients with rectoceles often complain of the need for frequent sequential episodes of defecation, and even for manual compression or splitting of the anterior perineum or posterior vagina in order to completely evacuate. Additionally, patients may experience incontinence with relaxation, leading to reduction of the rectocele and return of the sequestered stool to the lower rectum.

Van Dam and associates investigated the utility of defecography in predicting the outcome of rectocele repair. Rectocele size, barium trapping, intussusception, evacuation, and perineal descent were measured during defecography examinations of 74 consecutive patients with symptomatic rectoceles. The patients then underwent a transanal/transvaginal repair, followed by 6-month-postoperative defecography and reassessment of the five most common presenting symptoms (excessive straining, incomplete evacuation, manual assistance required, sense of fullness, bowel movement less than three times per week). No postoperative defecograms demonstrated a persistent or recurrent rectocele; however, one-third of patients had a poor result based on persistent symptoms. There was no association between defecography measurements and outcome of the repair. Still, the authors concluded that defecography serves three major purposes in the evaluation of a rectocele: preoperative evidence of its presence and size, documentation of additional pelvic floor abnormalities, and an objective assessment of postoperative changes.

An abnormal increase in perineal descent (typically greater than 2 cm) has been described among both incontinent patients and continent patients who strain during defecation. These conflicting data underscore the poorly understood relationship between neuropathic pelvic floor damage and symptomatology.

Bartolo and associates evaluated patients with perineal descent using manometric, radiographic, and neurophysiologic studies. When comparing 32 patients with incontinence and increased perineal descent with 21 patients with obstructed defecation and increased perineal descent, the authors found no significant difference in the extent of perineal descent or neuropathic damage to the EAS. Patients who were incontinent had lower manometric pressures (both resting and squeeze pressures) whereas those with obstructed defecation had normal manometric pressures. In a separate study, these authors also found that incontinent patients with increased perineal descent had severe denervation of both the puborectalis and the external sphincter compared with continent patients with increased perineal descent, who had
partial denervation of the external sphincter only.46 Miller and colleagues47 evaluated sensation in two similar patient groups. Patients who were frankly incontinent actually had less perineal descent than continent patients with descent, but had severely impaired anal sensation. Berkelmans et al.48 tried to determine whether women with increased perineal descent and straining at stool were at risk for future development of incontinence. The authors identified 46 women with perineal descent who strained during defecation but were continent. Twenty-four of the 46 were followed after 5 years and 13 of these (54%) had developed fecal incontinence, compared with 3 of 20 (15%) control patients. During their initial evaluation, the patients who previously strained and later developed incontinence had significantly greater perineal descent at rest and less elevation of the pelvic floor during maximal sphincter contraction than the women who strained but did not develop incontinence.

Thus, perineal descent may be a predictor of incontinence among patients with denervation of both the external sphincter and the puborectalis, and in patients with impaired anal sensation. Among patients with constipation, perineal descent and straining at stool may predict future fecal incontinence.

**Dyskinetic Puborectalis**

Dyskinetic puborectalis, paradoxical puborectalis, nonrelaxing puborectalis, and anismus are terms that describe the absence of normal relaxation of pelvic floor muscles during defecation, resulting in rectal outlet obstruction.49 Once diagnosed, dyskinetic puborectalis is usually treated with biofeedback and bowel management. Patients who fail conservative treatment have been offered botulism toxin injections into the puborectalis muscle with limited success.50

**Continence**

The dynamic intention of all the aforementioned anatomy and physiology ensures continence. It does not follow that a deficit in any one area ensures incontinence. Continence achieved in patients with an ileal pouch is proof the rectum is not essential. An intact and functional puborectalis muscle can provide continence in the patient with pediatric imperforate anus, but incontinence can ensue during adulthood. Even profound deficits do not necessarily lead to incontinence if stool consistency is solid, whereas minor deficits can easily lead to incontinence and gas. To determine and treat abnormal fecal incontinence requires a systematic approach focusing on identifying the specific deficits present, applying appropriate testing to elucidate anal physiology and anatomy, and then directing therapy accordingly.

**References**


