



Epidemiology, Anorectal Anatomy, Physiology and Pathophysiology of Continence

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2.1 Introduction and Epidemiology

Continence can be defined as the ability to retain flatus, liquid, or solid stools during normal daily life, including while physically exercising, coughing, sneezing, and changing position [1].

The anal canal is normally closed at rest and during sleep due to the steady activity of the internal anal sphincter (IAS) supported by the tonic activity of the external anal sphincter (EAS) and puborectalis.

The integrity of the continence and defecation mechanism is a multifactorial process that involves somatic and visceral functions and allows postponing defecation when necessary and avoiding the uncontrolled passage of feces or gas, causing patients to feel embarrassed with a negative impact on lifestyle, work, and interpersonal relationships [2].

Considering the heterogeneity of the problem, the incidence and prevalence of fecal incontinence (FI) are difficult to establish because they depend on the type and frequency of incontinence, age, and gender (Table 2.1) [3]. In fact, if the leading cause in women is post-obstetric injury, in men and the elderly, other factors such as anorectal surgery and diabetes mellitus must be considered [4, 5]. Moreover, the use of many terms to define FI has generated confusion and favored this trend.

Several authors have tried to quantify this phenomenon without achieving a definitive percentage [6–8]. According to Sharma et al. [6], the prevalence of FI ranges between 1.4% and 19.5%, whereas in the systematic review by Ng et al. [7] it has a median prevalence of 7.7%, without any difference between genders, but with a greater percentage in people older than 90 (15.9% vs. 5.7%) compared with people 15–34 years old.

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Table 2.1 Pathophysiological mechanisms leading to fecal incontinence

Structure		
Anal sphincter muscle	Obstetric injury Hemorrhoidectomy, anal dilatation, secondary to neuropathy	Sphincter weakness Loss of sampling reflex
Rectum	Inflammation, IBD, radiation, prolapse, aging, IBS	Sphincter weakness Loss of sampling reflex
Puborectalis muscle	Excessive perineal descent, aging, trauma	Obtuse anorectal angle Sphincter weakness
Pudendal nerve	Obstetrical/surgical injury Excessive straining/perineal descent	Sphincter weakness Sensory loss, impaired reflexes
CNS, spinal cord, ANS	Spinal cord injury, head injury, back surgery, multiple sclerosis, diabetes, stroke, avulsion injury	Loss of sensation, impaired reflexes, secondary myopathy, loss of accommodation
Function		
Anorectal sensation	Obstetrical, CNS, ANS injury	Loss of stool awareness Rectoanal agnosia
Fecal impaction	Dyssynergic defecation	Loss of stool awareness Rectoanal agnosia
Stool characteristics		
Volume and consistency	Infection, IBD, IBS, drugs, metabolic	Diarrhea and urgency Rapid stool transport Impaired accommodation
Irritants	Bile salt malabsorption, laxatives	Diarrhea
Hard stools/retention	Dyssynergia/drugs	Fecal retention with overflow
Miscellaneous		
Physical mobility/cognitive function	Aging, dementia, disability	Multifactorial changes
Psychosis	Willful soiling	Multifactorial changes
Drugs	Anticholinergics Laxatives Antidepressants Caffeine/muscle relaxants	Constipation Diarrhea Alter sensation/constipation Relaxes sphincter tone
Food intolerance	Lactose/fructose/sorbitol	Diarrhea/flatus Malabsorption

IBD, inflammatory bowel disease; *IBS*, irritable bowel syndrome; *CNS*, central nervous system; *ANS*, autonomic nervous system

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The higher prevalence among the elderly may be due to the physiological effects of aging on continence, such as impaired rectal sensation or dysfunction of both the IAS and EAS, which become thicker [9], as well as polypharmacy. In fact, the abuse of laxatives taken to avoid fecal impaction or constipation can exacerbate the condition. In this context, the highest prevalence currently recorded is among nursing home residents, where involvement reaches up to 50% [10].

The difficulty in establishing the extent of FI could be due to embarrassment in reporting the symptoms, as occurs with other proctological diseases [11]. Brown et al. [12], in an internet-based questionnaire study of 5817 women, showed that one-fifth of women over 45 in the USA suffer from at least one episode of FI per

year. A lack of knowledge and awareness of the problem as well as economic status represent further important barriers to patients' interaction with the health-care system.

2.2 Anatomy and Physiology of Continence

The continence process can be defined as a somatovisceral reflex and involves complex coordinated activity between several muscle groups – including the sphincter complex (Fig. 2.1) [3], ligaments and fascia of the pelvic floor—and rectal compliance, stool consistency and volume, and cognitive function. Thus, an abnormality in any of these factors may result in FI. In fact, up to 80% of patients with anal incontinence have more than one pathological abnormality in anorectal physiology.

The IAS (Fig. 2.2) [13] represents the continuation of the inner circular muscle layer of the rectum [14]. This smooth muscle under the control of the involuntary nervous system (sympathetic and parasympathetic) contributes to 80% of the resting anal pressure—normally 50–70 mmHg—and is the main barrier to the involuntary passage of gas and feces. The remaining 15–20% of resting anal pressure is regulated by the EAS, puborectalis, and hemorrhoids. All three contribute to the high-pressure zone, which is appreciated along the entire length of the anal canal and has 30% higher than found in the rectum. Interestingly, according to Penninckx et al. [15], the resting anal pressure is generated by the myogenic tone of the IAS (10%), by nerve-induced activity (45%), by the EAS (35%), and by the hemorrhoids (15%).

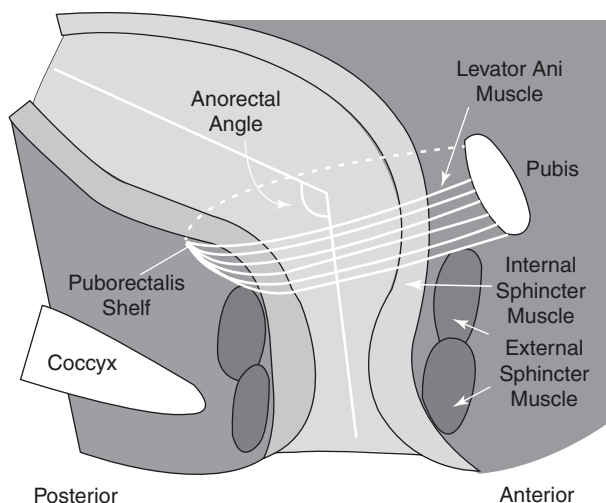


Fig. 2.1 Structure of the anorectum. The internal anal sphincter muscle provides between 70% and 85% of resting sphincter pressure. The anorectal angle, approximately 90 degrees at rest, becomes more obtuse during defecation. (Reproduced from [3] with permission from Elsevier)

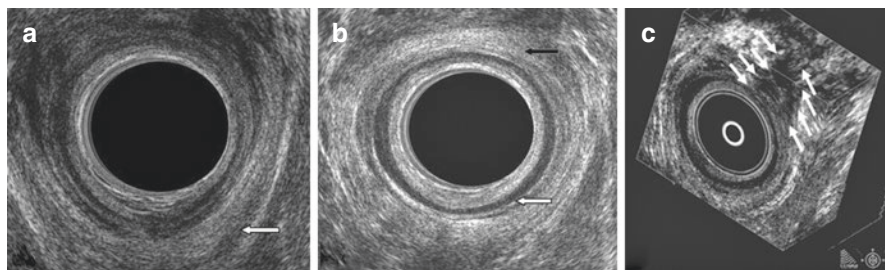


Fig. 2.2 (a) Endoanal scan demonstrating the U-shaped puborectalis muscle, which attaches to the pubic rami anteriorly. (b) Endoanal scan demonstrating the internal anal sphincter (*white arrow*) and the external anal sphincter (*black arrow*). (c) Three-dimensional endoanal ultrasound demonstrating the circumference/width as well as length of the anal sphincter defect. (Reproduced from [13] with permission from British Institute of Radiology)

The length of the IAS differs between genders and is smaller in women (2.0–3.0 cm vs. 2.5–3.5 cm), while its thickness varies between 2 and 4 mm. The IAS has a resting tone with cyclic variations, or rather short and ultrashort waves (1.5–3 cycles per minute). These waves are extremely important for reflex sampling. In fact, an intermittent relaxation of the IAS, lasting about 10–20 s, occurs approximately seven times per hour [16, 17] and allows rectal contents to come into contact with the sensitive mucosa of the anal canal.

The EAS (Fig. 2.2) [13] is a striated muscle that receives innervation from the pudendal nerve (S2, S3, S4), and its inhibition allows defecation [18]. It has a thickness of 4 mm and length of about 2.7 cm, although it is shorter anteriorly in women (1.5 cm), and it has three components: the subcutaneous, superficial, and deep anorectal elements. The EAS is responsible for the guarding reflex, a low spinal reflex, which contracts for 20–30 s after fecal contents arrive in the rectal ampulla.

The levator ani (Fig. 2.2) [13] has a pelvic floor support function and consists of three bundles: the pubococcygeus anteriorly, the iliococcygeus posteriorly, and the puborectalis inferiorly. The most important component for continence is the puborectalis muscle (PRM), which is a U-shaped loop muscle that forms a sling around the rectum, with an anorectal angle of approximately 90° that is at the basis of the flap-valve theory proposed by Parks [19], to which the three portions of the EAS also contribute. During normal defecation, relaxation of the PRM causes the anorectal angle to become more obtuse and widen ($>130^\circ$), facilitating the passage of the rectal content.

Recently, Broens et al. [20] introduced the concept of the “puborectalis continence reflex”, demonstrating that the PRM has also an involuntary contraction that is significantly stronger than the voluntary one with an equally increased pressure zone. This could confirm the role of the PRM in supporting dysfunctions of the other sphincter muscles in patients with impaired continence but who have not developed FI.

The conjoined longitudinal muscle, interposed between the IAS and EAS, consists of smooth muscle together with striated fibers from the levator ani, and its role

is controversial. It has a thickness of about 2.5 mm and seems involved in the shortening and widening of the anal canal. Shafik originally defined it as an “evertor ani muscle” [21] for its apparent action of eversion of the anal orifice. In addition, some studies have recently suggested its role in supporting continence after lateral internal sphincterotomy [22].

In addition to the muscular component, several other factors contribute to anal continence. The recto-anal inhibitory reflex (RAIR), also known as relaxation of the IAS, is certainly one of the best-established mechanisms linked to defecation and among the first to be activated.

It is regulated by the enteric nervous system, which is why it is absent in Hirschsprung’s disease, but it is still present after rectum or anus denervation or recovers with time [23].

The distension of the rectal wall caused by the arrival of the intestinal contents, made possible by the action of another important functional apparatus—the “recto-sigmoid sphincter” [24, 25]—thanks to colonic high-amplitude-propagated contractions, stimulates the multiple pressure receptors, allowing contact with the sensitive area of the anal canal. In this area, the stool contents are in contact with specialized sensory organs such as the Krause end-bulbs, Golgi-Mazzoni’s bodies and genital corpuscles, the sparse Meissner’s corpuscles, and Pacinian corpuscles [26, 27].

At this point, continence is maintained by the EAS (rectoanal excitatory reflex [RAER]) that may avoid anal leakage, and, to occur, intra-abdominal pressure should increase, with consequent relaxation of the PRM, widening of the anorectal angle, and rectification of the rectum. The reflex contraction of the EAS with the concomitant contraction of the PRM to restore the anorectal angle ends the process. If defecation is deemed inappropriate, it can be deferred by the voluntary contraction of both EAS and PRM [28]. In particular, the PRM induces closure of the pelvic diaphragm by generating horizontal forces with a consequent decrease of the anorectal angle. Both RAIR and RAER make up the sampling reflex, which has the fundamental function of advancing a part of the rectal content in the upper anal canal without causing an episode of incontinence.

Under physiological conditions, the rectum can passively undergo dilation without pressure changes. At a volume of approximately 200 mL, a sensation of urgency is perceived, with a maximum tolerated volume ranging from 300 to 500 mL.

Pathological alterations of the rectum, such as inflammatory bowel disease, in which rectal compliance decreases, or Hirschsprung’s disease, in which compliance is increased, favor altered continence.

Hemorrhoids are vascular cushions contributing to almost 15% of the anal resting pressure, and their importance has been seen in transient or permanent soiling episodes that can occur after excisional hemorrhoidectomy [29].

The volume and consistency of feces are essential, especially in elderly patients or patients with an already impaired continence, such as after low anterior resection [5].

Lastly, it is not only the peripheral nervous system with the sympathetic (L1–L3) and parasympathetic (S2–S4) components that is involved, but several studies have highlighted the role of the central nervous system. In fact, the sensation of rectal

filling or urgency has been shown to be associated with areas such as the insula, thalamus, secondary somatosensory cortex, or anterior cingulate gyrus [30], whereas Brodmann area 4, the primary motor cortex, seems involved in anal and rectal responses [31].

Onuf's nucleus, positioned at the level of the ventral horn gray matter of S2–S4, is in communication with the upper motor neurons responsible for voluntary sphincter complex contraction, usually located in the parasagittal motor cortex [32].

2.3 Pathophysiology of Fecal Incontinence

Given the complexity of the continence mechanism, several target areas of injuries can lead to the development of FI. Moreover, it appears that more than 80% of patients with FI have more than one alteration of the continence mechanism [33].

Depending on the type of muscle injury, FI can have different manifestations. Urge incontinence, or the loss of stools despite a voluntary attempt to avoid it, is caused by a lesion of the EAS. When the IAS is involved, it is referred to as passive incontinence, with involuntary loss of liquid or stool without awareness. Both conditions are very common after obstetric trauma, which can also be caused by the EAS, IAS, and the pudendal nerve being stretched, compressed, or suffering an ischemic injury (Fig. 2.2) [13]. The pudendal nerve, certainly the most studied but not the only nerve involved in the continence mechanism, can be damaged during childbirth at the exit from Alcock's canal, where its course is predominantly fixed on the pelvic sidewall [34].

Episiotomy appears to be related to damage to the sphincter complex, even if some studies show conflicting opinions [35, 36]. In particular, midline posterior episiotomy has been correlated with a higher incidence of sphincter trauma [37].

According to Dudding et al. [38], episiotomy and instrumental delivery, fetal occipito-posterior presentation, a prolonged second stage of labor, and birth weight greater than 4 kg are risk factors for injury. Interestingly, most women develop FI after menopause, maybe due to the deterioration of anorectal function with aging or withdrawal of hormonal input. Consequently, sphincter damage represents the first step for developing FI with the overlap of other factors [39].

Urge incontinence can also be secondary to anorectal surgery (hemorrhoidectomy, sphincterotomy, surgery for fistulas), albeit more frequently in men, with the contemporary loss of the sampling reflex.

Another component of the sphincter complex that can be damaged is the PRM, often following an accidental trauma, perineal descent, or aging, with loss of functionality of the anorectal angle.

Often, FI and constipation may coexist, and in this case we define incontinence as fecal seepage or the involuntary post-defecation loss of stool [40]. Fecal seepage results from an impaired rectal sensation plus an inappropriate increase of anal sphincter pressure with the contraction of the EAS after excessive straining.

In men, the causes of FI are less defined. Radical prostatectomy with consequent radiotherapy can lead to sphincter fibrosis, myenteric plexus degeneration, and a

reduction of the functionality of the EAS, which becomes thicker with reduced rectal compliance, especially in the elderly. The same phenomenon occurs during radiotherapy for anal or rectal cancer.

When the reservoir function of the rectum is no longer optimal, increased intrarectal pressure can cause FI. This scenario can occur in patients with inflammatory bowel disease, radiation proctitis, hysterectomy, rectal cancer, or spinal cord injury.

Among the functional mechanisms, the loss of anorectal sensation is certainly the most frequent.

In fact, an impaired anorectal sensation, which often occurs in children and the elderly, can lead to fecal impaction and a consequent fecal overflow. Some of the most common causes are represented by neuropathies such as multiple sclerosis, diabetes mellitus, and spinal cord injury or by inadvertent injury during colorectal surgery. Central nervous system diseases, such as Parkinson's disease, can alter cognitive functions, leading to pathological toilet habits due to the inability to carry out daily activities.

Hellström et al. [41] highlighted the influence of dementia on anal continence in a random sample of 485 subjects selected from the 85-year-old inhabitants of Gothenburg, reporting its presence in 34.8% of demented subjects and 6.7% of non-demented subjects.

Furthermore, fecal impaction, common in patients with obstructed bowel syndrome or pelvic floor dyssynergy, can result in stool leaks that bypass the impaction owing to persistent relaxation of the IAS. Overflow incontinence can also be present in patients with congenital malformations [42].

Another very common condition is soiling, which often occurs after anorectal surgery or in patients with obstructed defecation. Up to 63% of patients with grade II–IV hemorrhoidal disease or rectal prolapse may have soiling [43].

Finally, the change in stool volume and consistency due to inflammatory bowel disease, drugs (such as laxatives in the elderly), food intolerance, or metabolic disorders can cause diarrhea and urgency, fecal impaction, and malabsorption. In the case of diarrhea, increased activity of high-amplitude-propagated contractions can cause overwhelm of the reservoir capacity of the rectum [44, 45]. These motor complexes, extremely represented at the level of the rectosigmoid, are significantly increased in patients with urge incontinence [46].

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