Anorectal Disorders

Satish S. C. Rao,1 Adil E. Bharucha,2 Giuseppe Chiarioni,3,4 Richelle Felt-Bersma,5 Charles Knowles,6 Allison Malcolm,7 and Arnold Wald8

1Division of Gastroenterology and Hepatology, Augusta University, Augusta, Georgia; 2Department of Gastroenterology and Hepatology, Mayo College of Medicine, Rochester, Minnesota; 3Division of Gastroenterology of the University of Verona, Azienda Ospedaliera Universitaria Integrata di Verona, Verona, Italy; 4Division of Gastroenterology and Hepatology and UNC Center for Functional GI and Motility Disorders, University of North Carolina at Chapel Hill, Chapel Hill, North Carolina; 5Department of Gastroenterology/Hepatology, VU Medical Center, Amsterdam, The Netherlands; 6National Centre for Bowel Research and Surgical Innovation, Blizard Institute, Queen Mary University of London, London, United Kingdom; 7Division of Gastroenterology, Royal North Shore Hospital, and University of Sydney, Sydney, Australia; 8Division of Gastroenterology, University of Wisconsin School of Medicine and Public Health, Madison, Wisconsin

This report defines criteria and reviews the epidemiology, pathophysiology, and management of the following common anorectal disorders: fecal incontinence (FI), functional anorectal pain, and functional defecation disorders. FI is defined as the recurrent uncontrolled passage of fecal material for at least 3 months. The clinical features of FI are useful for guiding diagnostic testing and therapy. Anorectal manometry and imaging are useful for evaluating anal and pelvic floor structure and function. Education, antidiarrheals, and biofeedback therapy are the mainstay of management; surgery may be useful in refractory cases. Functional anorectal pain syndromes are defined by clinical features and categorized into 3 subtypes. In proctalgia fugax, the pain is typically fleeting and lasts for seconds to minutes. In levator ani syndrome and unspecified anorectal pain, the pain lasts more than 30 minutes, but in levator ani syndrome there is puborectalis tenderness. Functional defecation disorders are defined by ≥2 symptoms of chronic constipation or irritable bowel syndrome with constipation, and with ≥2 features of impaired evacuation, that is, abnormal evacuation pattern on manometry, abnormal balloon expulsion test, or impaired rectal evacuation by imaging. It includes 2 subtypes: dyssynergic defecation and inadequate defecatory propulsion. Pelvic floor biofeedback therapy is effective for treating levator ani syndrome and defecatory disorders.

Keywords: Anorectal Disorders; Fecal Incontinence; Constipation; Dyssynergic Defecation; Levator Ani Syndrome; Anorectal Pain; Biofeedback Therapy.

Anorectal disorders are defined by specific symptoms and, in the case of functional disorders of defecation, also with abnormal diagnostic tests. Our understanding of these disorders continues to evolve with the availability of newer techniques to characterize anorectal structure and function.1–3 Consequently, the distinction between “organic” and “functional” anorectal disorders may be difficult in individual patients.1–3

Anorectal disorders, such as fecal incontinence, are usually defined by specific symptoms, but functional disorders of defecation require symptoms and anorectal physiological testing.4 While bowel symptoms recorded by questionnaires and bowel diaries are correlated, some patients may not accurately recall bowel symptoms; hence, symptom diaries may be more reliable.

In this report, we examine the prevalence and pathophysiology of anorectal disorders, listed in Table 1, and provide recommendations for diagnostic evaluation and management. These supplement practice guidelines recommended by the American Gastroenterological Association7 and American College of Gastroenterology.8 We will not address anorectal symptoms secondary to a neurologic or systemic disorder. The revised diagnostic criteria include a minimum duration of symptoms that were selected arbitrarily to avoid the inclusion of self-limited conditions.

F1. Fecal Incontinence

Definition

Fecal incontinence (FI) is defined as the recurrent uncontrolled passage of fecal material for at least 3 months. We recognize that fecal staining of underwear may reflect poor hygiene, prolapsing hemorrhoids, or rectal prolapse rather than true FI, but for practical purposes it is included in the definition of FI. Clear mucus secretion must be excluded by careful questioning. Flatus incontinence is often included in the definition of anal incontinence but not in the current diagnosis of FI because it is difficult to define when isolated passage of flatus is abnormal. FI is often multifactorial and occurs in conditions that cause diarrhea, impair colorectal storage capacity, and/or weaken the pelvic floor (Table 2). FI is considered abnormal after toilet training has been achieved, generally around 4 years of age.9

Abbreviations used in this paper: ARM, anorectal manometry; CI, confidence interval; DRE, digital rectal examination; EMG, electromyography; FDD, functional defecation disorder; FI, fecal incontinence; IBS, irritable bowel syndrome; MRI, magnetic resonance imaging; OR, odds ratio.

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Disorders affecting rectal capacity and/or sensation reference time frame10,16,25 (1 year or past month), and fl

Pelvic not all16,24 studies reported a lower prevalence in African-across races in men.24 Interestingly, the majority of patients American than white women, but similar prevalence in men.24 Interestingly, the majority of patients seen in clinical practice are women.

Variations in the prevalence of FI among studies may reflect differences in survey methods, screening questions, reference time frame10,16,25 (1 year or past month), and definition of incontinence. Two studies evaluated the incidence of FI.23,25 In a community study (65 years and older), the incidence of FI at 4 years was 17%, with 6% having FI at least monthly.23 In a follow-up community study (50 years and older), the incidence of FI was 7.0%.20

**Epidemiology**

**Prevalence.** Several large community-based studies10-17 have suggested that FI is common, with a prevalence ranging from 7% to 15% in community-dwelling women, 18% to 33% in hospitals, and 50% to 70% in nursing homes.18,19 The prevalence is either comparable6,20 or lower in men than women.21,22 Some11,13,17,23 but not all16,24 studies reported a lower prevalence in African-American than white women, but similar prevalence across races in men.24 Interestingly, the majority of patients seen in clinical practice are women.

Table 1. Functional Anorectal Disorders

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<thead>
<tr>
<th>F. Functional anorectal disorders</th>
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<tr>
<td>F1. Fecal incontinence</td>
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<td>F2. Functional anorectal pain</td>
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<td>F2a. Levator ani syndrome</td>
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<td>F2c. Proctalgia fugax</td>
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<td>F3a. Dyssynergic defecation</td>
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<td>F3b. Inadequate defecatory propulsion</td>
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**Common Causes of Fecal Incontinence**

Table 2. Common Causes of Fecal Incontinence

<table>
<thead>
<tr>
<th>Anal sphincter weakness</th>
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<tr>
<td>Traumatic: obstetric, surgical (eg, hysterectomy, internal sphincterotomy, fistulectomy)</td>
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<tr>
<td>Nontraumatic: scleroderma, idiopathic internal sphincter degeneration</td>
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<td>Neuropathy</td>
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<td>Peripheral (eg, pudendal) or generalized (eg, diabetes mellitus)</td>
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<td>Pelvic floor disorders</td>
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<td>Rectal prolapse, descending perineum syndrome</td>
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<td>Disorders affecting rectal capacity and/or sensation</td>
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<tr>
<td>Inflammatory conditions: radiation proctitis, Crohn’s disease, ulcerative colitis</td>
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<tr>
<td>Anorectal surgery (pouch, anterior resection)</td>
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<tr>
<td>Rectal hypomotility</td>
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<td>Rectal hypersensitivity</td>
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<td>Central nervous system disorders</td>
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<td>Dementia, stroke, brain tumors, multiple sclerosis, spinal cord lesions</td>
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<tr>
<td>Psychiatric diseases, behavioral disorders</td>
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<tr>
<td>Bowel disturbances</td>
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<tr>
<td>Irritable bowel syndrome, post-cholecystectomy diarrhea</td>
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<td>Constipation and fecal retention with overflow</td>
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</table>

Impact on quality of life and psychosocial factors. Persons with FI report that poor bowel control restricts their social life; other issues pertain to toilet location, hygiene/odor issues, coping strategies, fear, physical activities, embarrassment, and unpredictability of bowel habits.27 Co-existent psychological problems may include anxiety and depression,28,29 poor self-esteem, and problems with sexual relationships.30 Quality of life issues can be evaluated by generic or disease-specific instruments, such as the Rockwood Fecal Incontinence Quality of Life Scale, modified Manchester Health Questionnaire, Fecal Incontinence and Constipation Assessment Quality of Life scale. FI symptoms can also be assessed by Pelvic Organ Prolapse/Incontinence Sexual Questionnaire—IUGA (International Urogynecology Association).31-34 There is a significant correlation between symptom severity and QOL in FI.31,35 FI was associated with increased mortality in some, but not all studies.36-38 but whether it is due to FI per se or conditions associated with FI (age and comorbidity) is unknown.16

**Etiology and risk factors for fecal incontinence.** The etiology of incontinence is often multifactorial. Therefore, it is more appropriate to focus on associated conditions, especially when they precede the onset of FI, and on risk factors for FI. In community surveys, bowel disturbances, especially diarrhea and rectal urgency, and the burden of chronic illness were more important and independent risk factors for FI than obstetric-related pelvic floor injury (eg, forceps use, complicated episiotomy).2,16,26,39-41 In a community-based cohort of 176 randomly selected women with FI and 176 without FI, the independent risk factors for FI were diarrhea (odds ratio [OR] = 53; 95% confidence interval [CI]: 6.1–471), cholecystectomy (OR = 4.21 95% CI: 1.2–15), current smokers (OR = 4.7; 95% CI: 1.4–15), rectocele (OR = 4.9; 95% CI: 1.3–19), stress urinary incontinence (OR = 3.1; 95% CI: 1.4–6.5), and body mass index (per unit, OR = 1.1; 95% CI: 1.004–1.1).41 Smoking, external sphincter atrophy, and obesity are also risk factors for FI.2,11,13,17,41 Other conditions associated with FI include advanced age, disease burden (comorbidity count, diabetes), anal sphincter trauma (obstetrical injury, prior surgery), and decreased physical activity.11,16,17,42,43 Several diseases that affect anorectal sensorimotor dysfunctions and/or alter bowel habits are also associated with FI in clinical practice (Table 2). Some of these conditions do not emerge as risk factors in community studies, possibly because their prevalence is relatively low. Consistent with the findings of community-based studies, the vast majority of women with FI who consult a physician might not have a neurologic or inflammatory disorder, but rather have bowel disturbances, typically diarrhea, often associated with a history of obstetric risk factors. However, neurologic deficit can only be identified with neurophysiologic tests, and these are not widely available.

The incidence of FI after vaginal delivery was 8% in a recent series.64 This may reflect improvements in obstetrical practices, including decreased use of instrumented
vaginal delivery (eg, forceps), less frequent and more selective use of episiotomy, and increased use of cesarean sections, although a Cochrane review showed no demonstrable difference between cesarean sections and vaginal deliveries.\textsuperscript{45} Third-degree (ie, involving the external anal sphincter) and fourth-degree lacerations (ie, extending through the external and internal anal sphincters) are strong risk factors for anal and fecal incontinence.\textsuperscript{46} A prospective National Institutes of Health trial identified a nearly 2-fold increased OR of FI for women with sphincter injury during childbirth compared with a control group.\textsuperscript{47} The risk is highest for instrument-assisted deliveries, with increased odds of 1.5 for anal incontinence and a higher risk with forceps than vacuum extraction.\textsuperscript{48} Among women in the community, the median age of onset of FI is in the 7th decade, that is, many decades after vaginal delivery\textsuperscript{11} and, therefore, how obstetric injury predisposes to FI is unclear.

Anorectal surgery for fistula, fissures, or hemorrhoidectomy and anorectal carcinoma can damage the sphincters.\textsuperscript{49} Impaired rectal compliance, as can occur with proctitis or after creation of a pouch, and fecal impaction with overflow diarrhea, can all cause FI.\textsuperscript{50–52}

Justification for Changes in Diagnostic Criteria

The earlier definition of functional fecal incontinence was cumbersome, did not facilitate management, and was seldom used in clinical practice or research studies. Therefore, we recommend the generic term *fecal incontinence*.

We recognize that newer sensitive diagnostic tools (eg, anal ultrasonography, pelvic magnetic resonance imaging [MRI], and high resolution/3-dimensional high-definition anorectal pressure topography) often reveal disturbances of anorectal structure and/or function in a majority of patients with FI, but their relationship to symptoms is unclear, especially as some have more dysfunction(s) than others. Therefore, it can be challenging to attribute symptoms with confidence to an organic or functional cause and more studies are needed.

Pathophysiology

**Physiological factors.** Continence is maintained by several mechanisms, including anatomical factors (endovascular cushions, integrity of anal sphincter, and puborectalis muscle), rectoanal sensation, rectal compliance, neuronal innervation, stool consistency, mobility, and psychological factors (Figure 1).\textsuperscript{53}

**Anorectal and pelvic floor musculature.** Anal sphincter weakness is the most frequently identified abnormality in FI. Among older women, approximately 40% had reduced anal resting pressure and 80% reduced squeeze pressure.\textsuperscript{54} Internal anal sphincter dysfunction is characterized by exaggerated spontaneous relaxation of the internal anal sphincter (sampling reflex)\textsuperscript{55} or decreased resting pressure.\textsuperscript{54,55} The latter is associated with structural disturbances, that is, defects (after obstetric injury) and/or thinning (scleroderma, advanced age). This is best visualized by ultrasonography. Among postpartum women, the severity of FI was greater in women with internal anal sphincter defects.\textsuperscript{56}

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**F1. Diagnostic Criteria\textsuperscript{a} for Fecal Incontinence**

1. Recurrent uncontrolled passage of fecal material in an individual with a developmental age of at least 4 years

\textsuperscript{a}Criteria fulfilled for the last 3 months. For research studies, consider onset of symptoms for at least 6 months previously with 2–4 episodes of FI over 4 weeks.

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**Figure 1.** Anatomy of the anal canal and rectum, which displays the key physiologic mechanisms for continence and defecation.
External anal sphincter weakness can result from one or more of the following factors: sphincter damage, neuropathy, myopathy, or reduced corticospinal input. In addition to the anal sphincters, the levator ani muscles also contribute to the pelvic barrier.\(^7\) One study suggested that the reduced inward traction exerted by the pubourethralis in patients with FI correlated more closely with symptoms than did squeeze pressures, and improved after biofeedback therapy.\(^6\) Whereas the anal sphincters and endovascular cushions seal the anal canal, thelevator ani and pubourethralis maintain continence of solid stool by a flap-valve action.\(^59-62\) Patients with excessive perineal descent have a more obtuse anorectal angle, suggesting that the flap valve that normally maintains continence when intra-abdominal pressure increases is impaired.\(^57\)

FI in men who generally have fecal soiling or leakage rather than gross incontinence may be associated with normal sphincteric function.\(^63-67\) Iatrogenic anal injury (eg, after perianal procedures); or dyssynergic defecation,\(^68\) wherein high anal resting pressure entraps feces during defecation and subsequently expels them\(^69\); radiation therapy\(^70\); or isolated weakness of the internal anal sphincter.

Rectal compliance and rectoanal sensation. Stool is often transferred into the rectum by colonic high-amplitude propagated contractions, which tend to occur after awakening or meals.\(^71\) Rectal distention by stool is associated with reduced rectal compliance and repetitive rectal contractions during rectal distention.\(^54,76\) Rectal capacity is also reduced in women with FI and associated with the symptom of urgency.\(^54,77\) In addition, rectal hypersensitivity cannot be entirely explained by disturbances in rectal compliance. Anal sphincter relaxation may occur during, or independent of, rectal distention, or along with colonic high-amplitude propagated contractions, which enables the anal lining to periodically “sample” rectal contents and ascertain whether rectal contents are gas, liquid, or stool.\(^74,76\) Sampling occurred less frequently in incontinent patients, perhaps depriving them of sensory information.\(^74\) In addition to anorectal dysfunctions, continence can also be affected by disturbances of stool consistency and/or delivery, impaired mental faculties, and mobility. These observations confirm that FI is a heterogeneous disorder and that patients often exhibit more than one deficit (Table 2).

**Clinical Evaluation**

**History.** It is essential to develop a rapport with FI patients and, with tact and skill, evaluate its severity, awareness for stooling, and conditions that predispose, including the type (solid, liquid, and/or gas), quantity, and frequency. Staining, soiling, and seepage reflect the nature and severity of FI.\(^70\) Soiling indicates leakage that is more extensive than staining of underwear and can be specified further (ie, soiling of underwear or furnishing/bedding). Seepage refers to leakage of small amounts of stool.

Characterization of bowel habit is important and the Bristol Stool Form Scale and bowel diaries can be useful.\(^79\) Constipation with fecal impaction is a significant risk in nursing homes.\(^76,80\) Factors that cause or exacerbate incontinence via loose stools (eg, laxatives, artificial sweeteners) and anorectal surgical procedures (eg, lateral sphincterotomy) or other mechanisms (eg, smoking, obesity) should be considered. Conversely, agents that cause constipation may predispose to fecal retention and overflow. Recognizing the timing of incontinence (eg, whether predominantly during or after events such as meals, bowel movements, exercise, or at night) can provide clues to etiology and management.\(^90\)

History taking should also include consideration of conditions that are a secondary cause of FI, such as multiple sclerosis, diabetic neuropathy, or scleroderma.

Urge vs passive FI can provide clues to the pathophysiology. Incontinence for solid stool suggests more severe sphincter weakness than liquid stool alone.\(^81\) Patients with urge incontinence have a sensation of the desire to defecate before leakage, but cannot reach the toilet on time. Conversely, patients with passive incontinence have diminished or no awareness of the desire to defecate before the incontinent episode. Patients with urge incontinence often have reduced squeeze pressures\(^82\) and/or squeeze duration,\(^83\) reduced rectal capacity, and increased perception of rectal balloon distention,\(^54,84\) whereas patients with passive incontinence often have lower resting pressures.\(^82,85\)

Several FI instruments—Wexner (Cleveland Clinic), Vaizey (St Marks), Rockwood, Fecal Incontinence and Constipation Assessment, and the bowel version of the International Consultation of Incontinence questionnaire—are currently used in clinical studies to rate the severity of FI\(^10,31,35,85-87\) and are validated instruments. Currently, success in therapeutic trials is typically defined as a 50% reduction in the number of episodes of FI or days per week\(^88\) although a patient’s perspective may differ and more meaningful outcome measures are required.\(^39,90\)

Physical examination including digital rectal evaluation. A multisystem and abdominal examination
and focused neurologic examination is often necessary in FI patients with neurologic symptoms.

A digital rectal examination (DRE) should be conducted in the left lateral position and before enemas or laxatives are given. Inspection may reveal scars from previous surgery or obstetric injury or a patulous sphincter or perianal fecal soiling or dermatitis. An absent anocutaneous reflex in response to gentle stroking of the perianal region suggests nerve impairment. After inspection, anorectal digital palpation should be conducted. This may reveal external anal sphincter and/or puborectalis weakness or defects, stool impaction, and presence of dyssynergia during simulated defecation. A meticulous DRE performed by an experienced examiner had a positive predictive value of 67% and 81% for identifying low resting and squeeze pressures, respectively.92

Diagnostic testing. Testing should be tailored to the patient’s clinical problem, severity, possible etiology, impact on quality of life, and response to medical management.

Endoscopy. Endoscopic assessment of the rectosigmoid mucosa or full colonoscopy with biopsies may be considered in patients with diarrhea or recent change in bowel habit.

Manometry evaluation. Anorectal manometry (ARM) assesses continence and defecatory mechanisms by determining:

1. resting anal pressure, which is predominantly (ie, approximately 70%) attributable to internal anal sphincter function;
2. squeeze pressure: the strength and duration of voluntary external anal sphincter contraction and puborectalis contraction;
3. presence of an internal anal sphincter inhibitory reflex;
4. threshold volume of rectal distention required to elicit the first sensation of distention, a sustained feeling of urgency to defecate, and the maximum tolerable volume;
5. whether attempted defecation is accompanied by increased intra-abdominal pressure and relaxation of the pelvic floor muscles (normal), or by paradoxical contraction of the pelvic floor muscles, which may be relevant to symptoms; and
6. rectal compliance can be evaluated by assessing the pressure-volume relationship during stepwise distention of a latex balloon, but it is preferable to do so with an infinitely compliant polyethylene balloon and a barostat.

The methods used for ARM, including solid-state probe, high-resolution ARM, and 3-dimensional high-definition ARM systems, and its measurements and interpretation are detailed elsewhere.93–95

Anal endosonography. Anal endosonography identifies anal sphincter thinning and/or defects that are often clinically unrecognized and may be amenable to surgical repair.96,97 Endosonography reliably identifies anatomic defects or thinning of the internal sphincter, whereas interpretation of external sphincter images may pose technical challenges. In contrast, 3-dimensional endosonography can measure the length and volume of the external anal sphincter and atrophy.98 Endoanal MRI,99,100 and vaginal ultrasound can provide additional information.101

Defecography. Defecography is useful only for selected patients with FI, particularly before surgery, to identify or confirm structural alterations of the pelvic floor.

Pelvic magnetic resonance imaging. MRI is the only imaging modality that can visualize both anal sphincter anatomy and global pelvic floor motion (ie, anterior, middle, and posterior compartments) in real time without radiation exposure.102 Endosonography is the first choice for anal sphincter imaging in FI because it is widely available and the internal sphincter is visualized more clearly. MRI is more useful for identifying external sphincter atrophy and a patulous anal canal, which is a marker of not only anal sphincter injury, but disturbances beyond sphincter injury, such as damage to the anal cushions or anal denervation.54,103

Neurophysiologic tests. Neurophysiological tests can characterize disturbances in the motor and sensory innervation of the anorectum and pelvic floor muscles. These tests include pudendal nerve terminal motor latencies, electromyography (EMG), rectoanal sensory tests, and motor evoked potentials. There are several methodological limitations to pudendal nerve terminal motor latencies, and the utility of this measurement has been questioned. Needle EMG can identify normal, neurogenic, or muscle injury.57,104 Recently, prolonged rectal and anal motor evoked potentials have been shown in a majority of FI patients, suggesting that neurophysiologic dysfunction plays an important role.105

Treatment

Management of FI must be tailored toward correction of clinical manifestations.

Bowel habit modification with dietary or pharmacological interventions. Loose stools are a major risk factor for FI.2,40 Correction of reversible factors like laxatives or other medications can help. Dietary trials (eg, low lactose or low fructose) in selected patients can normalize stool form. Among fiber supplements, only psyllium but not gum arabic or carboxymethylcellulose, improved FI compared with placebo.106 Loperamide given at an adequate dose (ie, 2–4 mg, 30 minutes before meals) can improve stool consistency and increase internal sphincter tone, thereby reducing incontinence.107 Diphenoxylate, combined with atropine, is an alternative to loperamide, but there may be anticholinergic side effects.108 In an open-label study of 18 patients, amitriptyline (20 mg daily), which has anticholinergic effects, improved FI in most patients.109

Patients with constipation, fecal impaction, and overflow incontinence often benefit from a program to increase emptying of the colorectum by various means. For example, a regimen consisting of a daily osmotic laxative (lactulose 10 mL twice daily) plus a weekly enema was useful in the
majority of elderly patients with FI, including those with dementia. However, loosening the stool may aggravate FI. Other measures aimed at improving rectal emptying, such as the use of suppositories or enemas, fiber supplementation, oral laxatives, and correction of any abnormal toileting behavior, or positioning and biofeedback may be helpful.

Rectal cleansing and anal plug devices. In patients who fail bowel modification and biofeedback therapy, periodic rectal cleansing is a practical solution. It should be considered particularly in patients with neurogenic bowel dysfunction. Plug devices may also be useful in some patients with seepage.

Biofeedback therapy. Biofeedback is based on the principle of operant conditioning or instrumental learning. One randomized controlled trial showed that biofeedback therapy is superior to Kegel exercises.

Surgical approaches. Anal sphincter repair, although well established, does not appear to be effective in the long-term. Sacral nerve stimulation and anal submucosal injection of dextranomer in stabilized hyaluronic acid [NASHA Dx], a bulking agent, are both approved by the US Food and Drug Administration for the treatment of FI. In the pivotal US multicenter study of sacral nerve stimulation, at 5-year follow-up, 76 of 120 (63%) patients were available, of whom, 36% reported complete continence and 89% were deemed a therapeutic success. However, this and nearly all other studies with sacral nerve stimulation have been uncontrolled. In a crossover study of 34 patients, the number of episodes of FI declined by 90% during stimulation vs 76% without stimulation.

In the pivotal trial of NASHA Dx (206 patients), the proportion of patients achieving a 50% FI episode reduction was higher for NASHA Dx (52%) than sham injections (31%), this response was sustained up to 3 years in some patients.

F2. Functional Anorectal Pain
Three types of functional anorectal pain disorders have been described: proctalgia fugax, levator ani syndrome, and unspecified. They are primarily distinguished on the basis of the duration of pain and the presence or absence of anorectal tenderness. Despite some differences, there is significant overlap among these conditions.

F2a. Diagnostic criteria for Levator Ani Syndrome.
Must include all of the following:
1. Chronic or recurrent rectal pain or aching
2. Episodes last 30 minutes or longer
3. Tenderness during traction on the puborectalis
4. Exclusion of other causes of rectal pain, such as inflammatory bowel disease, intramuscular abscess and fissure, thrombosed hemorrhoids, prostatitis, coccygodynia, and major structural alterations of the pelvic floor.

Criteria fulfilled for the last 3 months with symptom onset at least 6 months before diagnosis.

F2b. Diagnostic Criteria for Unspecified Functional Anorectal Pain
Symptom criteria for chronic levator ani syndrome but no tenderness during posterior traction on the puborectalis muscle

Justification for Changes in Diagnostic Criteria
The previous classification included chronic proctalgia that was subcategorized into levator ani syndrome, unspecified anorectal pain, and proctalgia fugax. Because chronic proctalgia includes many other conditions, it has been deleted, but the 3 subentities are retained. There are very limited published data on the duration of pain, but we believed the revised duration may facilitate better distinction between these entities. Reflecting the limited spatial discrimination of visceral pain in humans, the location of pain in proctalgia fugax has been revised to “rectum” instead of “anal canal or lower rectum.”

Pathophysiology. Physiological factors. Levator ani syndrome is hypothesized to result from spasm of pelvic floor muscles and elevated anal resting pressures. However, a recent randomized controlled study found features of dyssynergic defecation and a majority (85%) had levator muscle tenderness. The dysynergia reversed after successful biofeedback, suggesting that rectoanal incoordination may be a pathophysiological explanation for levator ani syndrome.

Clinical evaluation. Diagnosis is based primarily on the presence of characteristic symptoms and physical examination findings (see definition). Evaluation often includes sigmoidoscopy, ultrasonography, and pelvic imaging to exclude alternative diseases.

Treatment. Treatments include electrogalvanic stimulation; biofeedback training; muscle relaxants, such as methocarbamol, diazepam, and cyclobenzaprine; digital massage of the levator ani muscles; and sitz baths. However, only 2 randomized controlled trials have been reported. In one, 157 patients with chronic proctalgia received either electrical stimulation or digital massage of the levator ani...
F2c. Proctalgia Fugax

**Definition.** Proctalgia fugax is defined as sudden, severe pain in the rectal area, lasting for a few seconds to several minutes (rarely up to 30 minutes), and then disappearing completely. Pain is localized to the rectum in 90% of cases. Attacks are infrequent, typically occurring fewer than 5 times per year in 51% of patients. The pain has been described as cramping, gnawing, aching, or stabbing and may range from uncomfortable to unbearable. Almost 50% of patients had to interrupt their normal activities during an attack. The symptoms may awaken the patient from sleep.

**Epidemiology.** The prevalence of proctalgia fugax has ranged from 8% to 18% with no difference between the sexes. Symptoms rarely begin before puberty, but there have been cases reported in 7-year-old children.

**F2c. Diagnostic Criteria** for Proctalgia Fugax

Must include all of the following:

1. Recurrent episodes of pain localized to the rectum and unrelated to defecation
2. Episodes last from seconds to minutes, with a maximum duration of 30 minutes
3. There is no anorectal pain between episodes.
4. Exclusion of other causes of rectal pain, such as inflammatory bowel disease, intramuscular abscess and fissure, thrombosed hemorrhoids, prostatitis, coccygodynia, and major structural alterations of the pelvic floor.

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**Pathophysiology.** *Physiological factors.* The short duration and sporadic, infrequent nature of this disorder have made the identification of physiological mechanisms difficult, but abnormal smooth muscle contractions may be responsible for the pain. Two studies cited families in which a hereditary form of proctalgia fugax was found to be associated with hypertrophy of the internal anal sphincter and comorbid constipation. Attacks of proctalgia fugax are often precipitated by stressful life events or anxiety. In an uncontrolled unblinded study, a majority of patients were perfectionistic, anxious, and/or hypochondriacal.

**Clinical evaluation.** Diagnosis is based on the presence of characteristic symptoms as described and exclusion of anorectal and pelvic pathophysiology.

**Treatment.** For most patients, the episodes are so brief that remedial treatment is impractical and prevention is not feasible, and because it is harmless, treatment will normally consist of reassurance and explanation. However, patients with frequent symptoms will require treatment. A randomized controlled trial showed that inhalation of salbutamol was more effective than placebo for shortening the duration of episodes of proctalgia for patients in whom episodes lasted 20 minutes or longer.

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**F3. Functional Defecation Disorders**

**Definition.** Chronic constipation is commonly classified as either slow colonic transit or outlet dysfunction, although some patients may have neither and others fulfill criteria for both. A large subset of outlet dysfunction has a functional defecation disorder (FDD), which is characterized by paradoxical contraction or inadequate relaxation of the pelvic floor muscles during attempted defecation and/or inadequate propulsive forces during attempted defecation. These disorders are frequently associated with symptoms such as excessive straining, feeling of incomplete evacuation, and digital facilitation of bowel movements. However, symptoms (eg, digital disimpaction, anal pain) do not consistently distinguish between patients with FDDs from those without. Thus, the criteria for FDDs must rely on both symptoms and physiological testing.

Several investigators have described the association of paradoxical anal contraction with constipation and have described dyssynergia patterns. Likewise, studies have shown inadequate propulsive forces as identified by decreased or absent intrarectal pressure during attempted defecation. These patients are clinically indistinguishable from patients with dyssynergic defecation. Recently, a large controlled study showed that dyssynergic defecation, inadequate propulsive forces, and a hybrid of both disturbances were uncorrelated, suggesting that the pathophysiology of dyssynergic defecation and inadequate propulsive forces are distinct. Also, these patterns are observed in asymptomatic controls, and by themselves they have limited utility for discriminating between health and defecatory disorders. Hence, FDDs are best identified by a combination of dyssynergic patterns during attempted defecation and other findings (see diagnostic criteria).

**Epidemiology.** The prevalence of FDDs in the general population is unknown because the diagnosis requires laboratory testing.
At tertiary referral centers, the prevalence of dyssynergic defecation among patients with chronic constipation has ranged widely, from 20% to 81%. However, the prevalence of dyssynergia may have been overestimated due to the high false-positive rates seen in some studies. In one tertiary care center, the prevalence of dyssynergia was 3 times higher in women than men, but was similar in younger and older individuals.

### F3. Diagnostic Criteria for Functional Defecation Disorders

1. The patient must satisfy diagnostic criteria for functional constipation and/or irritable bowel syndrome with constipation.
2. During repeated attempts to defecate, there must be features of impaired evacuation, as demonstrated by 2 of the following 3 tests:
   a. Abnormal balloon expulsion test
   b. Abnormal anorectal evacuation pattern with manometry or anal surface EMG
   c. Impaired rectal evacuation by imaging

Subcategories F3a and F3b apply to patients who satisfy criteria for FDD.

#### F3a. Diagnostic Criteria for Inadequate Defecatory Propulsion

Inadequate propulsive forces as measured with manometry with or without inappropriate contraction of the anal sphincter and/or pelvic floor muscles.

#### F3b. Diagnostic Criteria for Dyssynergic Defecation

Inappropriate contraction of the pelvic floor as measured with anal surface EMG or manometry with adequate propulsive forces during attempted defecation.

Criteria fulfilled for the last 3 months with symptom onset at least 6 months before diagnosis.

These criteria are defined by age- and sex-appropriate normal values for the technique.

### Clinical Evaluation

A detailed assessment of bowel symptoms (e.g., prolonged or excessive straining, feeling of incomplete evacuation after defecation, digital facilitation of defecation) and a meticulous DRE often raise suspicion for an FDD. Bowel diaries avoid the limitation of recall bias inherent to questionnaires and an interview. In a single study, the DRE has a sensitivity of 75% and specificity of 87% for detecting dysynergia, which is associated with contraction or failure to relax the puborectalis and/or anal sphincter muscle and reduced perineal descent when patients try to expel the examining finger.

Physiologic studies should be considered if there is insufficient response to conservative treatment, for example, education regarding normal bowel habits, increased dietary fiber and liquids, and elimination of medications with constipating side effects whenever possible. These studies should include balloon expulsion test, anorectal manometry, and if necessary, defecography-imaging to aid in diagnosis of FDD. There is no single “gold standard” diagnostic test to diagnose FDD and limited agreement among various tests.

**Balloon expulsion test.** Rectal expulsion can be evaluated by asking patients to expel balloons filled with water or air from the rectum. The time required to expel the balloon depends on the method used and ranges from 1 minute to expel a 50-mL balloon filled with water to 2 minutes. It is recommended that the patient sit on a commode chair behind a privacy screen. The balloon expulsion test is a useful screening test for FDD, but it does not define the mechanism of disordered defecation. Because the balloon may not mimic the patient’s stool, a normal balloon expulsion study does not always exclude a defecation disorder.

**Manometric assessment.** Traditionally, ARM has been considered essential for diagnosis of FDDs. This assessment includes measurement of intrarectal pressures during attempted defecation, and measurement of anal pressures and/or EMG activity during attempted defecation. However, given the overlap of findings in asymptomatic people and patients with FDD, the precise criteria and utility of manometry for diagnosing defecatory disorders is in evolution. Also, body position and manometry systems may influence findings.

A normal pattern is characterized by increased intrarectal pressure associated with anal relaxation. A study of 100 patients with a 6-sensor, solid-state manometry system identified 4 patterns of FDD. Two patterns, types I and III, describe dyssynergic defecation. Type I pattern is characterized by increased intrarectal pressure (≥45 mm Hg) and increased anal pressure reflecting contraction of the anal sphincter. Type III pattern is characterized by increased intrarectal pressure (≥45 mm Hg) with absent or insufficient (<20%) relaxation of the anal sphincter. Inadequate propulsion (intrarectal muscles” are no longer specified because they vary among different techniques.
pressure <45 mm Hg) may be associated with paradoxical contraction (type II pattern) or insufficient relaxation (<20%) of anal sphincter (type IV pattern). During testing 1 month later, the abnormal patterns were reproducible in 51 of 53 patients. Levels of inter-observer agreement for identifying these patterns was substantial for types I and IV dyssynergia, moderate for normal defecation pattern, and fair for types II and III dyssynergia. A study using high-resolution manometry in 62 healthy women and 295 women with chronic constipation identified 3 phenotypes (high anal, low rectal, and hybrid) that discriminated among patients with normal and abnormal balloon expulsion time with 75% sensitivity and 75% specificity.

**Defecography.** Defecography is a radiologic technique used to evaluate the rectum and pelvic floor during attempted defecation. This test can detect structural abnormalities (rectocele, enterocele, intussusception, rectal prolapse, and megarectum) and assess functional parameters (anorectal angle at rest and during straining, perineal descent, anal diameter, indentation of the puborectalis, degree of rectal emptying).

The diagnostic value of defecography is unclear, but is still employed when ARM and balloon expulsion test are equivocal, or for patients who are unable to evacuate a balloon, but who relax the pelvic floor normally during simulated defecation. In several European countries, defecography is the primary modality for identifying FDD.

Magnetic resonance defecography images anorectal motion and rectal evacuation in real time. Advantages include better resolution of soft tissue surrounding the rectum, improved ability to visualize anal sphincter and levator ani muscles, and lack of radiation. MRI is particularly useful in patients with normal balloon expulsion to identify structural lesions and disordered defecation, and to guide surgical therapy, for example for rectoceles and cystoceles.

**Radio-opaque marker test of whole gut transit time.** By itself, slow colonic transit is not diagnostic of a primary colonic motility disorder because slow transit constipation exists independent of, or co-exists with, FDDs and up to two-thirds of patients with a defecation disorder also have delayed colonic transit. In one study, colonic transit improved after biofeedback therapy for outlet dysfunction, which suggests that outlet dysfunction was responsible for delayed colonic transit. Colonic transit time can be measured by obtaining abdominal radiographs after patients ingest radio-opaque markers, a wireless motility capsule, or by scintigraphy. The wireless motility capsule and scintigraphy can also measure gastric emptying and small intestinal transit, which may also be delayed in constipated patients.

**Utility of anorectal testing for functional defecation disorders.** The role of diagnostic testing was evaluated by assessing anorectal manometry, balloon expulsion test, defecography, and colonic transit in 100 consecutive patients with symptoms of difficult defecation. In this group, anal manometry and balloon expulsion were normal in 30%. Among 70 patients with abnormal manometry, balloon expulsion was abnormal in 42 patients (60%) indicative of FDD. Among 28 patients with abnormal manometry and normal balloon expulsion, defecography showed features of dyssynergic defecation in 7 patients (25%). Because a considerable proportion of healthy people exhibit dyssynergia when tested with high-resolution manometry, the utility of high-resolution manometry for identifying DD is unclear. Based on these results, abnormal findings in 2 of 3 tests (ie, anorectal manometry, balloon expulsion test, and defecography) are required to confirm the diagnosis of FDD.

**Pathophysiology**

FDDs are probably acquired but subliminal behavioral disorders, particularly in patients who learn to relax the external anal sphincter and puborectalis muscles appropriately when provided with biofeedback training.

Anxiety and/or psychological stress may also contribute to the development of dyssynergic defecation by increasing skeletal muscle tension and one study found that patients with dyssynergic defecation had higher scores for anxiety, depression, paranoid ideation, hostility, and obsessive compulsiveness than those patients with slow transit constipation. Psychological distress seem to have a negative impact on the outcome of biofeedback therapy. Uncontrolled studies have reported sexual abuse in 22% of women with FDD, and 40% of women with functional lower gut disorders, including FDD.

**Treatment**

Historically, 2 types of pelvic floor training involving behavioral modification have been advocated: biofeedback training in which pressure sensors or EMG placed inside the anus and rectum provide feedback to the patient on muscle activity and simulated defecation in which the patient practices evacuating an artificial stool surrogate. Simulated defecation has been combined with diaphragmatic muscle training by some investigators. Recent randomized controlled trials have used multicomponent biofeedback treatment, which includes the following four steps (Table 3):

1. Patient education: Explain to patients that they inadvertently squeeze or fail to relax their anus when they are straining.
2. Enhance push effort: Teach the patients to effectively push, when straining, by appropriately increasing the intra-abdominal pressure; use feedback from rectal sensor regarding abdominal and diaphragmatic push effort to expel stool.
3. Train to relax pelvic floor muscles: Teach patients to relax their pelvic floor muscles when straining. This skill can be taught by providing visual
feedback regarding anal canal pressure or EMG activity (Figure 2).

4. Practice simulated defecation: Educate patient to practice defecation and expulsion of a lubricated, inflated balloon while the therapist assists by gently pulling on the catheter.

Several randomized controlled trials have demonstrated that biofeedback is safe and effective treatment for dyssynergic defecation (Table 3). Biofeedback therapy was more effective than sham feedback, pelvic floor exercises, laxatives, and muscle relaxant drugs, both on a short- and long-term basis without side effects. Biofeedback therapy is to be regarded as first-choice treatment for FDD whenever dedicated expertise is available. Biofeedback therapy is not effective for constipated patients without FDD. Whether biofeedback is as effective for altered defecatory propulsion as it is for dyssynergic defecation is not known.

### Table 3. Summary of Randomized Controlled Trials of Biofeedback Therapy for functional defecation disorder

<table>
<thead>
<tr>
<th>Variable</th>
<th>Chiarioni et al(^{164})</th>
<th>Rao et al(^{183})</th>
<th>Chiarioni et al(^{168})</th>
<th>Heymen et al(^{186})</th>
<th>Rao et al(^{185})</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trial design</td>
<td>EMG Biofeedback vs PEG 14.6 g</td>
<td>Biofeedback (manometry pressure) vs standard treatment vs sham biofeedback</td>
<td>EMG biofeedback for slow transit vs dyssynergia</td>
<td>EMG biofeedback vs diazepam 5 mg vs placebo</td>
<td>Biofeedback (manometry pressure) vs standard therapy</td>
</tr>
<tr>
<td>Subjects and randomization and intervention(s)</td>
<td>109 (104 women)</td>
<td>77 (69 women)</td>
<td>52 (49 women)</td>
<td>84 (71 women)</td>
<td>52 = short-term therapy</td>
</tr>
<tr>
<td></td>
<td>54 biofeedback</td>
<td>1:1:1 distribution</td>
<td>34 dyssynergia</td>
<td>30 biofeedback</td>
<td>26 = long-term study</td>
</tr>
<tr>
<td></td>
<td>55 polyethylene glycol</td>
<td>Standard: diet, exercise, laxatives Sh: progressive muscle relaxation with anorectal probe</td>
<td>12 slow transit</td>
<td>30 diazepam</td>
<td>12 = biofeedback</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>6 mixed</td>
<td>24 placebo</td>
<td>13 = standard therapy</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Standard: diet, exercise, laxatives (titrated)</td>
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<tr>
<td>Duration and no. of biofeedback sessions</td>
<td>6 mo, 1 y, 5 weekly, 30-min training sessions performed by physician investigator</td>
<td>3 mo, every other week, 1 h, maximum of 6 sessions over 3 mo, performed by biofeedback nurse therapist</td>
<td>1-6-12-24 mo</td>
<td>6 every other week, 1-h sessions</td>
<td>1 y</td>
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<td></td>
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<td></td>
<td></td>
<td>6 active therapy sessions and 3 reinforcement sessions at 3-mo intervals</td>
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<tr>
<td>Primary outcomes</td>
<td>Global improvement of symptoms Worse = 0</td>
<td>Presence of dyssynergia Balloon expulsion time</td>
<td>Symptom improvement None = 1 Mild = 2</td>
<td>Global symptom relief No. of CSBMs Secondary outcome; Presence of dyssynergia Balloon expulsion time</td>
<td>Global satisfaction</td>
</tr>
<tr>
<td></td>
<td>No improvement = 1</td>
<td>No. of CSBMs</td>
<td>Fair = 3 Major = 4</td>
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<tr>
<td>Dyssynergia corrected or symptoms improved</td>
<td>79.6% reported major improvement at 6 and 12 mo 81.5% reported persistent major improvement at 24 mo</td>
<td>Dyssynergia corrected at 3 months in 79% with biofeedback vs 4% sham and 6% in standard group: CSBM = biofeedback group vs sham or Standard; (P &lt; .05)</td>
<td>71% with dyssynergia and 8% with slow transit alone reported fair improvement in symptoms both at short and long-term follow-up intervals</td>
<td>70% improved with biofeedback compared to 38% with placebo and 30% with diazepam; (P &lt; .01)</td>
<td>No. of CSBMs/wk increased significantly in biofeedback; (P &lt; .001)</td>
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<td></td>
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<td>Dyssynergia pattern normalized; (P &lt; .001)</td>
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<td></td>
<td></td>
<td>Balloon expulsion improved; (P &lt; .001)</td>
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<td>Colonic transit normalized; (P &lt; .01)</td>
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</table>

CSBM, complete spontaneous bowel movement; PEG, polyethylene glycol.
Recommendations for Future Research

1. Multicenter studies of normal physiology of defecation and fecal continence using newer diagnostic modalities in large groups of subjects stratified for age, sex, and parity.

2. Define the role of rectal contraction and sensation in disordered defecation, especially to understand dyssynergic defecation vs inadequate propulsion.

3. Evaluate interaction(s) between stool consistency, sphincter weakness, sphincter defects, rectal sensation and compliance, and neurogenic sphincter injury in FI.

4. Randomized, blinded controlled study of biofeedback treatment for dyssynergic defecation, especially to examine its generalizability for FDD.

5. Examine the natural history, duration, and phenotype of anorectal pain syndromes and perform randomized studies of drugs, biofeedback, and other treatments for levator ani syndrome.

6. Randomized controlled trials of bowel management, biofeedback, sacral nerve stimulation, anal bulking agents and sphincteroplasty in FI, including mechanistic understanding and well-designed outcome measures.

Supplementary Material

Note: The first 50 references associated with this article are available below in print. The remaining references accompanying this article are available online only with the electronic version of the article. Visit the online version of Gastroenterology at www.gastrojournal.org, and at http://dx.doi.org/10.1053/j.gastro.2016.02.009.

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Reprint requests
Address requests for reprints to: Satish SC Rao, MD, PhD, FRCP (LON), Augusta University, Division of Gastroenterology and Hepatology, AD 2238, Digestive Health Center 1481 Laney-Walker Blvd, Augusta, Georgia 30912. e-mail: srao@gru.edu; fax: (706) 721-0331; office: 706-721-2238.

Conflicts of interest
The authors disclose the following: AEB is an inventor of the portable anorectal manometry catheter that has been licensed to Medispira Inc; AEB and Mayo Clinic have contractual rights to receive royalties from the licensing of this technology. GC is an advisory board member and speaker for Shire Italia and Takeda Italia. The remaining authors disclose no conflicts.

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