Functional disorders of the anus and rectum

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Abstract

In this report the functional anorectal disorders, the etiology of which is currently unknown or related to the abnormal functioning of normally innervated and structurally intact muscles, are discussed. These disorders include functional fecal incontinence, functional anorectal pain, including levator ani syndrome and proctalgia fugax, and pelvic floor dyssynergia. The epidemiology of each disorder is defined and discussed, their pathophysiology is summarized and diagnostic approaches and treatment are suggested. Some suggestions for the direction of future research on these disorders are also given.

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A functional gastrointestinal disorder is defined as "a variable combination of chronic or recurrent gastrointestinal symptoms not explained by structural or biochemical abnormalities."[1] In keeping with this definition, this report addresses anorectal symptoms, the etiology of which is currently unknown or is related to the abnormal functioning of normally innervated and structurally intact muscles, or is attributed to psychological causes.

The functional anorectal disorders are defined primarily on the basis of the symptoms (table 1). Retrospective reports are unreliable but can be improved by interviewing the patient and by prospective symptom diaries.

This review and the associated recommendations are based on an authoritative review of the world literature by experts. (See acknowledgments for a list of expert reviewers whose advice was sought by the authors.) The diagnostic criteria include minimum duration of symptoms, which were selected arbitrarily so as to exclude self-limiting conditions while avoiding unnecessary delays in evaluation.

F1. Functional fecal incontinence

Functional fecal incontinence is defined as recurrent uncontrolled passage of fecal material in a person who has no evidence of neurologic or structural etiologies. This is distinct from fecal incontinence due to neurological injury, seepage from prolapsed rectal mucosa, poor hygiene, and willful soiling. However, neurogenic and anatomic causes of fecal incontinence may coexist with functional causes of incontinence.

Fecal incontinence should not be considered a medical problem earlier than age four years, and depending on the cultural context, the age at which it is considered appropriate to initiate treatment may be later.

EPIDEMIOLOGY

The prevalence of fecal incontinence (including organic causes) in US and European adults ranges from 2 to 7% and is about 0.7% for incontinence of solid stool.[2 3] Major risk factors for functional fecal incontinence include retention of stool in the rectum, the irritable bowel syndrome, and diarrhea. Among the elderly, cognitive and mobility impairment, certain medications, as well as diarrhea are significant risk factors.

Functional fecal incontinence associated with fecal impaction occurs in about 1.4% of children aged seven years. The incidence of fecal incontinence from all causes in US and British nursing home populations is estimated to be 30%.[4]

DIAGNOSTIC CRITERIA

Recurrent uncontrolled passage of fecal material for at least one month, in an individual with a developmental age of at least four years, associated with:
(1) Fecal impaction; or
(2) Diarrhea; or
(3) Non-structural anal sphincter dysfunction.

It is recognized that functional causes of fecal incontinence such as constipation and diarrhea may overlap with structural abnormalities (e.g., sphincter muscle injury, nerve injury).

RATIONALE FOR CHANGES IN DIAGNOSTIC CRITERIA

- The lowest age for inferring that the uncontrolled passage of stool constitutes fecal incontinence has been increased from three to four years to bring these diagnostic criteria into line with the *Diagnostic and Statistical Manual of the American Psychiatric Association*.
- The criteria for functional fecal incontinence have been broadened to include incontinence associated with diarrhea as well as constipation. This acknowledges that about 25% of patients with diarrhea-predominant irritable bowel syndrome experience incontinence.
- Previously, an elevated threshold for perception of rectal distension was included as a diagnostic criterion. Because elevated sensory thresholds are associated with organic causes of fecal incontinence including spinal cord injury, stroke, and diabetic peripheral neuropathy, this finding was felt to be too

Abbreviations used in this paper: EMG, electromyography; CT, computed tomography.

Table 1 Functional gastrointestinal disorders

A. Esophageal disorders
A1. Globus
A2. Rumination syndrome
A3. Functional chest pain of presumed esophageal origin
A4. Functional heartburn
A5. Functional dysphagia
A6. Unspecified functional esophageal disorder
B. Gastroduodenal disorders
B1. Functional dyspepsia
B1a. Ulcer-like dyspepsia
B1b. Dysmotility-like dyspepsia
B1c. Unspecified (non-specific) dyspepsia
B2. Aerophagia
B3. Functional vomiting
C. Bowel disorders
C1. Irritable bowel syndrome
C2. Functional abdominal bloating
C3. Functional constipation
C4. Functional diarrhea
C5. Unspecified functional bowel disorder
D. Functional abdominal pain
D1. Functional abdominal pain syndrome
D2. Unspecified functional abdominal pain
E. Biliary disorders
E1. Gall bladder dysfunction
E2. Sphincter of Oddi dysfunction
F. Anorectal disorders
F1. Functional fecal incontinence
F2. Functional anorectal pain
F2a. Levator ani syndrome
F2b. Proctalgia fugax
F3. Pelvic floor dyssynergia
G. Functional pediatric disorders
G1. Vomiting
G1a. Infant regurgitation
G1b. Infant rumination syndrome
G1c. Cyclic vomiting syndrome
G2. Abdominal pain
G2a. Functional dyspepsia
G2b. Irritable bowel syndrome
G2c. Functional abdominal pain
G2d. Abdominal migraine
G2e. Aerophagia
G3. Functional diarrhea
G4. Disorders of defecation
G4a. Infant dyschezia
G4b. Functional constipation
G4c. Functional fecal retention
G4d. Non-retentive fecal soiling

non-specific to serve as a diagnostic criterion for functional fecal incontinence.

CLINICAL EVALUATION

The diagnosis of functional fecal incontinence due to constipation or diarrhea can often be made by history and physical examination. Three alternative causes of soiling must be excluded: (1) rectal prolapse with mucus secreted onto the underclothes, (2) mental incompetence, and (3) willful soiling.

Physical examination

The initial examination should be done without prior enema or laxatives. A characteristic finding in constipation-related fecal incontinence is a large mass of stool in the rectum on digital examination and/or in the colon on abdominal palpation. If the patient is able to contract the external anal sphincter, efferent denervation is unlikely. Rectal prolapse can be evaluated by asking the patient to strain as if defecating while seated on a commode chair. The digital examination should assess for pelvic floor dyssynergia (decreased anal canal pressures reliably exclude the diagnosis of pelvic floor dyssynergia, but abnormal findings require confirmation). If the history and physical examination do not support a diagnosis of functional fecal incontinence, further examination may be required.

Diagnostic tests used to identify anatomical and neurological causes of fecal incontinence

Anoscopy is recommended over flexible sigmoidoscopy to examine the anal canal to determine whether fissures, inflammation, or mechanical obstruction are contributing to fecal soiling.

Manometry assesses continence mechanisms by determining: (1) the threshold volume of rectal distension required to produce the first sensation of distension and a sustained feeling of urgency to defecate; (2) rectal compliance as determined by the pressure:volume ratio during stepwise distension and maximum tolerable volume; (3) amplitude and duration of voluntary contractions of the external anal sphincter; and (4) resting pressure in the anal canal. This can be done using perfused catheters, solid-state pressure transducers, electromyography (EMG) electrodes in the anal canal, or balloons positioned in the anal canal.[5] The use of EMG alone is not recommended.

Anal endosonography allows imaging of both anal sphincters to identify structural defects in either muscle. The procedure is rapid and less invasive than pelvic computed tomography (CT), magnetic resonance imaging, or EMG. However, interpretation is very operator dependent and requires some experience.

Neurophysiological studies to evaluate the integrity of the pudendal nerve in patients with fecal incontinence include pudendal nerve terminal motor latencies and concentric needle EMG recordings from the external anal sphincter or puborectalis muscle. These studies are usually not indicated. However, surface EMG recorded in the anal canal or from perianal electrodes is useful as a biofeedback signal for pelvic floor retraining in patients with fecal incontinence or pelvic floor dyssynergia.

PATHOPHYSIOLOGY AND PSYCHOLOGICAL FACTORS

Impaired perception of rectal distension has been well documented in both children and adults with functional fecal incontinence. Sensory changes may be a consequence of fecal impaction, which alters the tone and viscoelastic properties of the bowel wall, or may alter mechanoreceptors. Decreased anorectal sensitivity may contribute to incontinence by causing the threshold for reflex inhibition of the internal anal sphincter to occur before the patient perceives the presence of stool in the rectum.[6]

Two types of internal anal sphincter dysfunction have been described in patients with idiopathic incontinence: (*a*) decreased resting pressure in the internal anal sphincter and (*b*) increased frequency of spontaneous internal anal sphincter relaxation (sampling reflex).[7]

Adult patients with fecal incontinence show elevated levels of psychological distress as well as elevations on scales measuring physical functioning, mental health, and social functioning. It is believed that these reflect primarily the consequences of having fecal incontinence.

TREATMENT

For patients with constipation-related fecal incontinence, habit training after bowel cleansing results in complete continence for roughly 60% of children and substantial reduction in frequency of soiling for another 23%.[8] For elderly incontinent patients, a daily osmotic laxative (e.g., lactulose 10 ml twice daily) plus a weekly enema was reported to be effective in more than 90% of patients.[9] For patients with diarrhea-related incontinence, loperamide is recommended. Biofeedback should be considered only in patients who do not respond to habit training or medication.

F2. Functional anorectal pain

Two forms of functional anorectal pain have been described: levator ani syndrome and proctalgia fugax. These two types of pain frequently coexist, but they can be distinguished on the basis of duration, frequency and characteristic quality of pain. It is necessary to exclude other causes of anorectal pain such as ischemia, fissures, inflammatory bowel disease, and intramuscular abscess.

F2a. Levator ani syndrome

The levator ani syndrome is also called levator spasm, puborectalis syndrome, chronic proctalgia, pyriformis syndrome, and pelvic tension myalgia. The pain is often described as a vague, dull ache or pressure sensation high in the rectum, often worse with sitting or lying down, which lasts for hours to days.

The prevalence of symptoms compatible with levator ani syndrome in the general population is 6.6%, and it is more common in women.[3] Only 29% consult a physician, but associated disability appears to be significant. More than half of affected patients are aged 30–60 years, and prevalence tends to decline after age 45.

DIAGNOSTIC CRITERIA

At least 12 weeks, which need not be consecutive, in the preceding 12 months of:

(1) Chronic or recurrent rectal pain or aching;
(2) Episodes last 20 minutes or longer; and
(3) Other causes of rectal pain such as ischemia, inflammatory bowel disease, cryptitis, intramuscular abscess, fissure, hemorrhoids, prostatitis, and solitary rectal ulcer have been excluded.

CLINICAL EVALUATION

The diagnosis of levator ani syndrome is made on the basis of symptoms alone, However, confidence in the diagnosis is substantially increased if posterior traction on the puborectalis reveals tight levator ani muscles and tenderness or pain. Tenderness may be predominantly left-sided, and massage of this muscle will generally elicit the characteristic discomfort. Two levels of diagnostic classification are proposed: a "highly likely" diagnosis of levator ani syndrome if symptom criteria are satisfied and these physical signs are present, or a "possible" diagnosis if the symptom criteria are met but the physical signs are absent. Clinical evaluation will usually include sigmoidoscopy and appropriate imaging studies such as defecography, ultrasound, or pelvic CT to exclude alternative diseases.

PATHOPHYSIOLOGY

Levator ani syndrome has been hypothesized to result from spastic or overly contracted pelvic floor muscles.[10] However, the etiology is unknown. Some reports suggest that levator ani syndrome is associated with psychological stress, tension, and anxiety.[11]

TREATMENT

A variety of treatments directed at reducing tension in the levator ani muscles have been described: digital massage of the levator ani muscles; Sitz baths; muscle relaxants such as methocarbamol, diazepam, and cyclobenzeprine; electrogalvanic stimulation; and biofeedback training. None of the treatment studies included a control group, and patient selection criteria varied. Many patients fail to respond to treatment. Surgery should be avoided.

F2b. Proctalgia fugax

Proctalgia fugax is defined as sudden, severe pain in the anal area lasting several seconds or minutes, and then disappearing completely.[12] Attacks are infrequent, occurring less than five times a year in 51% of patients.

Community prevalence estimates range from 8 to 18%. Only 17–20% of those affected report the symptoms to their physicians. Prevalence rates in men and women vary.

DIAGNOSTIC CRITERIA

(1) Recurrent episodes of pain localized to the anus or lower rectum; and
(2) Episodes last from seconds to minutes; and
(3) There is no anorectal pain between episodes.

CLINICAL EVALUATION

Diagnosis is based on symptoms alone. There are no physical examination findings or laboratory tests that support the diagnosis.

PATHOPHYSIOLOGY AND PSYCHOLOGICAL FACTORS

The short duration and sporadic, infrequent nature of this disorder has made the identification of physiological mechanisms difficult. Several studies suggest that smooth muscle spasm may be the cause of proctalgia fugax.[13 14] Psychological testing suggests that many patients are perfectionistic, anxious, and/or hypochondriacal.

TREATMENT

For most patients, episodes of pain are so brief that treatment consists only of reassurance and explanation. However, a small group of patients have proctalgia fugax on a frequent basis; a recent study shows that inhalation of salbuta-

mol (a beta adrenergic agonist) shortens the duration of episodes of proctalgia.[15] Others have recommended clonidine or amylnitrate.

F3. Pelvic floor dyssynergia

Pelvic floor dyssynergia is characterized by paradoxical contraction or failure to relax the pelvic floor during attempts to defecate.[16 17] It is frequently associated with symptoms of difficult defecation including straining, feeling of incomplete evacuation after defecation, and digital facilitation of defecation.

The prevalence of pelvic floor dyssynergia in the population is unknown, because the diagnosis requires physiological testing. However, in patients referred for evaluation of chronic constipation, pelvic floor dyssynergia is found in 25–50% of both children and adults.[18] This may be an overestimation due to the high false-positive rates seen in some studies.[19] No information is available on gender differences.

DIAGNOSTIC CRITERIA

(1) The patient must satisfy diagnostic criteria for functional constipation in Diagnostic Criteria C3;
(2) There must be manometric, EMG, or radiologic evidence for inappropriate contraction or failure to relax the pelvic floor muscles during repeated attempts to defecate;
(3) There must be evidence of adequate propulsive forces during attempts to defecate; and
(4) There must be evidence of incomplete evacuation.

Diagnostic criteria for functional constipation are: at least 12 weeks (which need not be consecutive) in the preceding 12 months of two or more of: (1) straining in >1/4 defecations; (2) lumpy or hard stools in >1/4 defecations; (3) sensation of incomplete evacuation in >1/4 defecations; (4) sensation of anorectal obstruction/blockage in >1/4 defecations; (5) manual maneuvers to facilitate >1/4 defecations (e.g., digital evacuation, support of the pelvic floor); and/or (6) <3 defecations/week. Loose stools are not present, and there is insufficient evidence for irritable bowel syndrome.

RATIONALE FOR CHANGES IN DIAGNOSTIC CRITERIA

In the previous working team report, symptoms of difficult defecation were used to define an independent diagnostic entity, which was called dyschezia. However, differentiation of subtypes of constipation based on symptoms alone is not reliable. Consequently, in the revised working team reports, a diagnosis of pelvic floor dyssynergia depends primarily on physiological findings.

The previous working team report recommended diagnosing pelvic floor dyssynergia on the basis of symptoms of difficult defecation plus manometric, EMG, or radiologic evidence of failure to relax the pelvic floor when attempting to defecate. However, one study[19] suggests that these criteria are too non-specific, and more restrictive diagnostic criteria have been recommended.[20] The working team therefore recommends augmenting the diagnostic criteria by requiring evidence of adequate propulsive forces and evidence of incomplete evacuation in addition to evidence of paradoxical contraction. However, there is insufficient empirical evidence to justify recommending specific tests or specific cut-off points on those tests.

In the previous working team report, a diagnostic category existed for difficult defecation which was associated with manometric evidence of internal anal sphincter dysfunction. The working team recommends dropping this category until it is confirmed by further studies.

CLINICAL EVALUATION

The physiological investigations considered useful for making a diagnosis of pelvic floor dyssynergia are: (1) anorectal manometry, (2) electromyography of the external anal sphincter, (3) balloon defecation (simulated defecation), and (4) defecography. Finding, on physical examination, that the patient is able to decrease anal canal pressure when straining is useful for ruling out pelvic floor dyssynergia, but an increase in anal canal pressure when straining during physical examination is not a reliable indication of the presence of pelvic floor dyssynergia.

The measurement of anal canal pressure and EMG activity from the external anal sphincter during straining to defecate is especially helpful in identifying patients with pelvic floor dyssynergia. There should be evidence of adequate propulsive forces during straining, measured as increased intra-rectal pressures and/or abdominal wall contraction.

Efforts to measure defecation include introducing lubricated balloons attached to thin catheters into the rectum, filling them with 50 ml water or air, and asking the patient to defecate them. Latencies less than 60 seconds are considered normal. However, additional research is needed to standardize this test. Many investigators use the balloon defecation test as a screening tool which, if positive, leads to further testing.

Defecography is a radiological technique to evaluate the rectum and pelvic floor during an attempted defecation. This test provides information on the presence of structural abnormalities and functional parameters such as the anorectal angle at rest and during straining, diameter of the anal canal, indentation of the puborectalis, and degree of rectal emptying. However, its value has been questioned because agreement between investigations on the interpretation of findings is low.[5] Defecography is principally useful for identifying structural causes of obstructed defecation and for quantifying rectal emptying.

The most frequently used technique for measuring colonic transit time involves having the patient swallow radio-opaque rings and taking abdominal radiographs on one or more days thereafter.[21 22] Although of no value in diagnosing pelvic floor dyssynergia, this test is

useful for determining whether the patient has colonic inertia as an alternative or comorbid cause of constipation.

PATHOPHYSIOLOGY

Pelvic floor dyssynergia is not attributable to a neurological lesion as at least two-thirds of patients can learn to relax the external anal sphincter and puborectalis muscles appropriately when provided with biofeedback training.[23] Anxiety and/or psychological stress may contribute to the development of pelvic floor dyssynergia by increasing skeletal muscle tension. Adults with difficult defecation have exhibited significantly higher scores for anxiety, depression, interpersonal sensitivity, obsessive compulsive traits, phobic anxiety, and somatization. Pelvic floor dyssynergia is more common in women with a history of sexual abuse.[24]

PATHOPHYSIOLOGY

Two types of training have been described for pelvic floor dyssynergia: (1) biofeedback training in which sensors in the anal canal or adjacent to the anus, monitor and provide feedback to the patient on striated muscle activity or anal canal pressures[23]; and (2) simulated defecation in which the patient practices defecation of a simulated stool.[25] Both seem to be effective. A systematic review published in 1993 gives an overall success rate of 67%.[26] However, recent but uncontrolled studies of large series of patients report improvement in 48[27] to 62%[28] of patients (intent-to-treat analyses).

Directions for future research

- Multicenter studies of the normal physiology of defecation in large groups of subjects stratified by age, gender, and (in women) by parity. This would (*a*) help to define the normal ranges for diagnostic tests of fecal incontinence and pelvic floor dyssynergia, and (*b*) help establish standardized technology for assessment of these conditions.
- Diagnostic criteria for pelvic floor dyssynergia must be validated and should include definite changes in anal canal pressure during straining, propulsive forces, and per cent evacuation of the rectum.
- A randomized, blinded study of the efficacy of biofeedback treatment for pelvic floor dyssynergia should be carried out. The study design should control for the placebo response.
- Psychological characteristics of patients with levator ani syndrome and proctalgia fugux may help to define the etiology of these disorders. These studies should compare medical clinic patients to individuals in the community.
- Studies are needed to determine whether proctalgia fugax and levator ani syndrome are separate disorders. These should include detailed symptom reports supplemented by symptom diaries in large groups of patients.
- A randomized, blinded multicenter study to compare electrogalvanic stimulation, biofeedback, and muscle relaxant drugs for the treatment of levator ani syndrome should be performed.

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