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Disorders of ano-rectal motility: functional defecation disorders and fecal incontinence

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ABSTRACT

Functional defecation disorders (FDDs) and fecal incontinence (FI) are common anorectal disorders often distressing and significantly add to the health care burden. They present with multiple, overlapping symptoms that can often obscure the underlying pathophysiology and can pose significant management dilemmas. A detailed history, stool diaries and visual scales of stool form, a careful digital rectal examination are needed to guide anorectal physiology tests. With high-resolution (3-D) anorectal manometry, anal ultrasonography, (magnetic resonance) defecography and imaging, and neurophysiological tests, it is possible to define and characterize the underlying structural and functional abnormalities more accurately. In this review, we present a succinct update on the latest knowledge with regards to the pathophysiology, diagnosis and management of FDDs, fecal incontinence and abnormalities of rectal capacity (i.e; megarectum, microrectum).

Key words: anorectal disorders – defecation disorders - constipation – anismus- recto-anal dyssynergia- fecal incontinence – megarectum – microrectum

INTRODUCTION

Functional defecation disorders (FDDs) and fecal incontinence (FI) are among the most prevalent gastrointestinal conditions. FDDs can affect up to 50% of patients with chronic constipation and FI varies from 2 to 21% of the general population, with greatly worsened quality of life and increased healthcare burden for both conditions[1]. Thus, the annual direct and indirect costs per person related to FI are estimated to be up to \$3521 in Europe and \$4410 in the USA[2]. This review focuses on the definition, epidemiology, diagnostic criteria, explorations and management of these disorders according to Rome IV classification, followed by a focus on rectal capacity disorders. It will provide a valuable resource for clinicians to approach and manage functional anorectal disorders in a stepwise and logical manner. Areas for future research are also additionally highlighted.

I. FUNCTIONAL DEFECATION DISORDERS.

1. Definitions:

Functional defecation disorder (FDD) is characterized by the presence of constipation symptoms with paradoxical contraction or inadequate relaxation of the pelvic floor muscles (also named dyssynergic defecation or anismus) and/or inadequate propulsive forces during attempted defecation[1]. These disorders are frequently associated with symptoms such as excessive straining, feeling of incomplete evacuation, and digital facilitation of bowel movements[1]. However, symptoms alone do not consistently distinguish patients with FDDs from those without[1]. Thus, the Rome IV criteria have recently defined FDDs based on both symptoms and physiological testing.

To be diagnosed with FDD according to Rome IV criteria, patients must satisfy clinical criteria for Functional Constipation (FC) and/or Irritable Bowel Syndrome with Constipation (IBS-C; box 1), but also have to demonstrate impaired rectal evacuation in at least 2 of the following 3 tests: (i) abnormal balloon expulsion test, (ii) abnormal anorectal evacuation

pattern with anorectal manometry or anal surface electromyography (EMG), (iii) impaired rectal evacuation on defecography without structural lesions.

2. Epidemiology and pathophysiology:

Chronic constipation represents the most prevalent gastrointestinal disorder in general population, with a prevalence of 11-18% among studies[3]. However, the prevalence of FDDs in community remains still unknown, mainly because of the necessity of laboratory testing in order to establish positive diagnosis[1],[4]. Up to 50% of patients with chronic constipation can be diagnosed with dyssynergia with anorectal manometry in tertiary centers[5], but the rate of false positive remains high[1]. The prevalence of dyssynergia was 3 times higher in women than in men in one study from a tertiary care center, but the prevalence was similar between old and young patients in the same study[5].

Potential contributors to the development of FDDs seem to be anxiety and/or psychological stress, resulting in an increase in skeletal muscle tension[5]. Indeed, the prevalence of anxiety, depression, obsessive compulsiveness, psychoticism and paranoid ideation are higher in patients with dyssynergic defecation. Lastly, sexual abuse are reported more frequently in patients with dyssynergic defecation, representing up to 48% of patients[6].

3. Clinical and paraclinical evaluation

The diagnostic approach for FDDs is similar to the evaluation of any other gastrointestinal (GI) disease. Firstly, the clinician has to search for alarm signs, such as acute change in bowel habits (in particular after the age of 50), hematochezia, iron deficiency, unexplained weight loss or familial history of colorectal cancer. In one of these cases, specific investigations are needed to exclude pathologic or metabolic disease. Clinicians should also have in mind that constipation can arise from neurological disorders like Parkinson's disease and medications,

such as opiates, calcium channel blockers and tricyclic antidepressants[1], [7]. In the absence of any organic or medication causes or anatomical pelvic floor disorders, functional constipation (FC, IBS-C, FDD) should be suspected[7].

In the evaluation of FDD, duration and nature of symptoms should be evaluated. Visual scales of stool form, especially the Bristol Stool Form Scale (BSFS) [8], and bowel diaries are very useful to describe patients stool form and accurately characterize bowel movements[7]. Indeed, the stool consistency is a better marker of colonic transit than stool frequency and can therefore help to diagnose slow transit constipation rather than FDD[4]. Additional symptoms like bloating, abdominal pain, distension and non-GI symptoms such as asthenia, psychological distress should also be noted in order to assess if the diagnosis criteria for IBS-C have been met. Lastly, history of sexual abuse or sexual traumatism can be informative regarding the etiology of the troubles but without clear distinction between IBS-C, FC or FDD[1] .

However, it is of importance to know that symptoms alone (excessive straining, feeling of incomplete evacuation and/or the use of digital maneuvers) cannot clearly differentiate between IBS-C, FC and FDD[5],[9]. Indeed, slow transit constipation can co-exist in up to 2/3 of patients with dyssynergia[5]. Moreover, coexistent difficult defecation was frequent in both patients with IBS-C (84%) and FC (68%) in one study[10].

Physical examination should search for alarm signs (abdominal mass, lymphadenopathy), musculoskeletal or neurogenic disease and a complete digital rectal examination (DRE) is mandatory[7],[11]. A first step of visual inspection, searching for anal fissures, rectal, pelvic organ or hemorrhoidal prolapse, is necessary. The DRE assesses the basal tone of the anal canal, the presence or absence of stool and search for masses, strictures and rectocele. Voluntary contraction of the anal sphincter and puborectalis muscle is then assessed while asking the patient to squeeze. Lastly, patient is asked to push like in an attempt to defecate, to

evaluate the recto-anal coordination. The clinician should put his left hand on the patient's abdomen in this stage in order to assess abdominal contraction. Abdominal muscle contraction, relaxation of anal sphincter and perineal descent is the normality of this maneuver. Absence of these normal findings, such as contraction or failure to relax puborectalis and/or anal sphincter muscles and/or absence of contraction of abdominal muscle, should lead to the suspicion of FDDs. Indeed, DRE has a specificity of 87% and a sensitivity of 75% to predict dyssynergia findings on anorectal manometry[11]. This step of physical examination is instructive for the clinician and should therefore not be neglected.

Physiologic studies should be performed after the failure of conservative therapy (exclusion of medication with constipation as side effect if possible, basic lifestyle and dietary modifications) and bulking agents or osmotic laxatives, suppositories and enemas, as FDDs may be less likely to respond to first-line treatment[7]. Physiologic studies include balloon expulsion test, anorectal manometry and in second line defecography-imaging if necessary[7], but to date, there is no single gold standard diagnostic test, as notified in the Rome IV criteria[1]. The purpose of these testing is to diagnose FDD and identify its mechanisms (i.e. inadequate propulsive forces with or without inappropriate contraction of the anal sphincter and/or pelvic floor muscles[1]) and thus to guide therapeutic measures.

Balloon expulsion test (BET):

This test assesses the capacity and time to evacuate from the rectum a 50 mL air- or warm water-filled balloon. Normal time to expel the balloon varies from 1 to 2 minutes, depending on the method used[12]. However, if the BET has been shown to be abnormal in a large proportion of patients with FDDs, this test may also be normal in patients with FDDs who are able to compensate by excessive straining and abnormal in asymptomatic subjects[7]. Moreover, the agreement of results of BET and anorectal manometry to diagnose FDD is low, explaining why this test is not used as a single diagnostic test and is now part of the London

classification for the interpretation of anorectal manometry (ARM) results as detailed below[13].

Anorectal manometry:

This testing includes assessment of intra-rectal pressure and measurement of anal pressures during attempted defecation. The recent London classification[13] provides interpretation for the disorders of anorectal coordination and requires the results of both the expulsion test (either BET or defecography) and the “push” maneuver on ARM (box 2). This classification uses two descriptive terms *propulsion* (i.e. adequacy increase in rectal pressure during “push”) and *dyssynergia* (i.e. finding of abnormal anal contraction or abnormal relaxation; Figure 1). If the BET result is abnormal with ARM showing either abnormal propulsion or dyssynergia or both, the diagnosis of FDDs is possible and should be confirmed according to the Rome IV criteria (2 out of 3 positive tests among the following tests: ARM, BET, defecography, EMG).

Defecography:

This radiological technique evaluates in a dynamic manner morphological changes of the rectum and pelvic floor during attempted defecation. In consequence, this procedure can detect anatomical abnormalities (rectocele, enterocele, intussusception, rectal prolapse and megarectum) and also investigate functional parameters like the anorectal angle and canal anal opening at rest and during straining. Defecography is especially useful when BET and ARM results are equivocal or contradictory[7], but this test is employed in several institutions as primary modality to identify FDDs[14]. Fluoroscopy defecography is traditionally used, but magnetic resonance defecography is a more recently available technique with advantages such as better resolution and lack of radiation but is performed in a non-physiologic position[1]. Magnetic resonance imaging machines for sitting imaging exist but are not yet currently used in this indication.

Colonic transit studies:

Colonic transit time can be measured using radio-opaque marker test, wireless motility capsule[15] or colonic scintigraphy[16]. The value of these tests in patients with FDDs is poor[7]. Indeed, slow transit constipation can exist in an independent way or co-exist with FDDs. Delayed colonic transit time, can be found in up to two third of patients with untreated FDDs among studies[1]. Moreover, colonic transit time can be improved after biofeedback in case of dyssynergic defecation, suggesting that this trouble can be secondary to the rectal evacuatory disorder[17].

Anal surface electromyography:

electrodes mounted on an acrylic anal plug or taped to the perianal skin can record average anal electromyographic activity. This technic is used to identify dyssynergic defecation, when an impaired reduction (<20%) of the signal is observed during attempted defecation[18].

4. Treatment:

Management of FDDs should be performed in a logical step-wise manner (Figure 2) and take in consideration patient's symptoms, underlying pathophysiology, age, co-morbidities, patient's concerns and expectations[7].

1. General conservative approach:

First of all, the clinician should listen to the patient's concerns, give a clear understanding of the underlying pathophysiology and establish realistic treatment goals. Patient's satisfaction and compliance to therapy are improved with tshis approach[4]. Initial therapy consists of the correction of coexisting issues such as exclusion of constipating medications if possible. The benefit of supplementing diet with fiber, in case of failure of general conservative approach, seems to be limited to soluble fibers (i.e. bulking agents like ispaghula husk and psyllium), but not to insoluble fibers[7]. It also should be recommended to start with low (such as 3-4g

per day) and gradually dose of supplementing (up to 20-30g per day), as abdominal pain and bloating can be worsened by the fermentable properties of the fibers. Suppositories and enemas are largely used by patients with FDDs. However, no controlled study assessing their utility or safety in patients with FDDs is available[18].

2. Osmotic laxatives:

Osmotic laxatives have not be systematically assessed in patients with FDDs, but they can be effective and are currently recommended in case of failure of general conservative approach[7]. Other more recent drugs (like stimulant laxatives, secretagogues, prokinetics) have not been evaluated and are not recommended in the case of FDDs.

3. Biofeedback therapy:

Biofeedback therapy is the preferred and most effective treatment for constipation due to FDDs[7]. Anorectal biofeedback is a behavioral therapy where information about a physiological process is converted to a simple signal to allow the patient to learn how to control the disabled function. Instrumented biofeedback can ameliorate symptoms and accelerate transit by improved defecation effort in up to 70% of patients with slow transit constipation due to dyssynergic defecation [17]. The efficacy of this treatment (up to 70-80% in dyssynergic defecation) has been proved by multiple randomized controlled trials (RCTs)[5] and seems to be effective at long-term without side-effect[7]. This treatment requires at least 4-6 intensive sessions and both patient's motivation and therapist skills can affect response to biofeedback therapy. Moreover, the outcome of biofeedback therapy seems to be worsened in patients with psychological distress[1]. However, there is a lack of studies in patients with ineffective rectal propulsion with or without dyssynergic defecation and a lack of standardization in biofeedback protocols in literature.

4. Other treatments:

Transanal irrigation showed efficacy in patients with chronic constipation of various etiologies, however no study was performed in the particular subpopulation of patients with FDDs.[19] Other therapies like botulinum toxin injection (in anal sphincter or puborectalis muscle) were tried in a few studies[5]. Botulinum toxin injection results are inconstant[5] with an efficacy in up to 50% in patients with dyssynergic defecation[20], but with secondary effects (up to 12.5%) like pain, anal incontinence and/or urgency[5],[20]. Surgical treatments are currently not recommended in the case of FDDs. Based on limited evidence, sacral nerve modulation should not be used in critical practice for management of patients with FDDs [21]. Lastly, surgically-created appendicostomy for delivering antegrade colonic enemas does not seem to be an effective long term solution for patients with FDDs[21].

5. Future research:

Diagnosis of FDDs remains difficult, mainly due to the large overlap between the results of explorations in patients and those of asymptomatic subjects. The recent years provided more accurate definition of FDD. Further research will need to assess and develop more accurate tools to help to diagnose these dysfunctions such as the simulated stool including many pressure, bending and shape sensors named Fecobionics[22]. Possible treatments of FDDs are still scarce in case of failure of biofeedback and should be developed according to the pathophysiological mechanism of FDD.

II. FECAL INCONTINENCE

1. Definition

Fecal incontinence (FI) is defined as at least 2 episodes per month of involuntary loss of feces (solid or liquid) for at least 3 months by the Rome IV criteria[1] (box 3). This definition excludes mucus secretion which should be excluded using careful questioning. Flatus incontinence is included in the definition of anal incontinence but not in those of FI. Lastly, FI

is considered abnormal after a developmental age of at least 4 years, which corresponds to the age of achievement of toilet training.

2. Epidemiology and pathophysiology:

The estimated prevalence of FI varies from 2 to 21% in general population, with differences among studies explained by the definition used, reference time frame and survey methods[23]. FI is associated with worsened quality of life, social isolation and negative economic impact on patients and healthcare systems[24]. Risk factors for FI have been assessed in a few number of studies, but age, bowel disorders in particular diarrhea, symptom of rectal urgency, stress urinary incontinence and burden of chronic illness (co-morbidities count, diabetes) were independent risk factors for FI[25].

Pathophysiology of FI is complex and etiology of FI in one patient is often multifactorial[1]. Common etiologies of FI are summarized in box 4. Anal sphincter dysfunction is the most common abnormality found in patients with FI and can affect internal anal sphincter (e.g. exaggerated spontaneous relaxation or decreased resting pressure) or external anal sphincter (e.g. decreased resting and/or voluntary contraction pressure) or both[1] (box 4). However, isolated sphincter pressures measurement does not always distinguish between continent and incontinent patients. Rectal hyposensitivity can lead to delayed external anal sphincter contraction while stools enter the anal canal and also to fecal retention, in particular when associated with increased fecal compliance[1]. Rectal hypersensitivity may be associated with repetitive rectal contractions during rectal distension and rectal reduced compliance, the latter being more present in patients with urgency symptoms[26].

3. Clinical and paraclinical evaluation:

Management of FI needs a clear appreciation of the type (solid, liquid and/or flatus), the frequency and the quantity to assess the severity of the condition. Therefore, the clinical needs to establish a tactfully relation with the patient to obtain correct information. The amount of fecal leakage can be differentiated between staining, soiling and seepage. Bowel habits characterization is also important and assessed using visual scales of stool form, like the Bristol Stool Form Scale (BSFS) [8], and bowel diaries. Several scales are validated to rate the severity of FI, like the Wexner (Cleveland Clinic), Vaizey (St Marks), Rockwood and the Fecal Incontinence and Constipation Assessment scale[25], and can be easily used in clinical practice at baseline and to assess treatment efficacy (Table 1) . Secondary etiology for FI should also be considered, like the use of drugs that may interfere with transit, anorectal surgical procedures, neurogenic diseases, diabetes mellitus, or scleroderma. Precise description of the fecal leakage can also be helpful, like circumstances, patient's awareness and timing of FI episodes. Two subgroups of FI are commonly found in literature[27]. Urge FI is known as “the inability to defer defecation once the urge is perceived for long enough to reach a toilet”. The second subtype of FI is passive FI, described as “the unvoluntary leakage of feces without forewarning”. By analogy with urinary incontinence, subtypes of FI are commonly used, but their utility regarding pathophysiology and etiologies of FI is still debated[28].

Physical examination is important in investigation of FI, with complete abdominal and neurological examination to determine the possibility of neurological disease. A complete digital rectal examination should be done[1]. A visual inspection may reveal scars resulting from prior obstetric trauma or anorectal surgery or fecal soiling or secondary dermatitis. Anocutaneous and sacral motor reflex should be tested, such as sensitive examination of the perineal area to look for a neurologic disease. Then, meticulous anorectal digital palpation

investigates rectal vacuity, anal sphincter and/or puborectalis weakness at rest and during voluntary contraction, presence of dyssynergia during attempt to defecate and sacral motor reflexes.

Endoscopic evaluation should be proposed in case of diarrhea or recent change in bowel habits. Physiologic testing should be balanced in regard to FI severity, possible etiology, *impact on quality of life and response to first line medical therapy*[1].

Anorectal manometry:

This testing is important in FI as it allows evaluation of anorectal motor and sensory function. Resting anal pressure and squeeze pressure during voluntary contraction can investigate respectively internal anal sphincter function and both external anal sphincter and puborectalis function. Rectal sensory test used different volumes of distension to determine volume thresholds (first constant sensation volume, desire to defecate volume and maximum tolerated volume) and, with rectal pressure measurement for each distension volume, allows calculation of rectal compliance. The recent London Classification[13] provides interpretation regarding disorders of anal tone and contractility and disorders of rectal sensations (box 5). Conventional ARM records pressure data from single points in the canal anal using water perfused or solid-state system, while high-resolution (HR) ARM records circumferential pressure data simultaneously. HR-ARM allows greater physiologic resolution and minimize artefacts movements[29].

Endoanal ultrasound (EAUS):

2D-EAUS is the gold standard for anal imaging and can perform anatomic assessment of the sphincteric complex (i.e. IAS, EAS and puborectalis sphincter) by detecting sphincter atrophy, scarring or defect, and the more recent 3D-EAUS can measure the length and volume of sphincters[29]. Results on IAS and EAS should be interpreted separately and the interpretation of EAS images remains challenging[1]. Defects of both IAS and EAS have

been associated with FI symptoms in literature. Due to the limited access to EAUS, other technics have been developed, like transperineal ultrasonography and vaginal ultrasound, with limited evidence[29]. Endoluminal and external phased-MRI are non-invasive technics that can give additional information on the pelvic floor motion, with similar accuracy as EAUS to detect sphincter defects [29].

Defecography:

This examination should be performed only in selected patients with FI, in particularly before surgery, to confirm or identify pelvic floor structural alterations.

Neurophysiologic tests:

These tests provide information regarding motor and sensory innervation of the pelvic floor area. These tests include electromyography, pudendal nerve terminal motor latencies, rectoanal sensory tests and motor evoked potentials. Electromyography recording correlate well with EAS sphincter pressures obtained using ARM[30]. Due to limited access, their invasive character and concerns about their accuracy[31], these technics are less frequently used in clinical practice.

4. Treatment:

1. Conservative therapy

Conservative therapy is the first step of the management of patients with FI (Figure 3), whose purpose is to treat constipation or diarrhea, optimize stool consistency and to slow down bowel motility, after correction of reversible factors like laxative therapy[1]. Supplementary fibers (psyllium preferentially) and osmotic laxatives should be limited as they can increase stool volume and alter stool consistency if used at high level, in particular in case of sphincter dysfunction[32]. Medications can be helpful to slow down bowel motility (anti-diarrheal medication like loperamide), improve stool consistency (cholestyramine), and to increase IAS

tone (loperamide)[1],[33]. Moreover, measures which aim at improving rectal emptying in case of constipation or fecal impaction, such as suppositories or enemas, fiber supplementation or laxative therapy, and correction abnormal of habit behavior, are recommended[1]. This conservative management step should not be overlooked as it allows to improve the quality of life and to treat approximately half of the incontinent patients[34].

The use of anal plug device is also potentially useful in patients with fecal seepage but maybe difficult to tolerate. However, new variant of a removable anal or vaginal insertion device could improve their results[35]. Physical therapy and biofeedback training are used to coordinate and strengthen the pelvic floor and sphincter function because its efficacy has been proven in approximately 70% of the incontinent patients[36]. In addition, periodic transanal irrigation is a simple and safe long-term treatment for patients who failed bowel modification and physical therapy, with a greatest benefit in patients with neurogenic bowel dysfunction[19]. Trans(per)-cutaneous tibial nerve stimulation cannot be recommended because of its disappointing results on FI[35].

2. Non-conservative approaches

Correctable structural deformities (like rectal prolapse, cloaca, perirectal or rectovaginal fistula) should always be addressed first and surgically treated if possible. In case of their absence, sacral nerve modulation (SNM) is currently recommended as first-line of surgical treatments by the International Continence Society[37]. It is particularly attractive for patients who have both urinary and fecal incontinence because of its established efficacy in many patients with urge urinary incontinence[38]. SNM has an excellent short and medium-term success rate ranging from 81 to 86%[37]. However, only a few data are available regarding long-term efficacy[39]. In these studies, the long-term success rate was estimated at approximately 50% which is not so bad for the treatment of a chronic pathology that may worsen over time[39]. SNM indication is also facilitated by the new compatible MRI

stimulators. Anal sphincter repair (sphincteroplasty), proposed in case of significant sphincter defect, has shown disappointing results with failure rates of greater than 50% after 40-60 months and even greater deterioration at long-term [40]. For this reason, anal sphincteroplasty is largely reserved for women with immediate postpartum sphincter injury.

The use of artificial bowel sphincter and simulated graciloplasty is now limited, due to high rates of infections, erosion and dysfunction of the device [21]. The overall experience with the magnetic anal sphincter has been disappointing and is no longer available[41]. Injection of bulking agents (dextranomer stabilized in hyaluronic acid, NASHA/Dx) showed efficacy in one RCT[42] but these results need to be reproduced before including this treatment in the management algorithm of incontinence. The SphinkeeperTM artificial bowel implant is a new surgical technic but its efficacy remains to be proved[43]. Lastly, in case of failure or contra-indication of all these therapies, fecal diversion with the creation of a diverting colostomy can be proposed to patients.

3. Further research:

Further research is needed in FI, in particular in case of failure of first-line treatments because we only have one recommended treatment (i.e; SNM). Cell therapy (i.e. autologous myoblasts injection in EAS) is currently evaluated in this indication, with promising preliminary results[44]. Rectal injections of botulinum toxin in case of urge FI could also be proposed to patients in the future[45].

III. Focus on rectal capacity: megarectum and microrectum

1. Idiopathic megarectum

Idiopathic megarectum (IMR) refers to a persistent dilation of the rectum in the absence of organic cause (i.e., exclusion of congenital innervation defects such as Hirschsprung's disease) and can be found in a proportion of patients with refractory constipation. From a

clinical perspective, IMR is usually considered in the context of problematic management of patients with recurrent fecal impaction that may begin during childhood or adult life. However, the incidence of IMR is unknown and considered to be an uncommon condition[46]. Even if its pathophysiology is still not fully understood, evaluation of anorectal function has revealed that patients with IMR may have increased rectal compliance, hypomotility and/or sensorimotor dysfunction (enteric nervous system, smooth muscle and cells of Cajal) of the rectum[46]. Consequently, patients also have impaired anorectal evacuatory function, often associated with secondary delay in colonic transit studies.

Historically, diagnosis of IMR was made during double-contrast barium enema or during evacuatory proctography, when the rectal diameter at the pelvic brim was found to be superior to 6.5 cm or 8.5 cm respectively. Alternatively, more recent studies recommend the use of balloon distension during anorectal manometry as a first screening tool, to reduce exposure to ionizing radiation[46]. If elevated maximal tolerable volume could reflect rectal capacity, this method overestimates the presence of IMR. A more reliable technic may be fluoroscopic screening during isobaric distension using a barostat, to identify patients with IMR when the maximum rectal diameter is superior to 6.3 cm at the minimum distending pressure (i.e. the pressure at which sufficient air is insufflated to prevent collapse of the wall but without causing active distension of the rectum) of the rectum[47].

The first-line treatment of IMR is medical involving behavioral retraining techniques[46]. However, these conservative therapies remain ineffective in 50-70% of patients[48]. Moreover, medical therapy must be continued all along life to prevent recurrence of symptoms, with a high risk of poor acceptance. Conservative therapy also fails to achieve restauration of a rectal caliber to normal, even after several years of treatment. Consequently, numerous surgical procedures have been attempted in patients with IMR. Rectal procedures includes the Duhamel procedure and proctocolectomy in case of concomitant megacolon[49].

But such procedures achieved variable results with efficacy varying from 67% to 72% for proctectomy and from 50% to 98% for Duhamel procedure. Moreover, they were associated with substantial morbidity (6.5% and 60% respectively) and mortality (6.5% and 3% respectively)[49]. Vertical reduction rectoplasty (VRR) is a more recent procedure that consists of transection of the dilated rectum in a vertical direction and excision of the anterior portion, thus reducing rectal capacity. VRR involves less radical pelvic dissection and could be a safer alternative in patients with IMR, but there is a need for further studies to confirm its long-term efficacy[49].

2. Microrectum

Microrectum refers to a reduced rectal reservoir. This condition appears to be secondary to a reduced anatomic rectal capacity and/or to an increased rectal wall stiffness and consequently reduced rectal compliance. The etiologies are numerous, such as following rectal surgery (low anterior resection syndrome), irradiation (prostatic, uterine cervical or rectal carcinoma), inflammation of the rectum (ulcerative recto-colitis or Crohn's disease) and IBS. The global prevalence of microrectum is unknown to our knowledge as it can also be an asymptomatic condition. From a clinical point of view, this condition can be found in up to 15% of patients with fecal incontinence and reduced rectal compliance was found to be associated to fecal urgency in one study[50].

The barostat allows the assessment of rectal capacity by measuring rectal compliance. Rectal compliance provides an indirect assessment of rectal wall stiffness (resistance to distension) by using the volume increase for a unit pressure rise. But due to difficult access to this technic, most centers use intra-rectal balloon distension during anorectal manometry to evaluate rectal capacity and perception of rectal filling using the maximal tolerated volume which is decreased in case of microrectum[51]. One limit to this assessment is that it can

underestimate the rectal capacity due to the use of non-compliant bag in most studies and that it is based on rectal sensory function which can be altered in several conditions. Lastly, a few studies also used stool substitute retention test to assess rectal capacity at sensation of desire to defecate [52].

No guidelines are available regarding management of symptomatic microrectum. If possible, treatment of the disease responsible of the reduced rectal capacity should be conducted. In case of FI, conservative treatment is comparable to management of FI previously described. For patients with FI due to low anterior resection syndrome (LARS) following surgery, symptoms can spontaneously and/or in association with biofeedback sessions improve up to 1 year after the procedure, without any correlation with manometric findings[53]. However, in case of persistent LARS, transanal irrigation could be recommended but with a prudent insertion of the probe and inflation of the balloon because of the risk of perforation[54]. Regarding more invasive therapy, a recent meta-analysis including 114 patients with FI following LARS showed an overall success rate of 83.3% with sacral nerve stimulation[55].

CONCLUSION

FDDs and FI are prevalent and incur health impairment and health-care utilization. The last decade has seen an increase in the possibilities of therapy for these conditions, with promising treatments currently evaluated. Using a logical stepwise approach, the clinician has the opportunity to tailor therapy and improve symptoms, quality of life and patient satisfaction.

Highlights

- FDDs management uses a stepwise approach of which biofeedback therapy is an important part.

- The first line of surgical treatment for FI is sacral neuromodulation, even in the case of anal sphincter defect.
- Treatment of idiopathic megarectum is in most cases surgical and vertical reduction rectoplasty seems a promising procedure.
- Sacral neuromodulation seems to be efficient in FI patients with Low Anterior Resection Syndrome, but there is a need for further studies.

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Box

Box 1: Rome IV criteria of Functional Constipation and Irritable Bowel Syndrome with

Constipation

Symptoms must be present for the last 3 months with onset at least 6 months before and without organic pathology.

Irritable Bowel Syndrome with Constipation (IBS-C)

- Abdominal pain presents at least 1 day per week, where the pain is associated with at least 2 of the following symptoms:
 - Change in stool frequency, towards infrequent bowel movements
 - Change in stool form, towards harder stools
 - Related to defecation
- Additionally, predominant stool abnormality of patients on days they have abnormal stools is Bristol 1 & 2 types in more than 25% of all stools, and Bristol 6 & 7 types in less than 25% of all stools, in order to be diagnosed IBS-C.

Functional constipation (FC) :

- Patients do not fulfill diagnostic criteria for IBS-C or Opioid Induced Constipation.
- Symptoms of FC must include at least 2 of the followings:
 - Straining in more than 25% of defecations
 - Lumpy or hard stools (Bristol type 1 & 2) in more than 25% of defecations
 - Sensation of incomplete defecations in more than 25% of defecations
 - Sensation of anorectal obstruction/blockage in more than 25% of defecations
 - Manual maneuvers to facilitate in more than 25% of defecations
 - Fewer than 3 spontaneous bowel movements per week

Subcategories for FDD:

(a) Diagnostic Criteria for **Inadequate Defecatory Propulsion**: inadequate propulsive forces as measured with manometry with or without inappropriate e contraction of the anal sphincter and/or pelvic floor muscles.

(b) 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.

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

Box 2: London classification for the interpretation of disorders of rectoanal

coordination.

This classification requires results of both the expulsion test (using BET or defecography) and the “push” maneuver on manometry.

- **Abnormal expulsion with dyssynergia:** prolonged expulsion with a positive anal pressure change (anal contraction). Minor finding.
- **Abnormal expulsion with poor propulsion:** prolonged expulsion with a reduced rectal pressure change. Minor finding.
- **Abnormal expulsion with poor propulsion and dyssynergia:** prolonged expulsion with both a reduced rectal pressure and a positive anal pressure change (anal contraction). Minor finding.
- **Normal expulsion with abnormal manometric pattern or rectoanal coordination:** any of the 3 push findings described above with the presence of normal expulsion. Inconclusive finding.
- **Abnormal expulsion with normal manometric pattern or rectoanal coordination:** prolonged expulsion in the presence of normal rectal pressure and normal anal pressure change. Inconclusive finding.

BET: balloon expulsion test.

Minor findings: this is a pattern that is seen in patients with anorectal symptoms, and however, may also be seen in healthy control subjects and may represent a physiological alteration associated with symptom generation.

Inconclusive findings: this is a pattern that is seen in patients with anorectal symptoms, but also seen in control subjects.

Box 3: Rome IV criteria for fecal incontinence.

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

Criteria fulfilled for at least 3 months. For research studies, consider onset of symptoms for at least 6 months previously with 2-4 episodes over 4 weeks.

Box 4: common etiologies of fecal incontinence.

- Bowel disorders
 - Diarrhea (Irritable bowel syndrome, post-cholecystectomy diarrhea)
 - Constipation, fecal retention with overflow
- Anal sphincter dysfunction
 - Traumatic (both EAS and IAS): obstetric, surgical (e.g. fistulectomy, hemorrhoidectomy, internal sphincterotomy)
 - Non-traumatic (IAS): scleroderma, idiopathic internal sphincter degeneration
 - Peripheral neuropathy (EAS): localized (e.g. pudendal) or more generalized (e.g. diabetes mellitus, equina syndrome)
- Pelvic floor disorders
 - Rectal prolapse, descending perineum syndrome
- Central nervous system disorders
 - Dementia, multiple sclerosis, spinal cord injury, stroke
- Disorders affecting rectal capacity and/or sensations (may be associated with diarrhea)
 - Proctitis: radiotherapy, Crohn's disease, ulcerative colitis
 - Anorectal surgery
 - Rectal hyposensitivity or hypersensitivity
- Psychiatric disease, behavioral disorders

EAS: External anal sphincter; IAS: Internal anal sphincter.

Table 1: **fecal incontinence severity scores (56-57)**

Incontinent	Never	Rarely	Sometimes	Weekly	Daily
Solid stool ^a	0	1	2	3	4
Liquid stool ^a	0	1	2	3	4
Gas ^a	0	1	2	3	4
Alteration in lifestyle ^a	0	1	2	3	4
Wears pads ^b	0	1	2	3	4
			No	Yes	
Need to wear a pad or plug ^c			0	2	
Taking constipating medication ^c			0	2	
Lack of ability to defer defecation for 15 min ^c			0	4	

Never: no episode in the past 4 weeks; rarely: 1 episode in the past 4 weeks; sometimes: > 1 episode in the past 4 weeks but < 1 week; weekly: 1 or more episodes a week but < 1 a day; daily: 1 or more episodes a day.

^a Vaizey and Wexner items

^b only Wexner item

^c only Vaizey item

Box 5: London classification for the interpretation of A/ Anal tone and contractility and

B/ Rectal sensations:

A/ Manometric diagnoses:

- **Anal hypotension** (major finding) and **anal hypertension** (minor finding) describe respectively reduced or increased anal resting pressure.
- **Anal hypocontractility** describes reduced anal squeeze pressure. Major finding.
- **Combined anal hypotension and hypocontractility** describes co-existent reduction of in both anal resting pressure and anal squeeze pressure. Major finding.

B/ Manometric diagnoses:

- **Rectal hyposensitivity** (≥ 2 thresholds above the upper limit of normal; Major finding) or **borderline rectal hyposensitivity** (1 threshold above the upper limit of normal; Inconclusive finding) describes diminished rectal sensation.
- **Rectal hypersensitivity** (≥ 1 sensory threshold, including MTV, below the lower limit of normal; Major finding) describes heightened rectal sensation.

Major findings: this is a pattern that is not seen in healthy control subjects and seems to represent a physiological alteration associated with symptom generation.

Minor findings: this is a pattern that is seen in patients with anorectal symptoms, and however, may also be seen in healthy control subjects and may represent a physiological alteration associated with symptom generation.

FIGURES

Figure 1. High-resolution manometry tracings reveals patterns that are commonly seen during attempted defecation in a healthy individual (Fig A) and in patients with dyssynergic defecation (Fig B). In a normal pattern of defecation, the subject can generate a good pushing force (increase in intra rectal pressure) and simultaneously relax the anal sphincter. In contrast, patients with dyssynergic defecation exhibit a paradoxical increase in anal sphincter pressure during the attempts of defecation.

Figure 2: proposed algorithm for the management of functional defecation disorders.

Figure 3: proposed algorithm for the management of fecal incontinence.





