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Diagnostic Approaches of Dysfunctional Anorectum and Pelvic Floor Disorders

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Abstract

Various causes of neuromuscular disorders of the pelvic floor muscles may affect the functional integrity of the pelvic floor and anorectum leading to the different pathological conditions such as anorectal incontinence, urinary incontinence and constipation of obstructed defecation, sexual dysfunction, and perineal pain syndromes. Diagnosis of the dysfunctional anorectum and pelvic floor disorders is an integrated approach that includes a thorough history, a careful physical examination, and selected specialized tests as well as the exclusion of organic disease (anorectal or endopelvic) which complement the patient's evaluation contributing to objective and accurate diagnosis of their pathological cause leading to the optimal treatment.

Keywords: anorectal incontinence, constipation, dyssynergic pelvic floor, levator ani syndrome, myofascial syndrome, pelvic outlet obstruction, perineal pain

1. Introduction

Variety causes of neuromuscular disorders of the pelvic floor muscles may affect the functional integrity of the pelvic floor and anorectum leading to the different pathological conditions such as anorectal incontinence, urinary incontinence and constipation of obstructed defecation, sexual dysfunction, and perineal pain syndromes.

Anorectal incontinence is considered a severe condition that influences the patient's life causing mainly unsafety as well anxiety and fear to social contact. The exact incidence of anorectal incontinence is unknown but in a few epidemiological studies it has been reported 2.2–8.3% in the population. It is more frequent in women and particularly in the elderly. However, some studies reported an incidence >50% [1–3].

Constipation can significantly affect the quality of patient's life and its diagnosis may be done at every age but usually in the elderly and in women. The incidence of chronic constipation has been calculated in epidemiological studies 2–27% [4, 5]. The Rome III diagnostic criteria have been helped to constipation definition (**Table 1**) [6]. However, constipation is assumed a subjective symptom of varied pathological

conditions (**Table 2**) [7]. Furthermore, constipation can be distinguished in normal constipation, slow transit constipation, and constipation of pelvic outlet obstruction or obstructed defecation [8]. Constipated patients with obstructed defecation usually have a normal colonic transit time, but delayed transit in the rectosigmoid part [9]. Some of these patients are presented with a megarectum, large rectocele, enterocele, rectal prolapse, or perineal descent, while others maintain a spasm of the pelvic floor muscles leading to difficult evacuation [10].

Perineal pain syndromes are characterized by chronic perineal pain (anorectum and perineum) without anorectal or endopelvic organic disease [11]. These syndromes constitute an idiopathic multifactorial complex interaction between neurological, musculoskeletal, and endocrine systems that is more influenced by psychological and behavioral factors [12]. The lack of understanding of the etiology of perineal pain is evident in its many names (chronic perineal, pain chronic idiopathic anal pain,

Symptom onset at least 6 months before diagnosis
Presence of symptoms in the last 3 months
Diagnosis includes two or more symptoms at least 25% of defecations
<ul style="list-style-type: none">• Straining
<ul style="list-style-type: none">• Lumpy or hard stools
<ul style="list-style-type: none">• Feeling of incomplete evacuation
<ul style="list-style-type: none">• Feeling of anorectal obstruction or blockage
<ul style="list-style-type: none">• Manual maneuvers to facilitate evacuation

Table 1.
Rome III criteria to diagnosis of chronic constipation.

Anorectum and pelvic floor
<ul style="list-style-type: none">• Neoplasms, polyps
<ul style="list-style-type: none">• Megarectum
<ul style="list-style-type: none">• Anal stenosis
<ul style="list-style-type: none">• Mucosal rectal prolapse
<ul style="list-style-type: none">• Internal rectal prolapse
<ul style="list-style-type: none">• Complete rectal prolapse
<ul style="list-style-type: none">• Solitary rectal ulcer
<ul style="list-style-type: none">• Rectocele
<ul style="list-style-type: none">• Enterocele
<ul style="list-style-type: none">• Descending perineum syndrome
<ul style="list-style-type: none">• Hirschsprung’s disease
<ul style="list-style-type: none">• Anismus
<ul style="list-style-type: none">• Dyssynergic pelvic floor
<ul style="list-style-type: none">• Hereditary internal anal sphincter myopathy

Table 2.
Causes of constipation of obstructed defecation.

anorectal neuralgia, levator syndrome, spastic pelvic floor syndrome, spastic levator syndrome, and spastic piriformis) which endeavor to describe the problem [13]. McGivney and Cleveland published an article in 1993 entitled “Levator syndrome and its treatment” [14]. However, the knowledge of the anatomy and physiology of the pelvic floor is of the sine qua non to understand the pathophysiology of chronic perineal pain syndromes [15].

In this review study we present diagnostic approaches of the dysfunctional anorectum and pelvic floor disorders which are accompanied by elements of their etiology and pathophysiology contributing to objective patients’ assessment and to accurate diagnosis of their pathological cause that lead to the appropriate treatment selection.

2. Dysfunctional anorectum and pelvic floor disorders

2.1 Idiopathic anorectal incontinence

Neurogenic anorectal incontinence is considered idiopathic, especially in women, because it may be due to damage of the nerves innervating the pelvic floor muscles. Causes of idiopathic anorectal incontinence are usually associated with difficult childbirth, constipation with chronic straining at stool, rectal prolapse, descending perineum syndrome, and advanced age. Pathophysiology of idiopathic anorectal incontinence is attributed to traction of the pudendal nerve or compression of the sacral nerves by the pelvic floor that descends or to pressure by the fetal head. Evidence that idiopathic incontinence relates to denervation injury of the pelvic floor seems clearly in manometry, electromyography, and pudendal nerve latency studies. These studies show that idiopathic incontinence is characterized by weakness of the pelvic floor and anal canal musculature [16].

2.2 Constipation of functional obstructed defecation

Constipation of functional obstructed defecation may be associated with anismus or dyssynergic pelvic floor, megarectum, Hirschsprung’s disease, and descending perineum syndrome. Anismus is characterized as a pelvic floor dysfunction. The puborectalis muscle and external anal sphincter fail to relax or paradoxically contract during straining to defecate leading to a difficult or impossible defecation. Dyssynergic pelvic floor is characterized by incoordination of the abdominal, rectoanal, and pelvic floor muscles leading to difficult or incomplete evacuation. The pathophysiological mechanism of persistent constipation is the failure of the anorectal angle to open, of the perineum to descend and of the anal canal to shorten as a result of sustained contraction of the puborectalis muscle [17]. Megarectum is a rare condition that is differentiated from Hirschsprung’s disease with rectal biopsies. Patients with megarectum often suffer from constipation (fecal impaction). In these cases there may be an impaired rectal sensation and high distensibility. In addition, impaired rectal sensation and ignoring or resisting the physiological urge to defecate lead to accumulation of more stools in the rectum, which are difficult and painful to expel [18]. Hirschsprung’s disease is another type of pelvic outlet obstruction which is characterized by absence of rectoanal inhibitory reflex. Aganglionosis leads to loss of internal anal sphincter relaxation when the rectum is distended [19, 20]. Descending perineum syndrome is characterized

by a persistent and intractable difficulty to defecate. Abnormal perineal descent during straining to defecate is probably secondary to injury to pudendal and sacral nerves from trauma, childbirth, or chronic straining at defecation [21, 22].

2.3 Perineal pain syndromes

Perineal pain syndromes are characterized by anorectal and perineal pain without anorectal or endopelvic organic disease which should be excluded. The most common perineal pain syndromes are levator ani syndrome, proctalgia fugax, and myofascial syndrome which are characterized by a chronic or recurrent anorectal and perineal pain. Etiology of these syndromes is usually idiopathic. However, they may relate to pelvic injury (fall, accident, and childbirth), surgical procedures (prostatectomy, hysterectomy, low anterior resection, spinal column, and anal fistulae), prolonged sitting in a car or train or hard surface, excessive physical activity, psychological stress, anxiety, and sexual abuse. Pathophysiology of levator ani syndrome is similar to that of dyssynergic defecation (incoordination between anorectum and pelvic floor muscles during defecation). The failure of relaxation of levator ani (puborectalis) or the external anal sphincter muscles or paradoxical contraction of them during straining to defecate was called spastic pelvic floor syndrome. Pathophysiology of proctalgia fugax has been associated with spasm of pelvic floor, abnormal contractions of internal anal sphincter, and hypertrophy of internal anal sphincter (inherited myopathy). Pathophysiology of myofascial syndrome relates to “trigger points” which are connected with the disturbance of the nerve endings and an abnormal contractile mechanism at many dysfunctional endplates. These endplates constitute the sites of active trigger points [23].

2.4 Evaluating patients with dysfunctional anorectum and pelvic floor disorders

In the diagnostic approach of dysfunctional anorectum and pelvic floor disorders (idiopathic incontinence, constipation of functional obstructed defecation, and perineal pain syndromes) that contribute to the history, a careful physical examination, specialized investigations, and the exclusion of anorectal or endopelvic organic disease should be carried out. In incontinent patients, the history may elicit leaking of enteric content (gases, fluid stool, or formed stool) and record the frequency, duration, severity, and timing of incontinence episodes. The incontinence may be true or false (overflow diarrhea), passive (neurogenic incontinence) or uncontrolled (diarrhea, trauma of anal sphincter, or puborectalis). However, a past medical history (obstetric, anorectal surgeries, constipation with straining at stool, rectal prolapse, low back pain, sciatica, and medications) should carefully be assessed. On physical examination should be looked for signs of incontinence. Perineal inspection (at rest and strain) may show perianal soiling, patulous anus, scars, prolapsing hemorrhoids, perineal descent, or rectal prolapse. The absence of anocutaneous reflex indicates pudendal neuropathy. Digital rectal examination may reveal tumors or impacted stool and at the same time allows the internal and external sphincter function evaluation (at rest and squeeze) as well as anorectal ring of the puborectalis assessment by palpation. Rectosigmoidoscopy should always be carried out to exclude neoplasms, proctitis, internal rectal prolapse, or a solitary rectal ulcer. At the end of the patient's interview should be determined the degree of incontinence (mild, moderate, and severe) and then some specialized tests should be recommended, if necessary. This can be done using a proposed incontinence scoring

system (e.g., Wexner, Pescatori, and Altomare) [24–26]. Thus, clinical evaluation is complemented by anorectal physiology tests (anorectal manometry, anal endosonography (AES), pudendal nerve terminal motor latency (PNTML), defecography, electromyography (EMG), and MRI) which provide objective patients' assessment and accurate diagnosis of the incontinence cause contributing to the appropriate treatment. However, in clinical practice, anorectal manometry, AES, and PNTML have been shown to be the most useful tests in diagnosis and after-treatment follow up [27, 28]. Anorectal manometry is the first investigation for anorectal physiology which may assess the anal sphincters function (at rest and squeeze), rectal sensation, and rectoanal reflex. In incontinence, anal canal pressures (at rest and squeeze) are low with or without impaired rectal sensation. Rectal sensation disorder may be managed by biofeedback [29]. AES is another examination in the diagnosis of anorectal incontinence, providing information about the anal sphincters integrity [30]. PNTML assesses the pudendal nerve function. Prolongation of PNTML is considered the diagnostic evidence of idiopathic incontinence [31]. Defecography is a useful test which may show anatomical and functional abnormalities of the anorectum and pelvic floor contributing to the anorectal angle assessment that is obtuse in idiopathic incontinence patients [32]. However, the recently used dynamic MRI of the pelvic floor in defecatory disorders may be a more efficient alternative to traditional defecography [33]. Furthermore, recent studies suggest that the defecography can also be replaced by perineal ultrasound [34]. EMG may detect functional anal sphincter abnormalities in incontinent patients with normal AES [35]. In constipated patients, the history may derive valuable information concerning the characteristics of patient's symptoms, the duration, and severity (difficult, painful, incomplete, or impossible defecation) but also the stool frequency, stool consistency, and stool size. A past medical history, obstetric, surgery, neurological, psychological, or medicines should be recorded. However, a recent history of severe constipation or overflow diarrhea in elderly should be carefully investigated to exclude an organic pathology (neoplasm) or impacted stool. Physical examination includes examination of the abdomen, perineum, and anorectum. Abdomen examination should exclude an intra-abdominal mass or tenderness. Perineal inspection may reveal a patulous anus, soiling, scars, prolapsing hemorrhoids, fistulas, or fissure. Digital examination may detect stool (fecal impaction), stricture, or neoplasm and at the same time should be done an anal sphincter function assessment (at rest, squeeze, and straining). If not observed pelvic floor dysfunction (pelvic outlet obstruction or dyssynergia), unusual perineum bulging or rectal prolapse may be noticed. Furthermore, an anterior rectocele or enterocele should be sought. Physical examination should always be followed by rectosigmoidoscopy which can identify anorectal and colonic pathologies (stricture, neoplasm, internal rectal prolapse, megarectum, inflammatory bowel disease, or solitary rectal ulcer). However, diagnostic approach of constipated patient is completed by selected specialized investigations which can diagnose and differentiate with accuracy and objectivity the constipation causes of obstructed defecation leading to optimal treatment. Specialized tests include colonic transit time test, anorectal manometry, defecography, balloon expulsion test, and EMG. In addition, new specialized techniques as high-resolution anorectal manometry, dynamic MRI, and dynamic perineal ultrasound have been used and proved useful in the diagnostic attempt of the dysfunctional disorders cause of the anorectum, pelvic floor, and colon [36–39]. Colonic transit time is estimated by an abdominal X-ray 5 days after using radiopaque markers. Retention of the markers in the rectosigmoid colon suggests a dyssynergic pelvic floor and pelvic outlet obstruction [40].

Anorectal manometry (at rest and straining) may show motor dysfunction of the anorectum (impaired anal relaxation-anal resting pressure unchanged) or (paradoxical anal contraction-anal resting pressure increases), or both [41, 42] and sensory dysfunction of anorectum (impaired rectal sensation and high distensibility-threshold for first sensation and for call to defecate elevated) in constipation of obstructed defecation [43, 44]. Rectoanal inhibitory reflex is usually present except in Hirschsprung's disease that is absent [45]. However, the new technique of high-resolution anorectal manometry seems to have more advantages compared with conventional manometry (easier use, more accurate values of the anorectal pressures, complete anorectal imaging, and automatic analysis of the recording results with color morphology) allowing a most comprehensive diagnostic approach of the dysfunctional anorectum and pelvic floor disorders as idiopathic incontinence, dyssynergic pelvic floor, and Hirschsprung's disease [46–48]. Defecography, in patients with functional obstructed defecation, may show acute anorectal angle as an inability of puborectalis muscle relaxation or a spastic pelvic floor [49, 50]. However, recently, dynamic MRI of the pelvic floor and dynamic perineal ultrasound can be considered an alternative to traditional defecography [51]. In balloon expulsion test, patients with obstructed defecation are unable to expel the balloon [52]. EMG can recognize a dysfunctional puborectalis or/and external anal sphincter, in cases with obstructed defecation (anismus or dyssynergia) during straining, recording to increased their pathological activity [53, 54]. In patients with perineal pain syndromes, in the diagnostic approach of patients with chronic perineal pain an important place occupy the thorough history, careful physical examination, and selected specialized tests. Perineal pain syndromes are clinically distinguished by the duration of painful episodes, frequently, and characteristics. However, perineal pain of dysfunctional pelvic floor syndromes should be distinguished by pelvic pain. Pelvic pain usually relates to pathological conditions such as gynecological or urological diseases, infection, irritable bowel disease, and neurological disorders. All these pathological conditions may affect the perineal muscles (pelvic floor, anal, and urethral sphincters) and sometimes may pretend dysfunctional syndromes with perineal pain, as the levator ani syndrome. So, the history may give useful information about the characteristics of pain, the location, duration, frequency, provocative factors, and factors of worsening the pain. Furthermore, the past medical history (medicines, pelvic injury, surgical procedures, excessive physical activity, psychological distress, anxiety, psychical trauma, sexual abuse, and psychical disease) should be recorded. Physical examination includes inspection and palpation of the perineum. Digital rectal and vaginal examination is significant in the assessment of the anorectum and levator ani muscle (puborectalis). The diagnosis of dysfunctional pelvic floor syndromes is based on characteristics symptoms, digital examination findings of levator ani palpation (tenderness, contraction, or sensitive trigger points), pathological tests as electromyography and exclusion of the anorectal or endopelvic organic disease with perineal pain. Nevertheless, the diagnosis of these syndromes may be difficult because they constitute overlapping functional entities. The differential diagnosis of chronic perineal pain includes neoplasms of anorectum, anal fissure, anorectal abscess, thrombosed external hemorrhoids, proctitis, cystitis, endometriosis, internal rectal prolapse, descending perineum syndrome, solitary rectal ulcer, leukemia, and neurological disorders spinal column or spinal cord [23].

In conclusion, diagnostic approaches of dysfunctional anorectum and pelvic floor disorders include the history, a careful physical examination and selected specialized

tests as well as the exclusion of the anorectal or endopelvic organic disease that contribute to objective and accurate diagnosis of their pathological cause leading to the optimum treatment.

Conflict of interest

The authors declare that they have no competing interests.

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