# 4 Neurofunctional Diagnosis and Anorectal Manometry

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## 4.1 Introduction

Fecal incontinence (FI) is defined as the recurrent uncontrolled passage of fecal material for at least 3 months [1]. Anal incontinence is characterized by failure to control the elimination of stool and/or flatus [2]. The pathophysiology has a multifactorial etiology including anal sphincter lesions, rectal reservoir impairment, rectal sensation damage, loss of pelvic floor integrity, damage to the pelvic floor nerve supply, cortical awareness weakening, and stool volume and consistency. All these alterations combine variously with each other and provide different pathogenetic profiles of patients. The diagnostic work-up of FI is applied in any individual with a developmental age of at least 4 years [1] and it is based on imaging techniques, to study the morphological structure of the anal sphincters and pelvic floor, and on functional instrumental studies, to evaluate neuromuscular anorectum function. Among the latter, neurofunctional tests and anorectal manometry are the principal techniques used.

## 4.2 Neurofunctional Diagnosis

The pelvic floor muscles and the external anal sphincter (EAS) are innervated mainly by the pudendal nerve (S2–S4) branches: right and left inferior rectal nerves [3, 4]. The neurofunctional diagnosis, in patients affected by anal incontinence, uses tests that study these anatomic structures and may offer data that can be useful in identifying the pathophysiological profile of incontinence. A systematic literature review was recently published to provide clinicians with evidence-based recommendations on the use of neurophysiological tests in clinical practice [5]. It

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concluded that the actual clinical usefulness of these tests has not yet been completely clarified and recommends pelvic floor neurophysiology studies when anal incontinence is present in patients affected by cauda equina and/or conus medullaris lesions, pudendal neuropathy, muscular diseases, spinal cord lesions and parkinsonisms [5].

Therefore, the neurophysiological evaluation of the pelvic floor should be reserved only for patients with a negative instrumental workup for anal incontinence and in whom there is a suspected neurological pathogenesis. The individual tests are described below.

### 4.2.1 Anal Electromyography

Electromyography (EMG) of the external anal sphincter and puborectalis muscle measures amplitude as well as duration of muscular action potentials during rest, voluntary anal contraction and attempted defecation [6]. EMG can be performed via three methods: using a needle electrode, a surface electrode on the perianal skin, or a cone-shaped plug in the anal canal. The needle electrode usually analyzes the anal area, which is subdivided into four quadrants, and may reveal, in incontinent patients, areas of sphincter damage that will display prolonged or absent action potentials. This method can also be helpful in mapping sphincter defects when there is dense scarring that can cause artefacts on endoanal ultrasound. Polyphasic potentials are an expression of reinnervation [7]. Needle electrode EMG use is limited by the anal pain induced during examination. The surface electrode and cone-shaped plug techniques can provide a global anal assessment rather than assessment of each EAS quadrant, but they are painless and more comfortable. They can be used for EMG biofeedback sessions.

### 4.2.2 Sacral Reflexes

Electrophysiological sacral reflexes are responses of the pelvic floor striated muscles to electric stimulation of the perineal skin, mucosa, or pelvic nerves [8]. These involve a spinal reflex arc. Sacral evoked potentials are responses recorded after pudendal nerve stimulation or stimulation of the urethral or anal skin. Stimulation of the dorsal nerves of the glans penis and clitoris, the mucosa of the rectum, bladder, and urethra, and the skin of the distal parts of the legs may give rise to an EAS response, and to a reflex response of the bulbocavernosus muscle. The first is named "pudendoanal reflex" with anal recording of the reflex, the second is the "bulbocavernosus reflex" recorded either with a concentric needle electrode or by placing two surface electrodes over the muscle site just below the scrotum. Both reflexes have an afferent limb and efferent fibers, with the sacral cord as reflection center. For this reason, the pudendal nerve terminal motor latency (PNTML)—induced by a fingertip electrode and with responses captured by another electrode at the base of the finger—is considered only to be valid for the analysis of efferent pathways of the pudendal nerve and not for the evaluation of the entire spinal reflex arc. An abnormal prolonged PNTML can suggest a pudendal nerve lesion. This test is, however, considered to be operator-dependent because proper placement of the fingertip electrode over the pudendal nerve is critical.

In patients affected by anal incontinence, the latency of the pudendoanal reflex can be increased due to pudendal nerve damage causing slowed conduction; this reflex response is absent in conus medullaris and cauda equina lesions, but normal in spinal cord or brain injury [5].

### 4.2.3 Evoked Potentials

Evaluation of somatosensory evoked potentials (SSEP) provides information about the integrity of the somatosensory afferent pathways from the pudendal nerve to the parietal cortex. This test may provide a concrete demonstration of altered transmission of stool and gas perception to the cerebral cortex. Motor evoked potentials (MEP) are recorded after transcranial magnetic stimulation, and they may demonstrate the defective transmission of cortical signals to the pelvic floor muscles. SSEP and MEP can be useful in patients with spinal cord or cauda equina lesions and pelvic symptoms, but they are not routinely performed in anal incontinence.

## 4.3 Anorectal Manometry

Anorectal manometry (ARM) analyses fecal continence mechanisms and, when applied in anal incontinence patients, identifies anal sphincter weakness, anorectal reflex abnormalities, rectal sensation impairment, and poor rectal compliance.

Several systems and probes exist for performing ARM. The perfusion system, originally developed by Arndorfer et al. [9], employs catheters perfused with distilled water infused by a pneumohydraulic pump, to obtain a steady perfusion rate of 0.2–0.4 mL/min. Occlusion of the catheter channels increases intraluminal pressure and produces resistance to the flow of water. This resistance is measured by transducers, interpreted as compliance of the tissue and converted into pressure values shown on a trace. High sensitivity (92.2%) and good specificity (86.6%) are reached when ARM is applied in FI patients [10]. In 2007, high-resolution ARM (HRAM) was introduced into clinical practice [11], and a few years later high-definition 3D solid-state ARM (HDAM) was developed [12]. Currently, both these techniques employ multi-use solid-state catheters, and pressure values are expressed according to a color scale in ascending order from green, the lowest values, to purple, the maximum values. HADM, through dedicated software, displays 3D cylindrical topographical models of the anal canal which can be rotated and viewed from all sides; in this way focal sphincteric defects may be detected in anal incontinence [13].

Regardless of the manometric system adopted, the recording steps are identical.

*Anal resting pressure* (ARP) detects the basal tone of the anal canal: one study assigned approximately 55% of ARP to the internal anal sphincter (IAS), 15% to the vascular anal cushions and the remaining 30% to the EAS [14].

*Maximal voluntary contraction* (MVC) is the squeeze pressure obtained by asking the patient to contract the anus with maximal strength: it reflects the activity of the EAS and puborectalis muscle. ARP and MVC tend to be higher and the normal range of anal pressure is relatively wide when measured with HRAM.

*Rectoanal inhibitory reflex* (RAIR) is the reflex inhibition of IAS tone and is considered part of the sampling reflex. Transient relaxation of the IAS allows rectal content, with small fecal volumes or gas, to come into contact with sensory receptors placed in the upper anal mucosa. In this way, the rectal content may be cortically typified and the subject is alerted to discharge flatus or to defecate [15]. Detection of rectal sensation is obtained by inflating, with air, a balloon at the tip of the catheter. Increasing volumes are used until values are reached to identify the *conscious rectal sensitivity threshold* (CRST: the lowest volume for the first sensation), the *constant sensation* (CS: the volume with call to stool) and the *maximal tolerated volume* (MTV: the threshold volume for urgency to defecate and for pain). Finally, the monitoring of *rectal compliance* reflects the tonic adaptation of the rectal wall to increasing volumes, and is determined by the ratios $\Delta P/\Delta V$ measured at different volumes of inflated air.

According to the clinical guideline of the American College of Gastroenterology (ACG), ARM is worth implementing in patients affected by anal incontinence [16]. Recently, the International Anorectal Physiology Working Group (IAPWG) published standardized testing protocols for HRAM and the London Classification for disorders of anorectal function was developed [17]. In incontinent patients, HRAM provides data for identification/quantification of impaired anal sphincter function and abnormal rectal sensitivity (both hyper- and hyposensitivity types). ARM, as explained above, offers objective data on continence mechanisms and may suggest which mechanism may be malfunctioning in incontinent patients. However, the manometric findings must be complemented with data obtained using other diagnostic techniques assessing morphology (anoscopy, MRI, endoanal ultrasound) and function (anal neurophysiologic tests).

ARP may be lower in anal incontinence, and the related hypotonic anal canal is usually due to IAS impairment, especially if passive incontinence occurs [18]. Unfortunately, the discriminative power of ARP between continent and incontinent patients is low because of the wide range of normal pressures. HDAM could be useful in detecting structural anorectal abnormalities such as sphincter defects, descending perineum and rectoanal intussusception. Objective criteria for sphincter defects, moreover, might be useful to better select patients for endoanal ultrasound [13].

MVC is frequently impaired in patients affected by anal incontinence: the amplitude and duration of squeeze tone are lower than in healthy controls [19], or sometimes inappropriate relaxations may occur (Fig. 4.1). MVC impairment is related to EAS and puborectalis malfunctioning and is found in patients with urge incontinence who lose stools because of the failed suppression of defecation [18]. HADM

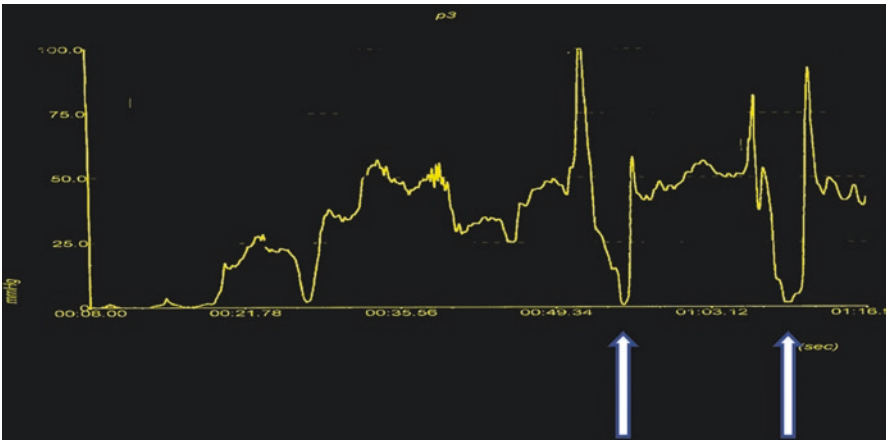


**Fig. 4.1** Maximal voluntary contraction. Spontaneous inappropriate relaxations occur (*white arrows*) during anal contraction

may detect EAS defects, for example after obstetric injury or anal surgery. The absence of EAS defects could suggest pudendal neuropathy and the pudendoanal reflex should be used for diagnosing pudendal nerve injury.

Detection of RAIR is an important tool for evaluation of rectoanal coordination. This reflex often cannot be elicited when ARP is extremely low (<10 mmHg) and, therefore, in this condition it is not possible to judge if the reflex is present or absent, normal or impaired. However, there are some reports of RAIR impairment. A longer RAIR duration has been detected in patients with fecal soiling and pruritus ani [20]. The duration of RAIR is also longer in patients affected by idiopathic FI than in controls [21]: the reflex exhibits a prolonged contraction time with a slow return to basal prestimulation values. This prolonged inhibition impairs the continence mechanisms. Small amounts of feces reach the inhibited proximal anal canal but there is poor distal EAS recruitment: therefore, in the presence of a sensitivity threshold higher than the RAIR threshold, fecal passive incontinence may occur. The absence of RAIR is a manometric sign that is often detected in ultralow rectal anterior resection [22]. It is the consequence of the total excision of the rectum such as in ultralow anterior resection with coloanal anastomosis: the descending recto-anal pathway is easily interrupted. Thus, the RAIR may be abolished, and the coordinated sensory-motor integration of the rectum is distorted with a possible negative influence on fecal continence.

The last but not the least step of ARM is the detection of rectal sensation and rectal compliance. Significant alteration of rectal sensation (hyposensitivity with high volumes or hypersensitivity with low volumes, according to the London classification) may be detected in patients with diabetes mellitus [23] or multiple sclerosis [24]. Impaired recognition of impending defecation can lead to FI. Hyposensitivity means passive incontinence if the EAS reflex contraction is weakened in the presence of rectal content. Hypersensitivity is present in urge incontinence when small volumes are involuntarily expelled because of impaired volumetric rectal capacity such as in patients with inflammatory disease (ulcerative colitis) or rectal fibrosis (actinic proctitis).

The compliance of the rectum expresses the elastic distensitivity of the rectal wall while the MTV measures the volumetric capacity of the rectum. Both values are an expression of rectal containment capacity, the first adapting the wall tension to the contents without increasing endorectal pressure, the latter as the expression of the maximum filling volume. Impaired compliance, often combined with low MTV, is present after sphincter-saving surgery [22] and explains urge incontinence that is the main symptom of anterior resection syndrome.

In addition to its diagnostic aspects, ARM may assist in the selection of therapy for patients with anal incontinence. The selection of incontinent patients for overlapping sphincteroplasty may be appropriate when low ARP (<10 mmHg) and low MVC (<40 mmHg) are detected [25]. The same cutoff values identify rectal prolapse patients who are at high risk for FI after surgical repair and suggest adoption of sphincteric correction combined with the chosen surgical technique [26]. ARM can also play an important role in multimodal rehabilitation of FI patients [27]. The algorithm of this rehabilitation program is based on manometric data. Biofeedback and pelviperineal kinesitherapy are suggested by low ARP or weak MVC. Volumetric rehabilitation (sensory retraining) is indicated for impaired rectal sensation and/or damaged compliance. Electrostimulation is only a preliminary step when patients need to better feel the anoperineal plane and increase their awareness. The usual sequence of techniques is: (1) volumetric rehabilitation; (2) electrostimulation; (3) pelviperineal kinesitherapy; (4) biofeedback. The same protocol has been used in patients with FI after sphincter-saving surgery. Many patients showed improvement of the Wexner Incontinence Score (58%): impairment of both MTV and compliance was associated with bad postrehabilitation results [28].

In conclusion, ARM may be considered an important tool in the diagnostic work-up of anal incontinence. It also offers data for understanding the pathophysiology of incontinence and can help physicians to modify the therapeutic strategy appropriately.

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