Coloproctology 1
Series Editor: Carlo Ratto

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Colon, Rectum and Anus: Anatomic, Physiologic and Diagnostic Bases for Disease Management

Springer

Coloproctology

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Coloproctology is a modern discipline covering a vast area of medicine, including all diseases and disorders of the colon, rectum, and anus. Physicians and non physician personnel are very interested in the field owing to the high prevalence of these clinical conditions in the general population, the severity of secondary symptoms and/or disabilities, the diagnostic and therapeutic issues, and the personal and social implications. In particular, a variety of specialties and subspecialties are involved in the clinical management of colon and anorectal diseases/disorders, which frequently entails a multidisciplinary approach. This book series will provide detailed coverage of a wide range of topics in Coloproctology, focusing particularly on recently introduced and emerging diagnostic and therapeutic techniques. Each volume will be a reference work on a specific disease or disorder. The core aim is to provide a sound and productive basis for clinical practice, and to this end some of the most highly regarded experts worldwide will contribute as co editors and authors. The series will also help researchers and all those interested in the field to identify key issues in Coloproctology in order to foster the development and implementation of further new technologies.

More information about this series at http://www.springer.com/series/13364

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Editors

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With 181 Figures and 54 Tables

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Foreword

The scientific publisher Springer is bringing out a new series of books on Coloproctology with Professor Carlo Ratto as the Editor. It will consist of several volumes dealing with broad subdivisions of the speciality with chapters written by internationally acknowledged experts in the field. As in all other areas of medicine, coloproctology has progressed to an extraordinary degree over the last 10 years.

While dealing with recent developments and the innovations that have occurred, the first volume maintains an emphasis on clinical assessment and decision taking. One of its themes is the integration of the results of different investigations and other forms of objective assessment with the clinical picture to allow the construction of treatment strategies applicable to the individual patient.

The book consists of four parts including an Introduction to Coloproctology, Anatomy, Physiology, and Diagnosis. Its emphasis is on proctology, and besides dealing with the actual diseases, there is a great emphasis on the clinical evaluation of anorectal disorders and their investigation. In a speciality where the diagnosis is made by clinical evaluation in over half the patients, the inclusion of a chapter describing the office visit in detail is very informative and will be most valuable to readers. The book deals with the anatomy and physiology of the colon and rectum, the instruments used in proctological practice, and the approach to investigation. The quantification of symptom severity is considered in a chapter on scoring systems. Data collection and analysis are dealt with in detail, and there are several chapters on physiological investigation, radiological imaging particularly ultrasound, and endoscopy. The chapters on electrophysiology of the pelvic floor and ultrasound and physiology of the rectum and anus are written by experts who have been at the forefront of research into these fields. Changes in practice are reflected by a chapter on technical advances in imaging which have resulted in changing concepts in the management of rectal cancer and organ preservation in this important disease in the light of recent developments.

Subsequent volumes will deal individually with all important proctological conditions such as hemorrhoids, anorectal sepsis, incontinence, and more. Others will focus on the major topics of inflammatory bowel disease and benign and malignant neoplasia.

This first volume of the series will be of great value to established specialists and trainees in this important field.

R. J. Nicholls
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Preface

Coloproctology is an amazing field of modern medicine that fascinates many surgeons, but also many others in the medical field. The evolution of knowledge and continuous progress in technologies has significantly changed this discipline over the last 20 years, and it is difficult to define its "state of art" today. To address this continuum of advances in this fascinating field, our book series is being published in both an electronic and printed format, allowing us to avoid trying to pinpoint the progress of coloproctology at this point in time (which would result in the information being "old" in a few months). Thanks to the "Major Reference Works" formula, in which living editions of reference works are updated by the publisher (Springer) as scientific developments warrant, readers can access further evolutions of each chapter after publication of the print edition by consulting the updated electronic contents.

The purpose of this project is to provide a panoramic view of the topic, ranging from the basics (including anatomy and physiology of the colon, rectum, and anus, oriented at immediate application in diagnosis and treatment) to principles of patient management. The first volume is dedicated to the basic anatomy, physiology, and principles of diagnosis in order to offer the keys of access to this specific discipline. Due to the variety of different clinical conditions, the following volumes of *Coloproctology* have been structured as several monographic books, dedicated to hemorrhoids, anal fistula and abscess, fecal incontinence, constipation and obstructed defecation, chronic inflammatory bowel diseases, miscellaneous benign colorectal and anal diseases, and neoplasms of the colon, rectum, and anus. Functional disorders, inflammatory diseases, benign neoplasms, malignant tumors, infectious diseases, and miscellaneous abnormalities and disorders affecting the colon, rectum, and anus are also all addressed.

Each book aims to discuss the main open questions regarding the pathophysiology and diagnosis of each topic along with current points of view, thereafter debating the actual strategies for treatment. Wherever the choice between a variety of diagnostic and therapeutic options would be controversial, a "virtual round table" has been set up, giving readers the pros and cons of different leading opinions. As would be expected, the panel of contributors is of the highest worldwide scientific level, reflecting the best clinical practice on each topic, and ranges from surgeons to gastroenterologists, oncologists, radiotherapists, radiologists, internists, specialists in abdominal and pelvic diseases and disorders, etc. When appropriate, an updated review of the literature is summarized in tables within the chapters, and a number of figures provide useful examples of cases diagnosed using different modalities of imaging and treated with different surgical approaches.

This book series aims to be a reference for not only coloproctologists, but for all specialists involved in the management of disorders and diseases of the large bowel and anus, medical students, and other professionals training in healthcare.

Finally, we are delighted that our *Coloproctology* book series is accessible to a wide audience through SpringerLink (http://link.springer.com/), the publishing platform for Springer's major reference works.

Carlo Ratto, M.D., FASCRS
Angelo Parello, M.D.
Lorenza Donisi, M.D.
Francesco Litta, M.D.

Contents

About the Editors

Carlo Ratto He was born in Naples, Italy, on July 6, 1962.

He was graduated in Medicine and Surgery at the Catholic University of Rome in 1987.

He is Researcher at the Department of Surgical Sciences, Catholic University, Rome.

His clinical practice, at the University Hospital "A. Gemelli" concerns particularly the colo-recto-anal disorders and diseases. He is the Chief of Proctology Unit.

He is actively involved in research on:

– Anorectal physiology, in particular concerning anorectal manometry, electrophysiology studies, and endoanal ultrasound in benign anorectal disorders. In particular, he is actively involved in the clinical application of three-dimensional endoanal ultrasound.
– Fecal incontinence and constipation, in particular concerning pathophysiology and treatment with traditional and novel therapeutic modalities (sphincteroplasty, graciloplasty, sacral neuromodulation, bulking agents).
– Hemorrhoids, in particular concerning pathophysiology and treatment with traditional and novel therapeutic modalities (THD procedure).
– Fistula-in-ano, in particular concerning the assessment of fistula and abscess with endoanal ultrasound related to surgery results.
– Rectocele, in particular concerning pathophysiology and modalities of clinical presentation and treatment options.
– Anal cancer, in particular concerning staging and restaging of the tumor and integrated therapies (chemoradiation).
– Colorectal cancer, in particular concerning diffusion modalities of the tumor and prognosis, integrated therapies (surgery, chemoradiation, intraoperative radiation therapy), and molecular biology.

He is author of a number of scientific publications on international journals and has presented results of his research at national and international scientific meetings.

He is Editor of the book entitled *Fecal Incontinence. Diagnosis and Treatment*, Springer Ed., May 2007.

He is active member of:

– American Society of Colon and Rectal Surgeons (ASCRS), Fellow
– European Society of Coloproctology (ESCP)
– Italian Society of Colorectal Surgery (SICCR)
– International Anal Neoplasia Society (IANS)

He was National Scientific Secretary of the Italian Group for Sacral Neuromodulation (GINS).

He was Delegate of Italy to the European Society of Coloproctology (ESCP).

He was General Secretary of the Italian Society of Colorectal Surgery (SICCR), 2006–2007.

He is Vice-President of the Italian Society of Colorectal Surgery (SICCR), 2015–2017.

He is Member of the Editorial Board of *Techniques in Coloproctology* and *World Journal of Gastroenterology*.

Angelo Parello He was born in Agrigento, Italy, on October 17, 1980. He was graduated with honors in Medicine and Surgery at the Catholic University of Rome in the first session of the academic year 2003–2004.

From 2004 to 2010, he was resident in General Surgery at Catholic University of Rome, which he attended with particular interest directed to diagnosis and treatment of coloproctologic diseases.

His clinical practice is mainly directed to diagnosis and treatment of colorectal-anal diseases, and he is an expert in performing both anorectal and pelvic floor diagnostics tests (e.g., anorectal manometry and endoanal and transrectal ultrasound) and treatment (e.g., transanal hemorrhoidal Doppler-guided dearterialization for hemorrhoidal disease, sacral neuromodulation for fecal incontinence and constipation).

He collaborated in the development of a novel minimally invasive surgical approach to treat fecal incontinence – THD implant Gatekeeper – now available for use in the world.

He is active member of the Italian Society of Colorectal Surgery (SICCR), and in 2007–2008 was member of the Guidelines Commission on behalf of this Society.

He was teacher and tutor in many national and international courses conducted in Italy and in other European countries.

He is author of several scientific publications on international journals, author of several chapters in books, and has presented results of his research at national and international scientific conferences.

Lorenza Donisi She was born in Caserta, Italy, on March 23, 1982. She was graduated in Medicine and Surgery at the Catholic University of Rome in 2007 with first class honors (110/110 cum laude). She is a specialist in General Surgery since 2014.

She is a PhD student in Oncological Sciences at the Catholic University, and performs daily clinical and research activity at the Proctology Unit of the "Fondazione Policlinico Universitario A. Gemelli," Rome, Italy, directed by Prof. Carlo Ratto, M.D., F.A.S.C.R.S.

Her daily activity and research are directed on the clinical aspects, diagnosis, and treatment of the anorectal disease, mainly with regard to fecal incontinence, obstructed defecation and colonic constipation, inflammatory bowel disease, hemorrhoidal disease, anal and perianal sepsis, and anal and rectal malignancies.

She is involved in studying the anorectal and pelvic floor physiology, by using tests as anorectal manometry and three-dimensional endoanal and transrectal ultrasound.

She is author of several scientific publications on international journals, author of several chapters in books, and has presented results of her research at national and international scientific conferences. She is a teacher in a series of national and international courses on the management of coloproctologic diseases.

She is active member of the Italian Society of Colorectal Surgery (SICCR) and is member of the Communication Commission of this Society.

Francesco Litta He was born in Matera, Italy, on April 25, 1983. He was graduated in Medicine and Surgery at the Catholic University of Rome in 2008 with first class honors (110/110 cum laude). He is a specialist in General Surgery.

He is an attending physician at the Proctology Unit of the "Fondazione Policlinico Universitario A. Gemelli," Rome, Italy, directed by Prof. Carlo Ratto, M.D., F.A.S.C.R.S.

His clinical practice focuses mainly on the diagnosis and treatment of disease of the colon and the rectum, and all proctological diseases. He is involved in the anorectal physiology testing, with particular interest in the evaluation of benign and malignant disease by means of anorectal manometry and three-dimensional endoanal ultrasound.

His research activity is mainly based on the evaluation and treatment of patients affected by fecal incontinence, constipation, anal fistula, hemorrhoidal disease, obstructed defecation, and inflammatory bowel diseases (Crohn's disease, ulcerative colitis), with studies concerning the pathophysiology and the surgical treatment by means of traditional and new minimally-invasive therapeutic options.

He is author of several scientific publications on international journals, author of several chapters in books, and presented results of his research at national and international scientific conferences. He is teacher in a series of national and international courses on the management of coloproctologic diseases.

He is active member of the Italian Society of Colorectal Surgery (SICCR) and is member of the Guidelines Commission on behalf of this Society.

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1 A Surgeon for the Pelvic Floor: Dream or Reality?

Tracy Hull

Contents

Abstract

As surgeons, we have evolved from barber surgeons to superspecialists. In the future, the pelvic floor surgeon will be even more specialized and most likely cross-trained so there is true understanding of all aspects of pelvic floor disease. Probably more importantly, there will be a team for the pelvic floor, and pelvic floor problems will be addressed by a true multidisciplinary team with the surgeon as a major stockholder in this process. All aspects of care will evolve to comprehensively consider the entire pelvis along with the entire GI tract and nervous input. This chapter reflects my crystal ball for the future care of our pelvic floor patients.

As surgeons, we all started as barber surgeons. Our tools were used to cut hair and then perform surgical procedures. As medical and surgical knowledge was advanced, we broke away from our barber heritage and concentrated on our surgical skills. Initially, surgeons were all generalists operating on the entire body. But the body is complex, and vast quantities of knowledge were deciphered for each specific region or organ system. With time, the combination of knowledge and technical demands were overwhelming for a true general surgeon, and therefore our predecessors began to focus their skills toward specific organ systems or regions. Thus, specialists were born. However, humans are curious and always striving to learn more and improve.

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Our knowledge and tools have exponentially advanced over the past 50 years, and superspecialists were needed as the problems continued to become more complex.

The pelvis is one of those extremely complex regions of the body. It is unique being a box made from the boney pelvis with a floor made of muscle. Piercing that muscular floor are the urinary and bowel systems in men and women (additionally the vagina in women). We have learned that all of these pelvic systems are interrelated. Changes or compromise in one area can lead to a problem in another area. For optimal pelvic health, all regions must work individually and together as a unit. When looking at the treatment of pelvic disorders, we are at a crossroads that is steering us toward further specialization by the current health care specialists that treat this area of the body. The multiple surgical specialists that have a stake in its treatment include general surgeons, colorectal surgeons, gynecologists, urologists, and some other branches. Each of these disciplines many times work in isolation when assessing and treating *their* compartment of the pelvis. Ironically, this contradicts the pelvis being a unit with each compartment – anterior, middle, and posterior – interrelated. Additionally at many current symposiums and educational meetings, it is promoted as being essential for all healthcare providers treating pelvic disorders to work together and treat the pelvis as one entity. However in practice, this does not seem to be a uniform reality. Therefore, the future of pelvic healthcare requires this area be treated by a multidisciplinary team (just like occurs with rectal cancer) in order to provide optimal care. This will require surgeons treating disorders currently designated as "pelvic floor problems" to think broadly and in concert with all providers that participate in treatment and care for this region of the body.

"Pelvic floor surgeons" may become true multidisciplinary surgeons. They could draw from the experience of each compartmentally trained surgeon (colorectal and general from the posterior compartment; obstetrical/gynecologists and urogynecologists from the middle compartment; and urologists and urogynecologists from the anterior compartment). This type of surgeon would be broadly trained and able to treat and operate on all aspects of the pelvis. Another option which may be more likely is that they will lead a genuine integrated multidisciplinary team with a common goal to provide expert care for the entire pelvis.

While nobody knows how we will care for these patients in the future, I have taken this opportunity to think of my "wish list" and look into my crystal ball. The following are my thoughts about idealistic future care for our pelvic floor patients.

1 The Office Setting

In the near future, surgeons will be only a part of this "pelvic team." When patients make an appointment, a specially trained nurse will contact the patient and review all symptoms, problems, previous testing, and previous treatments. Since all health care records will be computerized in the future, a patient will have a copy of all personal health records and can forward the appropriate information to this nurse. Additionally, the nurse will request that the patient log onto the computer and complete a series of questionnaires that cover routine health questions including other medical conditions, past surgeries, medications, and obstetric, social, and family history. Specific in-depth questions about bowel, bladder, and sexual function, pain, activity level, pelvic symptoms, and quality of life scores will also be part of this computerized intake process. The program will be set up so the data will automatically download onto the research system to allow all patients to be followed for research purposes. Then, the nurse can gather all previous testing and treatment results and review the questionnaires, ensure they are fully completed, and plan for possible testing that will be needed when the patient is seen. This will all occur before the patient reaches the clinic/office.

The clinic/office will be a self-contained area with friendly staff to put patients at ease since discussing problems in this area of the body can be embarrassing for patients. The décor will also promote a calm atmosphere. Literature and pamphlets available for patients to read while waiting for their appointment will focus on diet, exercise, nutrition, and strategies to promote physical and emotional health.

When the patient is seen in an exam room, all the outside information and results of the questionnaire will be available. A pelvic floor medical doctor or advanced midlevel provider (who has completed specialized training in pelvic floor pathology) will review the records and results with the patient and fill in any gaps in the information. Then, a focused physical exam will be done that includes an in-depth examination of the entire pelvis as patients may have unrecognized problems in one area which will lead to symptoms in another.

2 Testing and Planning

After the entire history and physical examination is completed, the pelvic medical specialist will order appropriate testing as needed. In the case of ambiguity, the surgeon and other team members will be consulted to review the history and physical findings and weigh in on the testing needed.

In the future, testing will also incorporate the entire pelvis (and actually body) as a whole unit. The MRI and defecating proctogram will be combined into a PETogram. This will look at defecation, voiding, and simulated sexual encounters using technology that allows the correlation of function and brain stimulation. All testing will be done in the physiological position (i.e., sitting for defecation and voiding). The scan will utilize technology more closely related to an MRI than a traditional defecating proctogram to decrease the radiation exposure. Sensors will also be placed on strategic areas in the pelvis to pinpoint muscle movement. Brain activity will be captured by a helmet that the patient wears. Afferent and efferent nerve impulses will also be captured with sensors attached to the skin over the spine and posterior tibial nerve. Sensors will also be placed in the bladder, urethra, vagina, anus, and rectum which will allow correlation when the patient is resting, asked to squeeze, and strain. Since the entire GI tract can affect bowel function (fecal incontinence, flatus production and control, and constipation), this will be also assessed by tiny nanosensors that the patient will swallow prior to the test. These temporarily attach to the mucosa in various portions of the GI tract and collect data regarding pressure, microbiome of the area, and coordination. All this will be correlated by a computer that has been programmed to incorporate these data points and assess for dysfunction. The patient will also be placed upright to simulate exercise and coughing – again looking to see how the pelvis and brain work together. While some forward thinkers hope this data will be able to be collected totally by nanosensors injected into the bloodstream or simply by moving a wand over the body, these advances in testing will not be a reality for another 50 years.

Virtual simulation assessing sexual responses and changes in pain perception will also be a feature of this new testing device. Instead of nanosensors, nanostimulators will be utilized for this aspect of testing.

Because the genetic makeup of the pelvic tissues influences their susceptibility to injury or dysfunction, tissue biopsy of the rectum, anal muscle, levator muscle, bladder, urethra, vagina, uterus, and other structures will be obtained as indicated utilizing a sharp but flexible needle about the size of a human hair. When there has been an acute injury such as during childbirth or surgery, stem cells which correct this defect or problem will be grown and injected into the appropriate tissue area to aid healing or correct genetic issues.

After all past and current records and testing are completed, patients will be discussed in the multidisciplinary management conference. This will consist of a team of health care providers including the specialist provider who did the initial history and physicians, nurses, surgeons (colorectal, gynecological, urogyncological, urological), physical therapists, radiologists, nutritionists, sexual therapists, gastroenterologists (specializing in bowel disorders), pain management doctors, social workers, psychologists, geneticists, and trainees. Individually patients will be presented and their testing reviewed as a group. Some patients will have relatively straightforward problems, and a treatment plan can be recommended. Others will be complex, and all meeting attendees may be required to provide input and recommendations for care. Detailed notes will be taken and a summary provided for each patient's chart. Then, the patients will be seen again in the clinic with the appropriate providers and the recommendations explained. Appointments will be made that complement the recommended treatment course.

3 Follow-Up

Since most treatment recommendations will continue to be initially nonsurgical in the future, the patient will participate in the treatment algorithm recommended by the group. Progress will be charted on an individual data tablet which will automatically download into the patient's chart. Patient adherence and improvement will be assessed. Repeated discussions will be undertaken in the multidisciplinary group when patients do not sufficiently improve, and the next phase of treatment will be initiated.

4 Operating Room

The operating room of the future will be quite different from what we have now. Patients will be positioned in a special device that will allow repositioning to optimize whichever area of the pelvis is being addressed. For instance, if the prone position is needed for an anorectal issue and then the patient needs to be repositioned to lithotomy, this device will allow the patient to be moved while protecting the endotracheal tube and pressure points. This will also allow positioning and easy repositioning for abdominal procedures.

When indicated, a 3-D pelvic model showing the deficient area of the pelvis will be displayed on a screen over the OR table. While operating, the surgeon will be able to manipulate the model to assist in visualization of the problem.

Since surgeons will be trained across surgical specialties in the pelvis, coordination in the OR will be much easier. Gone will be the problems of positioning for each area of the pelvis because the patient can simply be rotated. Procedures will be rehearsed and planned using the 3-D model so the movements of each surgeon will have been planned and thus there will be more efficient use of the OR time. Most surgery will be done by a hybrid laparoscopic approach that promotes better ergonomic health for the surgeon but allows for improved wrist mobility. The surgeon will be at the table and not removed to another area. Three-dimensional viewing will be possible due to new technology in eyewear the surgeon will wear. The camera will directly transmit the image to this eyewear (this will be a direct offshoot from the google glass technology). Tactile feedback will also be possible due to sensors that attach to the fingers and transmit this sensation from the tip of the instrument to the surgeon.

There will still be patients that have adhesions or other reasons that require the old-fashioned "open" approach. Since trainees will have limited experience in open surgery, fellowships in open surgery will be offered (very much like the minimally invasive fellowships of the early 2000s were needed to propel surgeons into mainstream minimally invasive approaches). Surgeons trained in the "open" approach will be more generalists and less specialized across all aspects of the pelvic floor since there will be fewer open cases versus the minimally invasive ones. By nature since these patients require an open approach, they naturally may be more complex, and the added fellowship training in areas such as adhesion dissection will be invaluable. The open and minimally invasive surgeons will therefore need to work as a team combining their skills in caring for these patients.

5 Post-op Care

The postoperative care will also be a coordinated effort. Many procedures will be same-day surgery as new medication will be given that accelerate the healing process and reduce postoperative tissue trauma and swelling. There will always be infirmed patients that require hospitalization, but with preoperative planning and optimization, these will be few. After the patient is discharged, preventative strategies will be implemented and taught or reinforced with the patient via the physical therapist, occupational therapist, nutritionist, personal trainer, and others.

6 Preventative Care

Preventative care for bowel, bladder, and sexual health will also receive more focus in the future. Starting in primary school, children will be assessed and taught the best way to eat and exercise according to their physical makeup. For instance, if their personal assessment shows a high probability they will be prone to rectal prolapse in the future, they will be counseled to avoid strenuous weightlifting or other activity which can also predispose to this problem. The individual makeup of their bacterial flora and gut mucosal mechanisms will be analyzed to recommend the optimal diet strategy to prevent many diseases including those associated with immune mechanisms such as irritable bowel syndrome.

One area that deserves special mention is childbirth injury. All women with demonstrable injury after childbirth will have immediate harvest of cells in order to produce “healing cells” made from their own genetic tissue. These cells will be grown and then within 36 h injected into the injured area to promote tissue regrowth and mitigate nonfunctional scar tissue.

7 Research

Research and hard work has gotten us to this future point in the care of patients with pelvic floor problems. It has also allowed us to implement preventative strategies starting at a young age. All patients will be part of a health database and their data reviewed so no patient is lost to follow-up. This will allow complete records and data collection.

To continue to push forward and progress, future research will be more global and include teleconferences with centers all over the globe. Prospective randomized trials will be a main focus and the ability to generate a three-dimensional model of the pelvis will assist in the preop and post-op assessment of repair. Functional improvement will be assessed with true validated questionnaires. All results, both positive and negative, will be published and available to guide future research and ideas.

8 Practical Application

I was asked to comment on a 32-year-old P2G2 woman who is actually the daughter of a surgeon (friend of mine). She was seen in her local ER due to progressive problems defecating and an inability to void. A foley was placed and 3,100 cc of urine drained from her bladder. She had not moved her bowels for 10 days. She was admitted to a gastroenterologist’s private service and an IV started. Her complete blood count, electrolytes, and urinalysis were unremarkable. She was given small quantities of an oral laxative and a urology consult obtained. There was no vaginal or digital exam done. A CT scan showed that her colon was filled with stool, and an MRI showed no spinal lesions. Two days later, she had a cystoscopy that showed a large bladder but no lesions. On hospital day 5, a gynecology consult was obtained. This woman still has not moved her bowels, and no digital exam has been done. The vaginal exam showed laxity of the anterior wall of her vagina with prolapse of what was felt to be the bladder toward the introitus, a mild rectocele, and a cyst on her cervix. Still no digital exam was done. She continues to receive oral laxatives.

At this stage (hospital day 7), her father sent me her records and asked me to comment (she remained hospitalized nearly 10,000 miles away in another continent). I was amazed that the GI doctor, gynecologist, and urologist never spoke directly to one another. She has now gone 17 days with no defecation, she states she is distended and has some cramps, she is anorexic, and still no digital exam has been done. She could not hold an enema, and therefore this treatment was abandoned. She is straight cathing as her bladder does not empty, and she is understandably very discouraged.

This is a true story about a patient with these pelvic floor problems that occurred while I was writing this chapter. It exemplifies the need for teamwork assessing pelvic floor problems. In this woman's case, at the least a thorough physical exam and collaborative discussion was needed to attempt to sort out her problem. This poor patient's situation highlights why we need to train pelvic floor doctors of the future who act and work together in the best interest of this group of patients. This *must* become a reality and move beyond the discussions which mostly occur at intellectual meetings. All healthcare providers with a stake in treating pelvic floor problems must be aware of the diagnostic power they would possess working as a group. Modern multidisciplinary centers need to actually be set up and staffed with a team of providers with an interest in collaborating and treating these patients. It is time to move beyond the dream and make this a reality.

Part I

Anatomy

Surgical Anatomy of the Colon, Rectum, and Anus

2

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Contents

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Abstract

Knowledge of the embryologic origins of the gastrointestinal tract is useful for the understanding of the anatomic relationships and surgical tissue planes used in abdominal surgery. The colon begins in the right lower quadrant where the terminal ileum enters the cecum and ends at the sacral promontory. The rectum follows the sacrum ending at the anal canal. The blood supply to the right colon comes from the superior mesenteric artery and to the left from the inferior mesenteric artery. Branches of the internal iliac artery supply the rectum. The veins and lymphatics run parallel to the arteries. Innervation comes from the parasympathetic and sympathetic fibers of the lower thoracic and lumbosacral region of the spinal cord.

1 Embryology

Knowledge of the embryologic origins of the gastrointestinal tract is useful for the understanding of the anatomic relationships and surgical tissue planes used in abdominal surgery and congenital abnormalities. The primitive foregut structures end at the second portion of the duodenum and are supplied by the celiac artery. The midgut starts at the duodenal ampulla and ends at the distal transverse colon. The small intestine, cecum, appendix, ascending colon, and proximal transverse colon are derivatives of the midgut and are

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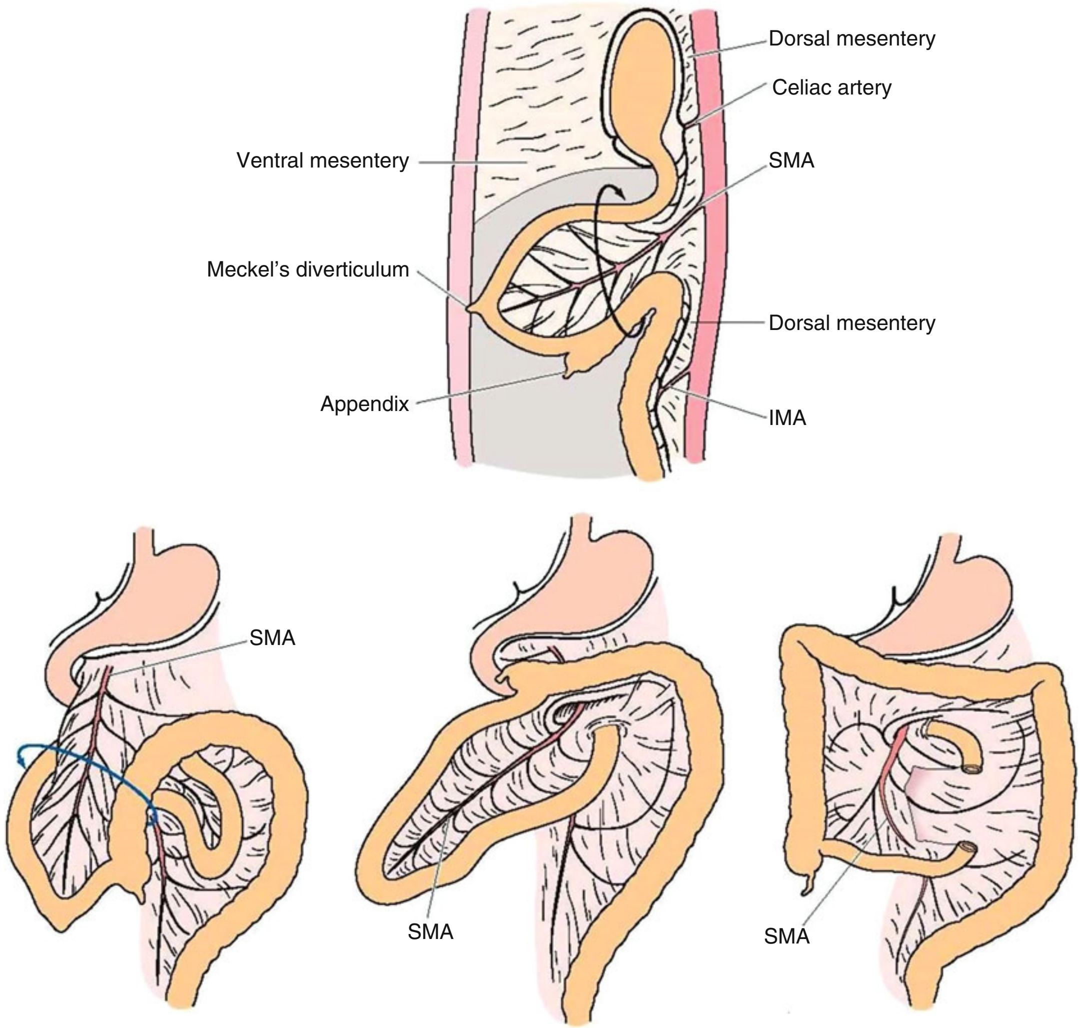

Fig. 2.1 During the sixth week in utero the midgut herniates out of the abdominal wall and rotates 90° counterclockwise. By the tenth week the midgut rotates an additional 180° counterclockwise as it returns to the abdominal cavity (Mike and Kano 2013)

supplied by the superior mesenteric artery. The hindgut begins at the distal third of the transverse colon and includes the descending colon, sigmoid colon, rectum, and the upper portion of the anal canal. The hindgut and part of the lower urogenital tract are supplied by the inferior mesenteric artery. The anal canal is formed at the end of the rectum where the endodermal tissues fuse with the ectoderm and is supplied by branches of the internal pudendal artery (Rowe et al. 1974; Dujovny et al. 2004). The dentate line is the approximate location of the transition from endoderm to ectoderm at the junction of the upper two thirds of the anal canal with the lower one third. The veins and lymphatics follow the same embryologic divisions as the arterial supply.

These structures develop during the fourth week in utero (Fig. 2.1). During the sixth week, the midgut lengthens, herniates out of the abdominal cavity, and rotates 90° counterclockwise around the superior mesenteric artery. The cloaca is the caudal part of the hindgut and begins to separate into two sections at the end of the sixth week of gestation. The ventral portion develops into the urogenital sinus and the dorsal portion develops into the anorectum (Rowe et al. 1974). This separation occurs as a result of either a cranially oriented septum growing down to reach the cloacal membrane or from lateral folds fusing in the middle (Stephens 1961). By the tenth week the midgut rotates an additional 180° counterclockwise as it returns to the abdominal cavity. The cecum descends to the right iliac fossa where it becomes fixed. The base of the small bowel mesentery is fixed from the ligament of Treitz in the left upper quadrant to the ileocecal valve in the right lower quadrant (Moore 1988).

2 Anatomy

The colon extends from the ileocecal valve to the proximal rectum and is approximately 3–5 f. in length. The terminal ileum enters the cecum on its posteromedial border at the ileocecal valve. The cecum is a large blind pouch approximately 7.5–8.5 cm in diameter which projects from the ascending colon below the ileocecal valve. It is covered by peritoneum and has a narrow diverticulum (the appendix) located in the lower portion. The ascending colon is approximately 20–25 cm in length and is retroperitoneal. The ascending colon ends at the hepatic flexure just under the liver. The transverse colon extends from the hepatic flexure to the splenic flexure and is attached to the gastrocolic ligament. The omentum is attached to the transverse colon on its anterior superior edge. The descending colon is approximately 10–15 cm in length. It is a retroperitoneal structure which begins at the splenic flexure and ends at the sigmoid colon. The sigmoid colon is a redundant loop of colon covered with peritoneum which can be variable in length and location.

The colon progressively diminishes in size from the cecum to the sigmoid colon. The sigmoid colon is approximately 2.5 cm in diameter and is the narrowest portion of the colon. This size discrepancy accounts for the frequent finding that cecal tumors can grow to be large and bulky prior to the onset of symptoms while sigmoid tumors are symptomatic at smaller sizes. In addition, tension on the bowel wall is directly proportional to the diameter of the bowel as explained by Laplace's Law ($T = PR$; T, tension in the wall of the bowel; P, internal pressure; R, radius of the bowel). Since the cecum has the largest diameter it is usually the first part of the bowel to rupture due to distal obstruction.

The ascending colon, descending colon, rectum, and posterior surface of the hepatic and splenic flexures are fixed retroperitoneal structures. The cecum, transverse, and sigmoid colon are intraperitoneal and are prone to volvulus because of their location and relative lack of fixation.

The colonic wall consists of mucosa, submucosa, inner circular muscle, outer longitudinal muscle, and serosa. The longitudinal muscle is an incomplete layer and is seen as three bands of muscle called taenia coli. The taenia coli are located 120° apart around the circumference of the colon. They converge proximally at the appendix and disappear as distinct bands at the level of the sacral promontory. Haustra coli are sacculations between the taenia and are separated by crescent shaped folds called plicae semilunares. Appendices epiploicae are fatty appendages covered by peritoneum and have no anatomical or pathologic significance.

The rectum is the last segment of the large intestine and is approximately 12–15 cm in length (Fig. 2.2). It begins at the rectosigmoid junction at the level of the sacral promontory (S3), follows the curve of the sacrum, and ends at the anal canal. Intra-abdominally the rectosigmoid junction is identified as the point where the discrete taenia coli of the sigmoid splay out to form a complete layer of longitudinal muscle in the upper rectum. The rectal wall consists of mucosal, submucosal, inner circular, and outer longitudinal muscular layers. There is no serosal layer in the rectum nor are there haustra which are present in the proximal colon. Folds of the rectal wall are seen endoscopically and are called Valves of Houston. They serve no physiologic function but are useful as landmarks for endoscopy. There are typically three valves and their location within the rectum can be variable. The upper valve is usually found on the left side of the rectum 8–16 cm from the

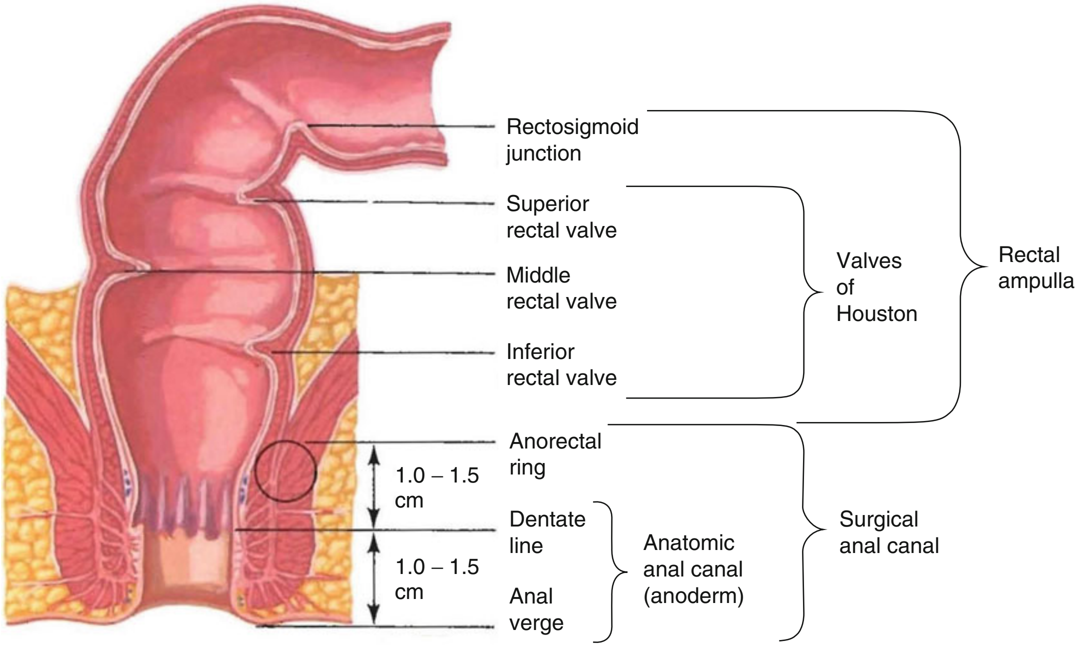

Fig. 2.2 The rectum begins at the rectosigmoid junction and ends at the anal canal (Fry and Kodner 1986)

anal verge. The middle valve is typically on the right side of the rectum 7–12 cm and is at the level of the peritoneal reflection. The lower valve is located in the left side 5–10 cm from the anal verge (Shafik et al. 2001).

The mesorectum is a fascial structure which surrounds the perirectal fat, blood vessels, and lymphatics of the rectum. Posterior to the mesorectum is the presacral fascia which covers the presacral venous plexus. The presacral fascial layer fuses with the posterior mesorectal fascia forming the rectosacral fascia or Waldeyer's fascia below S2-S4. Preservation of these layers is vital to proper surgical technique for proctectomies done for rectal cancer. The peritoneum extends along the anterior wall of the rectum covering one third to two thirds of the anterior rectal wall. Denonvilliers' fascia is just caudal to the peritoneal reflection. It separates the mesorectum from the prostate and seminal vesicles in men and the vagina in women (Lindsey et al. 2000; Chapuis et al. 2002; Lin et al. 2011).

The anal canal is the terminal structure of the gastrointestinal tract (Fig. 2.2). The surgical anal canal extends from the proximal internal anal sphincter to the anal verge and is approximately 4 cm in length. It is surrounded by the internal anal sphincter and the external anal sphincter. The surgical anal canal extends from the anal verge to the anorectal ring and is more of a functional description than anatomic. The anatomic anal canal extends from the dentate line to the anal verge. The dentate line is an important landmark and is a true mucocutaneous junction located 1–1.5 cm above the anal verge. A 6–12 mm transitional zone exists above the dentate line where the squamous epithelium of the anoderm becomes cuboidal and then columnar epithelium. Columns of Morgagni are 8–14 mucosal folds located just above the dentate line and surround anal crypts. Small rudimentary glands open into some of the crypts. These glands go through the internal sphincter into the intersphincteric groove but do not penetrate the external sphincter. Knowledge of this anatomy is useful in describing the origins of perianal abscesses. The dentate line is a transition point for the blood supply and innervation of the anal canal. The anus is innervated by sympathetic and parasympathetic nerves with no somatic pain fibers proximal to the dentate line. Distal to the dentate line there is somatic innervation which is particularly useful to explain the pain sensation for excisional hemorrhoidectomies and the relative lack of pain for elastic ligation done above the dentate line. The anal verge is the junction between anal and perianal skin. Anal epithelium (anoderm) lacks hair follicles, sebaceous glands, or sweat glands. The anal margin is identified at the border of the anal verge where hair follicles and keratinized epithelium begin.

The anorectal ring is 1–1.5 cm above the dentate line and is the palpable upper border of the anal sphincter complex. The internal anal sphincter, external anal sphincter, and levator ani make up the anal sphincter complex and are vital for maintenance of fecal continence. The internal anal sphincter is a continuation of the inner circular layer of the rectum. It extends beyond the external anal sphincter approximately 1 cm from the dentate line. It is a smooth muscle which is contracted at rest contributing to anal tone. The sympathetic innervation (T11-L2) and parasympathetic innervation (S2-S4) of the internal anal sphincter cause the internal anal sphincter to be contracted at rest (Freckner and Ihre 1976). The external anal sphincter is skeletal muscle innervated by branches of the pudendal nerve and the perineal branch of S4. It too is contracted at rest but it is a voluntary muscle which can be contracted further if necessary. The external anal sphincter wraps around the internal anal sphincter and is a continuation of the levator ani. The levator ani is actually three muscles (the puborectalis, pubococcygeus, and iliococcygeus) which make up the pelvic floor. The puborectalis is a continuation of the external anal sphincter and it creates a U-shaped sling around the anal canal. The muscle is not contiguous in the anterior quadrant but is fixed anteriorly to the pubis. Contraction of the puborectalis pulls the rectum anteriorly and contributes to fecal continence.

3 Arterial Supply

The superior mesenteric artery arises from the ventral surface of the aorta below the celiac axis, passes behind the pancreas, and crosses in front of the third portion of the duodenum (Fig. 2.3). The SMA is the central axis around which the midgut rotated during embryogenesis and gives off right and left branches which form the blood supply to the midgut. The right branches include the inferior pancreaticoduodenal artery, the middle colic artery, the right colic artery, and the ileocolic artery. The left branches are the jejunal and ileal branches and number 15–20 (Sagar and Pemberton 1997). The middle colic artery is the

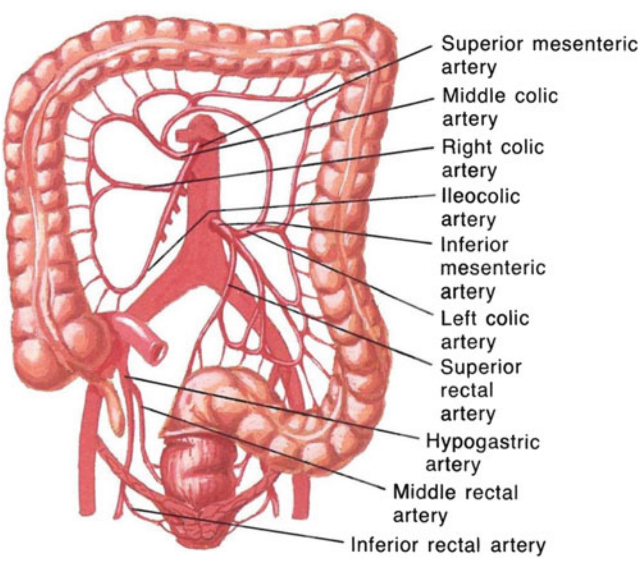

Fig. 2.3 Arterial supply of the *right* (ascending) colon via branches of superior mesenteric artery; *left* (descending) colon and rectum via branches of interior mesenteric artery. Distal rectum supplied by branches from hypogastric artery (Fry et al. 1989)

first branch of the SMA arising just below the uncinate process of the pancreas. It supplies the transverse colon and communicates with branches of the IMA. The middle colic artery typically branches into a right and left branch in the center of the transverse colon. The left branch forms collaterals with the left colic artery via the marginal artery. The right colic artery supplies the ascending colon but is the most variable of the colic arteries and is present in only 10–40 % of cadavers (Michels et al. 1965; Wenk 1995; Garcia-Ruiz et al. 1996). It may arise directly from the SMA, the middle colic, or the ileocolic artery. The ileocolic artery is the terminal branch of the SMA and supplies blood to the terminal ileum, cecum, and ascending colon.

The inferior mesenteric artery arises from the infrarenal aorta and supplies the descending colon, sigmoid colon, and upper rectum via its left colic, sigmoidal, and superior rectal branches (Fig. 2.3). The left colic artery is the first branch of the IMA and runs upward reaching the splenic flexure where it forms collaterals with the left branch of the middle colic artery. The sigmoid arteries can arise from the left colic artery or the descending branch of the IMA (Goligher 1967). The terminal branch of the inferior mesenteric artery becomes the superior rectal artery which descends in the sigmoid mesocolon and bifurcates at the level of S3. Branches of the superior rectal artery supply the upper and middle rectum. Ligation above the origin of the left colic artery has been considered “high ligation” and below this level “low ligation” (Surtees et al. 1990). It is probably more accurate to describe the artery proximal to the left colic artery as the IMA and caudal as the superior hemorrhoidal artery.

Middle rectal arteries arise from the anterior divisions of the internal iliac arteries and anastomose with branches of the superior and inferior hemorrhoidal arteries in the region of the lateral ligaments of the rectum. These arteries supply the lower two thirds of the rectum. The presence and location of these vessels is variable and may be absent (Goligher 1967; Lin et al. 2011).

Inferior rectal arteries are branches of the internal iliac arteries via the pudendal branches. Anastomosis exists between the middle and superior rectal arteries; there are no anastomoses with the inferior rectal arteries. The middle and inferior rectal arteries provide the blood supply to the rectal stump after division of the superior hemorrhoidal artery during sphincter saving procedures for rectal carcinoma (Boxall et al. 1963; Goligher 1967). Preservation of the middle rectal arteries is necessary to maintain viability of the rectal stump after high ligation of the inferior mesenteric artery (Boxall et al. 1963). The middle sacral artery arises just before the aortic bifurcation. It descends in the midline along the vertebrae, sacrum, and coccyx and provides small branches to the posterior surface of the rectum.

Collaterals exist between the superior and inferior mesenteric arteries in the region of the splenic flexure (Sonneland et al. 1958; Fig. 2.4). The arcades of the ileocolic, right, middle, and left colic arteries are peripherally connected by the marginal artery of Drummond which runs along the mesenteric border of the colon and provides the vasa recta to the colon (Kornblith et al. 1992). The marginal artery runs 2–3 cm from the edge of the colon and is better defined in the left colon than in the right (Krupski et al. 1997). The arc of Riolan is a tortuous, inconstant vessel which is located more centrally within the colonic mesentery. It forms a communication between the left colic branch of the inferior mesenteric artery and the middle colic branch of the superior mesenteric artery. It is frequently referred to as the meandering mesenteric artery and is best visualized when there is an occlusion of either the inferior or superior mesenteric artery (Michels et al. 1965). The presence of large marginal/meandering arteries may indicate occlusion or stenosis of the SMA or IMA.

There are two watershed areas in the colon where collateral blood flow may be variable: Grifith’s point at the splenic flexure and Sudek’s point near the rectosigmoid junction. (Goh 1997) Grifith’s point is where the branches of the middle colic and the left colic meet and Sudek’s point is where the sigmoid branch and superior hemorrhoidal artery meet. The importance of these sites is that they can be locations of ischemic colitis in low flow situations.

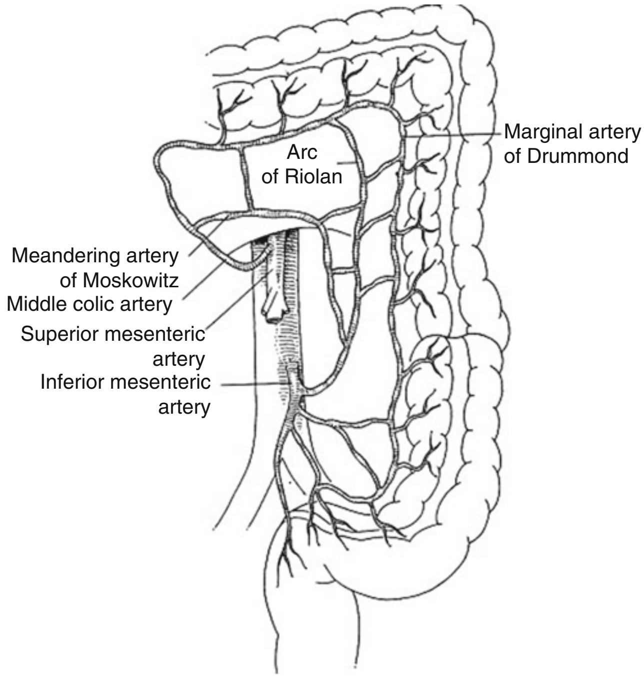

Fig. 2.4 Collaterals exist between the superior and inferior mesenteric arteries via the marginal artery of Drummond and the Arc of Riolan (Lin and Chaikof 2000)

4 Venous Drainage

The veins draining the colon follow the same course as the corresponding arteries except for the inferior mesenteric vein (Fig. 2.5). The inferior mesenteric vein drains the descending colon, sigmoid colon, and proximal rectum and runs in a retroperitoneal location to the left of the ligament of Treitz where it enters the splenic vein. The superior mesenteric vein drains the cecum, ascending colon, and transverse colon,and joins the splenic vein to form the portal vein. The veins from the right colon drain into the SMV which lies to the right of the SMA. The SMV joins the splenic vein to form the portal vein behind the neck of the pancreas (Goligher 1967).

The venous drainage of the rectum enters the portal or systemic (caval) system. The upper and middle rectum is drained by the superior rectal vein which enters the portal system via the inferior mesenteric vein. The lower rectum and upper anal canal are drained by the middle rectal vein which empties into the internal iliac veins and then into the caval system. The inferior rectal veins drain the lower anal canal and empty into the pudendal veins which empty into the caval system via the internal iliac veins. Rectal tumors can metastasize through venous channels into either the portal or systemic venous systems. This venous anatomy may explain why some patients with low rectal

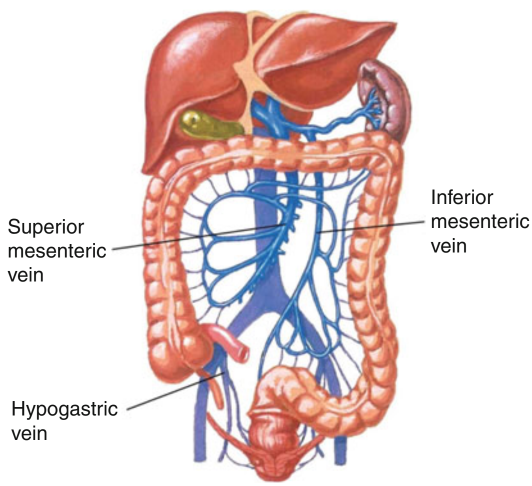

Fig. 2.5 Venous drainage parallels arterial supply. *Right* colon drains into portal system via superior mesenteric vein; *left* colon and rectum via inferior mesenteric vein. Distal rectum drains via hypogastric vein (Fry et al. 1989)

carcinomas can develop pulmonary metastases without hepatic metastases.

There are three submucosal internal hemorrhoidal complexes located above the dentate line. Hemorrhoidal tissue receives its blood supply from the superior, middle, and inferior rectal arteries. The left lateral, right posterolateral, and right anterolateral internal hemorrhoids drain into the superior rectal vein. The external hemorrhoids are located below the dentate line and drain into the pudendal veins. There is communication between the internal and external plexuses and mixed internal/external hemorrhoids will result if these communications become engorged.

5 Lymphatic Drainage

Colonic mucosa has rich vascular plexuses but no lymphatics. Lymphatic capillaries encircle the colon in the submucosal and the muscularis mucosal layers. This segmental architecture limits longitudinal intramural extension of tumors and circumferential extension results in annular lesions. Lymphatic vessels follow the blood supply of the colon (Fig. 2.6).

Lymph nodes are located on the bowel wall (epicolic), along the inner margin of the bowel (paracolic), around the named mesenteric arteries (intermediate), and along the origin of the superior and inferior arteries (main) (Goligher 1967). Lymph from the upper and middle rectum drains into the inferior mesenteric nodes. The lower rectal lymphatics follow the superior rectal artery and enter the inferior mesenteric nodes. Lymph from the lower rectum can also flow laterally along the middle and inferior rectal arteries, posteriorly along the middle sacral artery, or anteriorly through channels in the rectovesical or rectovaginal septum (Block and Enquist 1961). These channels drain to the iliac nodes and subsequently to periaortic lymph nodes.

Lymphatics from the anal canal above the dentate line drain via the superior rectal lymphatics to the inferior mesenteric lymph nodes or laterally to the internal iliac lymph nodes. Below the dentate line, the lymphatic drainage does not parallel blood vessels. The lymphatics below the dentate line drain primarily to the superficial inguinal nodes and then to the external iliac and lumbar trunks. Lymphatics from the anal canal can drain to the inferior or superior rectal lymph nodes as well (Block and Enquist 1961; Wenk 1995).

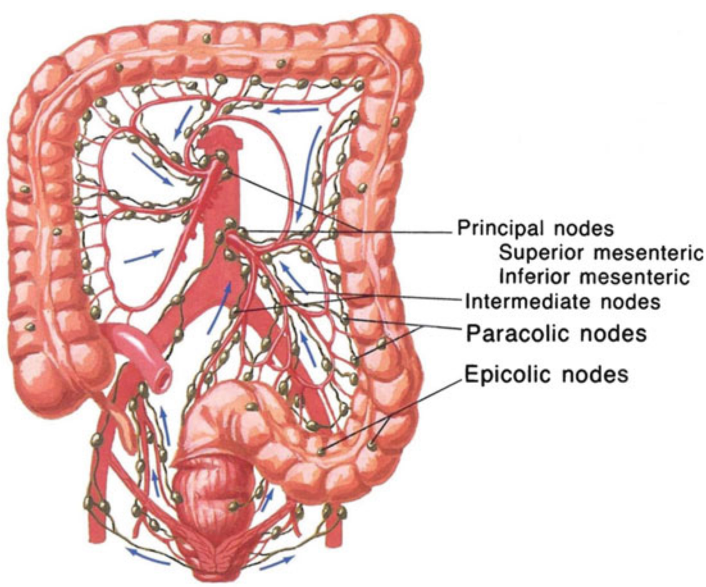

Fig. 2.6 Lymphatic drainage of colon centripetal. *Right* colon drains to superior mesenteric nodes, while *left* colon drains to inferior mesenteric nodes. Rectal drainage primarily via nodes along superior rectal artery. Distal rectal drainage may go to lateral pelvic wall and inguinal nodes (Fry et al. 1989)

6 Nerve Supply

The sympathetic and parasympathetic nerves to the colon follow the course of the blood vessels. Sympathetic nerves inhibit and parasympathetic nerves stimulate peristalsis. The foregut and midgut parasympathetic fibers originate from the vagus and the sympathetic fibers come from the lower six thoracic vertebrae (Longo et al. 1989). Sympathetic fibers to the right colon travel in the thoracic splanchnic nerves to the celiac and then to the superior mesenteric plexus. The hindgut is innervated by parasympathetic fibers from the sacral plexus and sympathetic fibers from the lumbosacral column. Sympathetic supply to the left colon and rectum originates in the first three lumbar segments. These nerves join the pre-aortic plexus, and become the inferior mesenteric plexus below the bifurcation of the aorta. The parasympathetic nerves to the left colon ascend from the pelvis, pass through the sigmoid mesocolon, and spread out toward the sigmoid and the descending colon.

The rectum is innervated by both sympathetic and parasympathetic nerves (Church et al. 1987; (Figs. 2.7, and 2.8). Sympathetic nerves from thoracolumbar segments unite below the inferior mesenteric artery forming the inferior mesenteric plexus and descend to the superior hypogastric plexus below the aortic bifurcation. The nerves bifurcate and descend in the pelvis as the hypogastric nerves supplying sympathetic innervation to the lower rectum, bladder, and sexual organs in both men and women. Sympathetic nerves stimulate the rectum and anal canal causing relaxation.

Parasympathetic fibers from the second, third, and fourth sacral roots (the nervi erigentes) unite with the hypogastric nerves anterior and lateral to the rectum forming the pelvic plexus (Church et al. 1987; Fig. 2.8). Parasympathetic stimulation of the rectum and anal canal causes relaxation of these muscles. The periprostatic plexus arises from the pelvic plexus. Mixed fibers from these plexuses innervate the rectum, internal anal sphincter, prostate, bladder, and penis. The pudendal nerve (S2, 3, 4) mediates sensory stimuli from the penis and clitoris via the dorsal nerve. Damage to the periprostatic plexus may occur during dissection of the rectum. Injury to the pelvic autonomic nerves may result in bladder dysfunction, impotence, or both.

Parasympthetic innervation (S2, 3, 4) of the internal anal sphincter causes the muscle to relax while sympathetic innervation (T11-L1) causes

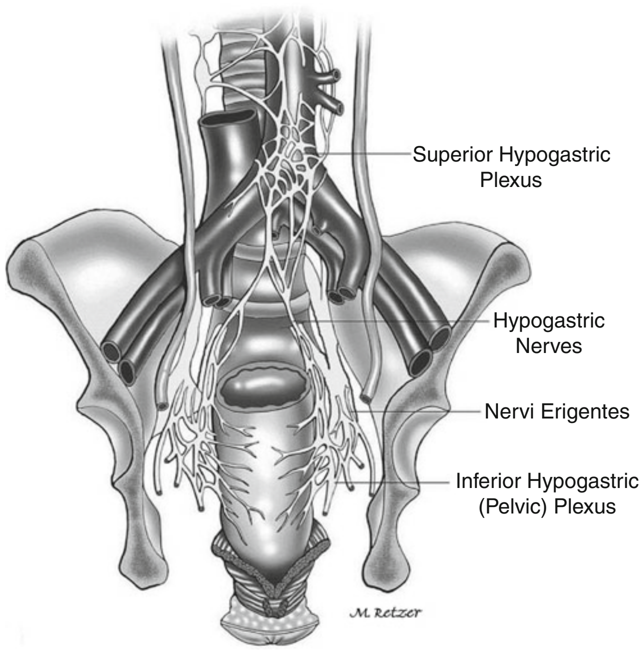

Fig. 2.7 Sympathetic innervation of the rectum (Jorge and Habr-Gama 2007)

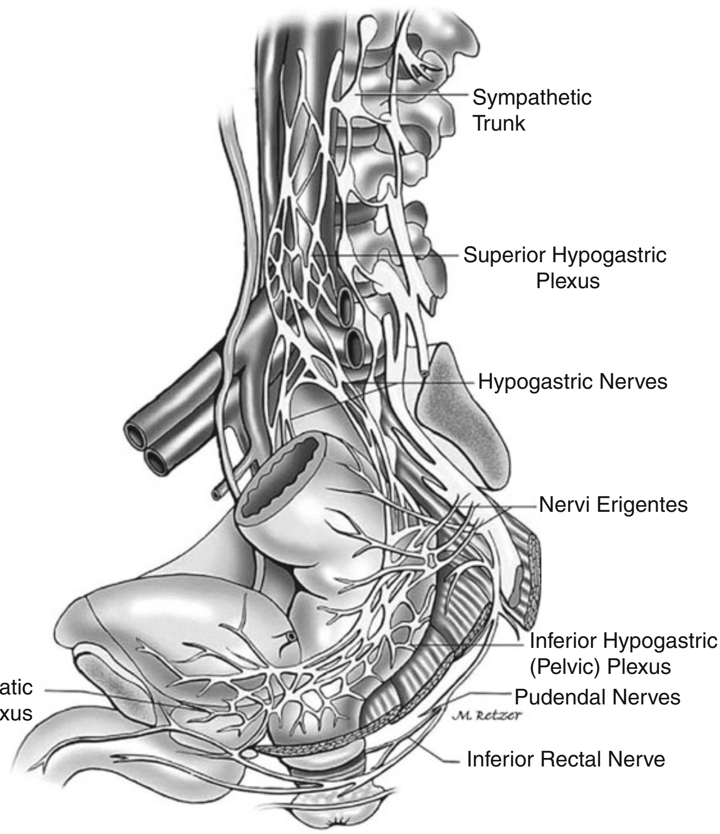

Fig. 2.8 Sympathetic and parasympathetic innervation of the rectum and anal canal (Jorge and Habr-Gama 2007)

contraction. It is made up of slow twitch muscles fibers and is fatigue resistant. The external anal sphincter is a voluntary muscle innervated by the pudendal nerve (S2, 3, 4), it is composed of slow and fast twitch fibers, and is a fatiguable muscle.

The pudendal nerve is a mixed nerve supplying both sensory and motor function. Below the dentate line, cutaneous sensations of temperature, pain, and touch are conveyed by afferent fibers of the inferior rectal and perineal branches of the pudendal nerve. Numerous free nerve endings make this area very sensitive to these modalities (Duthie and Gairns 1960). Above the dentate line, a poorly defined dull sensation, experienced when the mucosa is pinched or when internal hemorrhoids are ligated, is probably mediated by parasympathetic fibers.

There are several areas of potential neurogenic injury during abdominal surgery. The superior hypogastric plexus can be injured in the preaortic region during high ligation of the inferior mesenteric artery. The hypogastric nerves can be injured at the pelvic inlet. The inferior hypogastric plexus can be damaged along the pelvic sidewall during division of the lateral stalks. Dissection along Denonvillers' fascia and rectoprostatic space can injure the nerves during low rectal resections.

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Topographic Anatomy of the Colon and Rectum: Tips and Tricks in Laparoscopic and Robotic Surgery

3

Alessio Vinci, Mark H. Hanna, and Alessio Pigazzi

Contents

Abstract

Operative laparoscopy has progressed rapidly in recent years, and this alternative to the conventional approach for abdominal surgery has allowed the identification of new planes, spaces, and anatomic landmarks as a result of the artificial rupture of the "anatomical continuum" under endoscopic vision (Jimenez and Noguera Aguilar 2009). Magnified laparoscopic views and the ability to deeply explore anatomic features better demonstrate the basic anatomy. Therefore, even as laparoscopy requires a more profound knowledge of basic anatomy, it enhances our understanding of anatomy. Furthermore, laparoscopy requires a distinct vision and manipulation of the connective tissue, resulting in a different perspective on surgical anatomy than open surgery.

The laparoscopic surgeon must have a deep knowledge of basic anatomy to explore new surgical approaches better suited to the visual characteristics and handling of minimally invasive technology. In this respect, the concepts of basic anatomy are not modified, but horizons for the interpretation of anatomic structures are significantly expanded.

It has therefore been necessary to rediscover this anatomy to adapt basic anatomic education to the new and more demanding dynamics of surgical practice.

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1 Introduction

Minimally invasive colorectal surgery has gained increasing popularity and is widely accepted in the surgical community as a superior alternative for procedures involving benign and malignant colorectal diseases.

Minimally invasive surgery (MIS) offers several advantages over laparotomy. Pelvic and abdominal anatomy is magnified, allowing more precise identification and treatment of the disease adjacent to vital organs, blood vessels, and nerve structures. Other benefits of MIS include minimized bleeding from small vessels afforded by pneumoperitoneum, the elimination of large abdominal incision, less adhesion formation, early ambulation, and faster recovery and shorter hospital stay.

Most surgeons are familiar with the anatomy involved with colon and rectal resection. However, there is a different anatomical view with laparoscopic (LS) and robotic surgery (RS), and although the anatomy does not change, the view through which the anatomy is seen does (Bokey et al. 1997; Morris et al. 2010).

Since the first case of laparoscopic colon resection (Fowler and Jacobs 1991), the surgical technique has developed rapidly, and the feasibility of MIS in colorectal cases has been demonstrated.

However, the operative range of view for the colon and rectum is extensive and spread over the four abdominal quadrants. The blood supply and adjacent tissues of the colon and rectum are complicated and often variable. Moreover, operations of the colon and rectum involve not only simple resection but also reconstruction of the intestinal tract. These factors result in technical difficulties of MIS colorectal surgery (Feliciotti et al. 2003; Patankar et al. 2003; Minhua 2002).

The risk of surgical complications is linearly correlated with surgical technique, instrumentation, and knowledge of relevant anatomy (Rangel and Escobar 2005). By mastering the anatomical features of MIS colorectal surgery, operative mistakes and complications can be reduced, with no compromises in terms of oncological results when compared to open surgery.

The aim of this chapter is to discuss several critical areas related to the surgical anatomy of the colon and rectum, with special attention to the laparoscopic and robotic approach. Several colorectal surgical procedures will be presented offering an in-depth view of the anatomical details and the topographical correlations with the MIS approach.

2 Right Colectomy (RC)

2.1 Indications

The most common indication for RC is right-sided colon cancer. Nevertheless, patients with complete obstruction caused by the cancer, extensive invasion of adjacent organs, and cancer larger than 10 cm should only be approached laparoscopically in the hands of highly skilled operators.

Less common indications are benign tumors not resectable by an endoscopic procedure and inflammatory bowel disease strictures.

2.2 Laparoscopic Approach

2.2.1 Procedure and Anatomical Landmarks

The laparoscopic dissection of the right colon is generally thought to be more straightforward than the transverse colon, left colon, or rectum.

The patient is placed in the supine or lithotomy position. The surgeon stands on the patient's left, watching the monitor on the other side.

There are two general approaches, one where the colon is mobilized from its lateral attachment first (the lateral-to-medial approach) and one where the vascular pedicles are initially ligated, followed by colonic mobilization (the medial-to-lateral approach).

Medial-to-Lateral Technique

While both techniques accomplish the same dissection, a medial-to-lateral approach has the conceptual advantage of early ligation of the vascular

Fig. 3.1 Identification of the ileocolic pedicle

pedicles thus preventing any escape of tumor cells into the mesenteric circulation during tumor mobilization. The medial-to-lateral approach also allows preservation of the lateral colonic ligament till the end of the mobilization, which keeps the right colon fixed in place and thus limits the need for manipulation.

Supporting the patient on the operating table is important to prevent patient slippage and allow for tilting of patient during procedures. The patient is placed in Trendelenburg with the right side up to remove the small bowel from the pelvis and right lower quadrant.

The operation begins with the surgeon standing on the patient's left side to identify the ileocolic pedicle root and to mark its lower border. The terminal ileum is grasped with the left hand and pulled toward the anterior abdominal wall. Adequate traction of the mesocolon is needed to facilitate the mobilization of the ileocolic vessels (IC) from the retroperitoneal fascia leading onto the duodenum (Fig. 3.1).

According to the Okuda and Nobuhiko classification (Milsom et al. 2006a), the vascular anatomy of this area may be classified into two types (type A and type B). This classification addresses the need to achieve a complete lymphadenectomy around the origin of IC for advanced right colon cancer and is based on the position of the ileocolic pedicle with respect to the superior mesenteric vein.

– In type A, the ileocolic artery is found anteriorly to the superior mesenteric vein (SMV). After mobilization of the ileocolic pedicle from the duodenum, the dissection of the ventral side of the superior mesenteric vein leads to the dissection of the origin of the ileocolic artery.
– In type B, the ileocolic artery is behind the SMV. The dissection of the ventral side of the superior mesenteric vein leads to a complete dissection of the root of the middle colic artery and vein.

The right colic vessels are then exposed at the anterior part of the head of the pancreas, respecting Henle's gastrocolic venous trunk. If an accessory right colic vein is found, it is clipped and divided. If it is difficult to confirm the presence of this variant, this vein may be easily detected later during the mobilization of right flexure.

The patient is now placed in steep Trendelenburg position with the right side elevated to move the small intestine toward the right upper quadrant. The peritoneum is incised along the base of the ileal mesentery upward to the duodenum taking care of the right ureter and gonadal vessels. A wide window is made in the peritoneum inferior to the ileocolic pedicle as the retroperitoneal structures are gently swept away in a posterior direction. The pedicle should be isolated adequately and circumferentially to allow for easy vessel division. The surgeon should clearly identify the duodenum and gonadal vessels to avoid injury (Fig. 3.2). Identification of the right ureter is not always necessary in laparoscopic right colectomy. The division of the ileocolic pedicle can be performed using vessel-sealing energy devices, staplers, or clips per the surgeon's preference. The level of division of IC vessels will depend on the surgical indication. For malignancy, this pedicle should be proximally ligated so as to maximize the lymph node harvest (high ligation). In benign indications such as Crohn's disease where the mesentery may be thickened, the vessel is divided where it is soft.

A medial-to-lateral mobilization of the ileocecal region and right mesocolon is achieved.

Particular caution must be taken during the right flexure mobilization to avoid bleeding

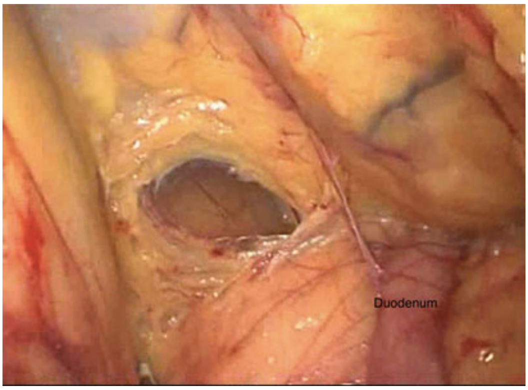

Fig. 3.2 Retromesenteric dissection

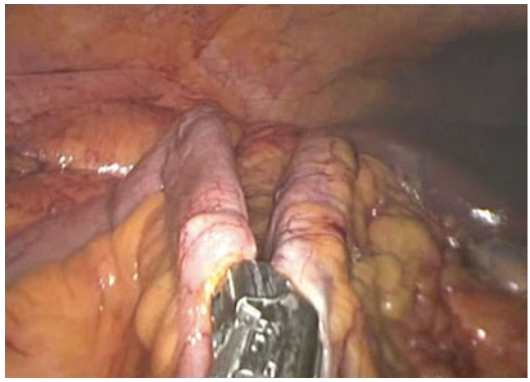

Fig. 3.3 Intracorporeal anastomosis

especially from around Henle's trunk. If the accessory right colic vein was previously difficult to detect, at this time it can be easily identified and divided. Accessory right colic vein and the right branches of middle colic vessels are clipped and divided.

Finally, the right flexure and right colon are completely freed laterally, which completes the mobilization of the entire right colon.

Once the entire right colon is mobilized, it is withdrawn through an enlargement of the umbilical port site or through a suprapubic incision. Prior to making the incision, however, the surgeon must ensure adequate reach of the transverse colon to the proposed incision site; otherwise, the surgeon risks an unnecessarily difficult anastomosis or undue tension and tearing of the middle colic vessels. A wound protector is placed to decrease the risk of extraction site recurrence and wound infection. The prepared colon is extracted. Ileal and colic resection is carried out.

The anastomoses are accomplished extracorporeally or intracorporeally (Fig. 3.3) with care to maintain the proper orientation of the duodenum and transverse colon, and avoid twisting of the mesentery.

The anastomosis is accomplished by aligning the terminal ileum and the transverse colon in an isoperistaltic fashion, then creating an enterotomy and a colotomy and firing a single 60 mm linear stapler cartridge to create the side-to-side anastomosis. Alternatively, a hand-sewn suture may be performed. The enterotomy can then be closed in a running fashion using absorbable sutures.

The bowel is put back into the peritoneal cavity in the case of extracorporeal anastomosis. The wound is irrigated and closed.

The abdominal cavity is reinflated. The ileocolic anastomosis is inspected and the proper positioning of the small bowel checked. The peritoneal cavity is irrigated with saline and checked for bleeding. The mesenteric opening does not usually require closure after a right hemicolectomy as the defect is large and it is uncommon to develop a mesenteric internal hernia. If concern arises for small bowel herniation into the mesenteric defect during the final laparoscopic inspection, the defect can be closed at the surgeon's discretion. A drain is left in the right parietal fossa per the surgeon's preference.

Lateral-to-Medial Technique

The dissection is begun either from the area inferior to the cecum and proceeds superiorly or begins at the hepatic flexure and proceeds inferiorly. Most surgeons prefer the cecum as a starting point; the dissection is started by putting appropriate tension on the cecum allowing an incision of the fascia of Toldt to be performed (Fig. 3.4). As the cecum is rotated medially, the right ureter is usually identified as it crosses the iliac artery bifurcation. The duodenum is identified medially as the dissection proceeds superiorly. The location of the ureter and the duodenum should be rechecked repeatedly throughout the procedure. The dissection then follows the cecum and hepatic flexure mobilization up to the identification of the

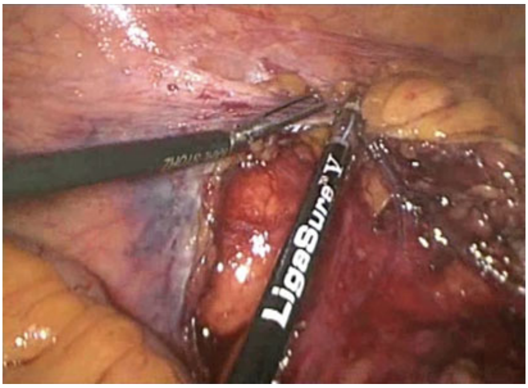

Fig. 3.4 Laparoscopic Toldt's fascia incision

ileocolic and right colic vessels. If clearly identifiable, those vessels may be dissected and divided with vascular staplers. The operation is now performed similarly to the medial-to-lateral technique.

2.3 Robotic Approach

After diagnostic laparoscopy confirming the feasibility of the colon resection, the patient is placed in the lithotomy position, and the robot is brought from the right side of the patient and docked onto the ports. Both medial-to-lateral and lateral-to-medial approaches can be performed following the steps underlined above for the laparoscopic procedure.

Although a complete understanding of the vascular anatomy is still required, the tridimensional vision and endowrist instruments offer a greater precision during the vascular dissection.

The dissection of the ileocolic pedicle as well as the identification of right gonadal vessels and right ureter may be particularly challenging, especially in obese patients (Witkiewicz et al. 2013).

Once the bowel is fully mobilized, it can be extracted via a minilaparotomy for extracorporeal anastomosis. However, the robotic system enables the surgeon to create an intracorporeal anastomosis more easily and extract the specimen via a Pfannenstiel incision.

The procedure allows a lower colon mobilization with fewer mesentery exteriorization complications such as bleeding and wound infections. A smaller incision is needed to retrieve the specimen with a better cosmetic result and reduced postoperative incisional hernia incidence (Hellan et al. 2009a; Lee et al. 2013a; Pigazzi et al. 2006).

2.4 Tips and Tricks for Right Colectomy

- The omental detachment must be begun in the medial part of the transverse colon to facilitate the dissection and should be done in all cancer cases.
- The colon may not be properly extracted without prior vessel control. Therefore, the vessels should preferentially be divided intracorporeally
- The intracorporeal anastomosis has shown many advantages, especially if performed robotically: ease creation of anastomosis in morbidly obese patients (Raftopoulos et al. 2006), flexibility in location of extraction sites, fewer mesentery exteriorization complications, and avoidance of accidental mesenteric twists (Lee et al. 2013b; Hellan et al. 2009b).

3 Left Colectomy (LC) and Low Anterior Resection

3.1 Indications

Minimally invasive left colon resection is indicated for both benign (Crohn's disease, diverticulitis, polyp unresectable by endoscopy) and malignant (colon cancer) etiologies.

3.2 Laparoscopic Approach

The patient is placed in the modified lithotomy position with carefully padded Allen stirrups. The legs are oriented so that the toes, knees, and shoulders are all in line. The knees should be slightly flexed and thighs lowered parallel to the bed to allow for a maximum range of motion of the laparoscopic instruments. Positioning of the patient in the operating room should include tucking of both arms by the patient's side to allow full access to the sides of the patient. The patient needs to be not only carefully padded to avoid any pressure injuries but also carefully secured to the bed to allow extreme positioning changes during the operation. Care should be taken to confirm that the patient's perineum is low enough off the edge of the table to allow for ~5 cm of exposure needed to allow easy passage of the circular EEA stapler.

The abdomen is then prepped and draped in the usual standard fashion with care taken to position the sterile towels along the anterior axillary line laterally, up to the xiphoid superiorly, and down to the pubis to allow for maximal exposure. The perineum is prepped if transanal extraction or a hand-sewn anastomosis is anticipated.

Note that the trocar positions are shifted down when a low anterior resection is performed.

An adequate exposure must provide an excellent view of the sacral promontory and of the aortoiliac axis. This exposure is particularly important for the medial-to-lateral vascular approach.

Similarly to the RC, the left colectomy can be performed in lateral-to-medial and medial-to-lateral approaches.

3.2.1 Medial-to-Lateral Approach

This approach is begun by placing the patient in steep Trendelenburg position with the left side tilted up to allow for the small bowel to be swept away from the root of the mesentery.

The sigmoid mesocolon is retracted anteriorly and the visceral peritoneum incised at the level of the sacral promontory. The incision is continued upward along the right anterior border of the aorta up to the ligament of Treitz. The mesentery is elevated and the IMV identified and the dissection begun there just lateral to the ligament of Treitz. The IMV is skeletonized circumferentially via blunt dissection from its attachments to the left mesocolon. Once this is achieved, the vessel is then ligated using a vessel-sealing energy device, stapler, or clips per surgeon's preference. The

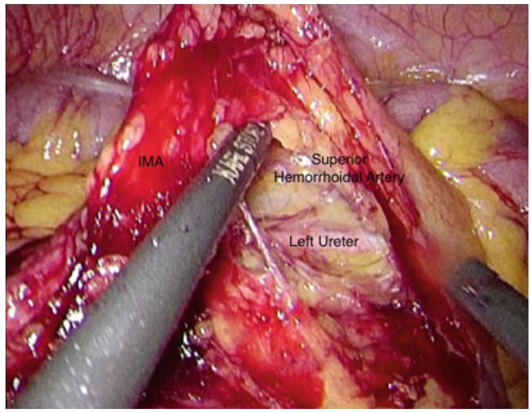

Fig. 3.5 Inferior mesenteric artery identification

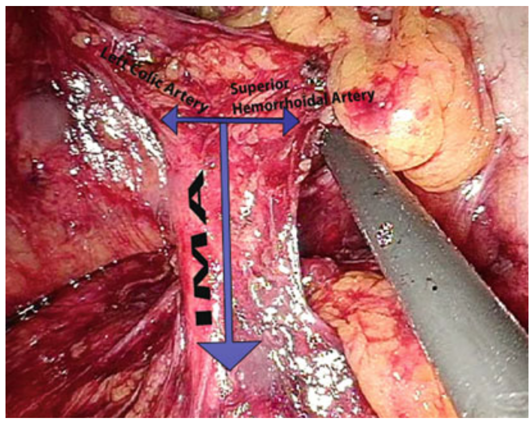

Fig. 3.6 The critical "T structure"

sigmoid colon is then elevated toward the abdominal wall and the overlying peritoneum medial to the right common iliac artery at the sacral promontory incised. The upward traction is maintained and a plane developed bluntly under the superior hemorrhoidal artery. The left ureter is again identified and swept posteriorly and the dissection continued to the origin of the IMA at the aorta (Fig. 3.5). The IMA is then skeletonized circumferentially and the critical "T"-shaped view of safety achieved. This is comprised of the junction of the left colic artery and superior hemorrhoidal artery with the IMA (Fig. 3.6). The IMA is then ligated using a vessel-sealing energy device, stapler, or locking hemoclips per surgeon's preference. The left colic artery is also divided in a similar fashion in most patients.

The sigmoid mesocolon is retracted anteriorly applying traction during dissection. The avascular plane between Toldt's fascia and the sigmoid mesocolon can then be identified and easily divided. At this point, it is important to identify the rectosigmoid junction and the ureters. The dissection continues posteriorly and laterally toward Toldt's line. The sigmoid colon is then completely freed, and the lateral attachments can then be divided using a lateral approach.

In the event that a long segment of sigmoid colon must be resected, mobilization of the splenic flexure is required. The small bowel is repositioned in the pelvic cavity. Traction on the mesocolon of the transverse colon and traction on the adhesions of the splenic flexure will lead to safe division of the splenic flexure.

In the medial approach to the splenic flexure, the posterior attachments of the transverse and descending colon are dissected first. The root of the transverse mesocolon is divided anterior to the pancreas. The dissection plane follows therefore the plane of the previous sigmoid colon mobilization.

The spleen should not be injured by excessive traction, and one should stay as close as possible to the colon. Dissection during this step has to be slow and meticulous with care taken not to damage the tail of the pancreas, splenic vessels, and spleen itself.

When the whole colon has been mobilized, it is possible to dissect down into the pelvis and decide on the site for the anastomosis.

The upper portion of the rectum is mobilized posteriorly and laterally through the presacral space, recognizing the boundary between the mesorectal fascia and presacral fascia itself.

This area of dissection should be approached carefully, particularly on the left side where the superior hypogastric nerve and the left ureter are situated.

The superior hemorrhoidal arteries are usually divided in the posterior upper mesorectum.

In case of left colectomy/sigmoidectomy, the avascular plane is followed, and the dissection is continued until a sufficient distal margin is achieved.

When a low anterior rectal resection (LAR) is performed, a total mesorectal excision (TME) is required. The dissection proceeds posteriorly into the pelvis to about the level when Waldeyer's fascia becomes visible as a thickening of the presacral plane.

The dissection is carried down to the peritoneal reflection in the cul-de-sac anteriorly. Particular attention to hemostasis permits better identification of the small nerve roots and branches of the pelvic nerves and helps avoid iatrogenic injury to the sacral venous plexus.

The dissection then proceeds by taking down the lateral rectal stalks with care to identify and avoid the lateral hypogastric nerve plexus in this region. The dissection is then shifted anteriorly through Denonvilliers' fascia in males and Pouch of Douglas in females by incising the peritoneal reflection between the anterior wall of the rectum and the posterior wall of the seminal vesicles or vagina. This circumferential dissection of the rectum down to 1–2 cm distal to the tumor completes the total mesorectal excision (TME), and the rectum is then clamped at the level of the desired transection and an intraoperative colonoscopy then used to confirm the tumor's distance from the dentate line. A distal margin of at least 1 cm is targeted. The distal rectum is divided with a reticulating 30-mm linear stapler. The fully mobilized left colon is exteriorized through a muscle-splitting incision using one of the left lower quadrant ports or a Pfannenstiel, and the specimen is resected (Fig. 3.7). The proximal division site should be located at around 8–10 cm proximal to the tumor. The anvil is then introduced into the bowel lumen and closed with a purse string; then, the colon is reintroduced into the abdominal cavity. The abdominal incision is closed to reestablish the pneumoperitoneum.

A circular stapler is then introduced into the rectum. The anvil is then connected to the shaft of the stapler and fired (Fig. 3.8).

The anastomosis is checked for leaks with intraoperatrive endoscopy by verifying the integrity of the proximal and distal rings, as well as performing an air test.

Fig. 3.7 Total mesorectal excision specimen

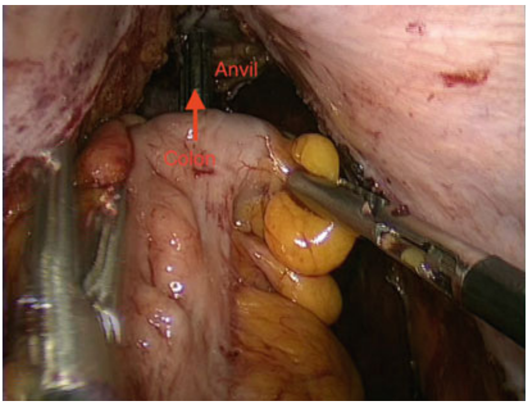

Fig. 3.8 Circular mechanical anastomosis

3.2.2 Lateral-to-Medial Approach

In the lateral-to-medial approach, the first step is to mobilize the sigmoid colon by applying traction and countertraction on the mesocolon. A good visualization of the rectosigmoid junction and the ureters is mandatory to avoid iatrogenic lesions. If a ureter is not clearly visible because of dense inflammation, it is possible to locate it by inserting a ureteral stent.

The lateral approach to the splenic flexure is more often used in open surgery but can also be suitable in laparoscopic colectomy. After the release of the lateral attachments of the descending colon, an ascending incision is made along the line of Toldt. Retraction of the descending colon and the splenic flexure toward the right lower quadrant helps to expose the correct plane. The attachments between the transverse colon and the omentum are divided and the lesser sac opened.

3.3 Robotic Procedure

Although both left colectomy and low anterior rectal resection are feasible using the da Vinci surgical system, the robotic surgery may offer more advantage when applied to the rectal resection.

Laparoscopic rectal surgery is challenging, and room for technical improvements exists. Because of the anatomy of the rectum, its tight boundaries within the pelvis, and its proximity to delicate structures such as the pelvic nerves and the reproductive organs, the application of robotics in rectal cancer surgery can overcome some of the pitfalls of laparoscopic surgery.

Both a totally robotic or hybrid approach have been described for low anterior rectal resection (Hellan et al. 2009c).

However, a hybrid approach utilizing a laparoscopic colonic mobilization and vessel ligation followed by the docking of the robot for the low pelvic part of the operation is a less cumbersome and more efficient use of the robot.

The robot is docked from a left hip approach. Dissection is carried first posteriorly toward the coccyx as distal as possible following the areolar plane, then laterally avoiding injury to the pelvic autonomic plexus, which lies laterally. Dissection is lastly carried anteriorly (Fig. 3.9) while the additional robotic arm pushes the rectum posteriorly. Denonvilliers' fascia/Pouch of Douglas (rectovesical/rectovaginal pouch) is entered by incising the peritoneal reflection between the anterior wall of the rectum and posterior wall of the vagina or the seminal vesicles. Dissection is carried below the level of the tumor (Fig. 3.10). At the most distal end of the rectum, the mesorectum ends and the naked surface of the rectum will become obvious. Once clear margins are achieved, the bowel is divided (Fig. 3.11).

Transabdominal extraction can be performed as previously reported. Transanal extraction is also a possibility by placing a wound protector through the rectum. The specimen is delivered via

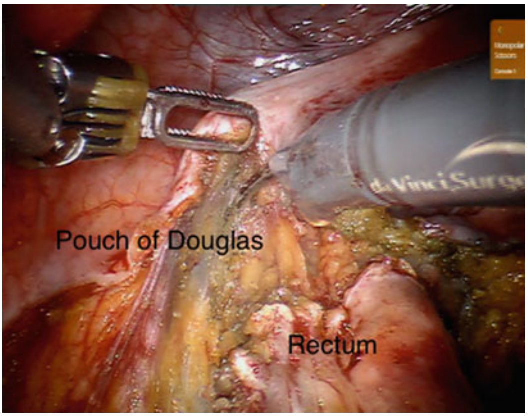

Fig. 3.9 Anterior dissection

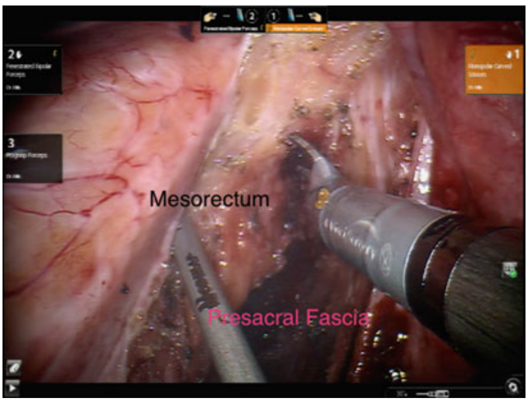

Fig. 3.10 Robotic total mesorectal excision

the anus, and extracorporeal resection can be achieved and the specimen placed back into the abdomen.

After insertion of the appropriate anvil, an anastomosis using an EEA stapler is then performed under laparoscopic vision.

For total mesorectal excision with splenic flexure mobilization, the total robotic approach can be disadvantageous in that it usually requires the surgeon to redock the robot in order to facilitate the splenic flexure mobilization. Therefore, the disadvantage of a total robotic approach is the time needed for redocking. Also the need for multiple redocking means that precise port placement is crucial to avoid collision between the different operative configurations of the robot.

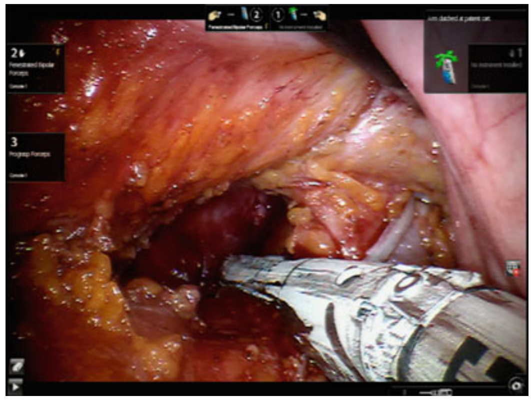

Fig. 3.11 Bowel division

This approach allows the robot to be used for its maximal benefit, which becomes apparent in the narrow confines of the pelvis. Robotic surgeons should be proficient in advanced laparoscopic surgery, and therefore, it may be easier to perform the portions of the operation that require change in patient position via laparoscopy.

3.4 Tips and Tricks for Left Colectomy

- The above comments relating to left colectomy also apply to the Hartmann procedure, except that there is no anastomosis and the mobilized colon is exteriorized through one of the port openings. A colostomy can then be performed at the site of the trocar orifice.
- In the cases of left colectomy for diverticular disease, the opening of the peritoneum can be limited to the mesosigmoid parallel to the colon at mid distance between the colon and the root of the mesosigmoid. The distal resection of the bowel is usually performed right below the rectosigmoid junction.
- Ureteral injuries are one of the most important complications in LC/LAR, which can be avoided by good and extensive exposure and paying respect to the correct plane of dissection. A dissection properly performed above Toldt's fascia does not expose the ureter to accidental injury. Difficult cases, such as those with dense inflammatory reaction, cancer

invasion, or adhesions, may alter the anatomy of the region and render the identification of the ureter problematic. In these special cases, prevention of ureteral injury may be facilitated by the use of ureteral stents.

- Though robotic surgery may overcome some of the intrinsic drawbacks of laparoscopic surgery, the loss of tensile and tactile feedback poses risks on tissue manipulation. Recognizing the visual cues of tissue tension and conceptualizing the spatial relationships of robotic instruments can minimize the risk of iatrogenic injuries.
- The hybrid approach (robotic-laparoscopic) is efficient and allows one-time docking of the surgical robot.
- Perioperative hemorrhage can also be a difficult problem in laparoscopic colorectal surgery. An accurate knowledge of the proper anatomical planes and where hemorrhaging can easily occur can minimize the risk of postoperative complications. Below is a summary of the anatomical planes that are important during dissection of the large intestine and the anatomical structures that can be injured whenever these planes are not respected (Milsom et al. 2006b).
  - The anterior layer of Toldt's fascia in the dissection of the colon → injuries to gonadal vessels and ureters
  - The plane between the mesorectum and the presacral fascia → injuries to hypogastric and pelvic plexus
  - The plane between the anterior rectum and Denonvilliers' fascia in dissection of the rectum → injuries to vagina, prostate, rectum

4 Abdominoperineal Resection

4.1 Indications

The main indication for the abdominoperineal resection (APR) is a malignant disease in which the tumor is invading the anal sphincters or the pelvic floor.

Adenocarcinoma of the rectum and less commonly epidermoid anal canal carcinomas are the most common histological types of cancer that require an abdominoperineal resection.

4.2 Laparoscopic Approach

The patient is placed supine in the modified lithotomy position using stirrups. Surgery is begun in the Trendelenburg position and after cannula insertion the patient tilted right side down. The surgeon and second assistant stand on the patient's right side.

A careful exploration of the entire peritoneal cavity is done, starting with the right upper quadrant and focusing on the liver, because this is an operation done only for malignancy.

Dissection and division began from the left lateral attachment of the sigmoid colon and identification of Toldt's fascia. This is done by retracting the mesosigmoid in a ventrolateral direction using bowel graspers from the left upper and lower quadrants. The surgeon incises the peritoneum to the right of the superior rectal artery starting at the sacral promontory.

Small visceral branches of the nerves, supplying the colon and upper rectum, may be safely divided while carefully preserving the main trunks leading into the pelvis, then the IMA is divided using clips and ultrasonic scalpel.

Dissection then is continued medially beneath the artery, and the left ureter and gonadal vessels are identified and swept posteriorly. If the left ureter cannot be identified easily from the medial approach, the lateral attachments of the sigmoid colon are incised, the sigmoid colon mobilized left to right, and the gonadal vessels and ureter identified and freed from the mesentery.

After the entire sigmoid and mesosigmoid colon are fully mobilized, the rectum is retracted upward and forward. The presacral plane is dissected as far as possible with the ultrasonic scalpel, and hypogastric nerves are easily visualized.

The dissection moves first to the right and then to the left of the rectum. Anterior dissection of the rectum is done in front of Denonvilliers' fascia and posterior dissection along the Waldeyer's fascia. Next, the lateral ligaments on either side of the rectum are divided. Posteriorly, the pelvic nerve is identified and preserved. The cycle of dissecting posteriorly, laterally first on the right, and then on the left is repeated over and over until the tip of the coccyx and beyond is reached, without any significant anterior dissection being done yet. Finally, the left colon is transected using an endostapler.

Division of the skin, subcutaneous fat, and levator ani muscle from a perineal approach allows a window to be made posteriorly through Waldeyer's fascia. The pelvic cavity is entered posteriorly initially, with release of the pneumoperitoneum, then perineal excision of the anus and rectum is completed out in a standard manner.

After inspecting and securing hemostasis, the perineal incision is closed in layers. An end colostomy is fashioned by bringing the colon out through the left iliac fossa trocar site.

4.3 Robotic Procedure

Although the intraperitoneal phase of APR is performed in a similar fashion with a laparoscopic and robotic approach, the robotic system may offer a real advantage during the pelvic phase of the procedure. A basic principle in total mesorectal excision (TME) is sharp dissection along the avascular planes between the presacral fascia and the fascia propria recti (Fig. 3.12). Therefore, especially in patients with a very narrow pelvis, dissection in this region is very difficult. A good operative view is essential for successful rectal surgery.

As illustrated by several studies, a total mesorectal excision plays a major role in reducing the rates of local recurrence and improving survival in rectal cancer (Rottoli et al. 2009; Guillou et al. 2005).

One reason for this is the higher frequency of complete resection of the tumor together with its lymphatic and venous drainage that is achieved by complete removal of the mesorectum (Kayano et al. 2011).

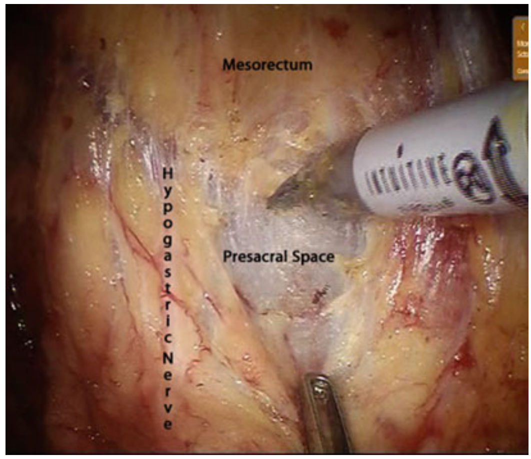

Fig. 3.12 Avascular planes between the presacral fascia and the fascia propria recti

During the TME, the upper and right anterior traction of the rectosigmoid can be achieved by the long grasper placed in the third robotic arm; this grasper remains motionless until further change of position. The anterior-superior traction of the mesorectum with arm 2 enhances even more the exposure of the presacral space and the precise dissection of the avascular space and without nerve branches between the fascias (Angel's hair). With the same exposure, the right side peritoneum is transected, with the hypogastric nerves used as anatomical reference (Fig. 3.13). The rectum and mesorectum are afterward raised anteriorly, allowing a safe dissection of the mesorectal space (Holly Plane) until reaching the pelvic floor (Ramos and Parra-Davila 2014).

In case of locally advanced rectal tumor, whenever an involvement of the circumferential resection margin (CRM) is highly predictable, an extralevator abdominoperineal resection (E-APR) should be performed. This technique aims to improve oncological outcome through removal of more tissue in the distal rectum and en bloc excision of the levator ani muscle creating a cylindrical specimen rather than an hourglass-shaped specimen (West et al. 2010; Bebenek 2009; Holm et al. 2007; Bebenek et al. 2007). The surgery, performed with the patient in lithotomy position, may be carried out as hybrid laparoscopic-robotic or fully robotic surgical procedure (Kang et al. 2012).

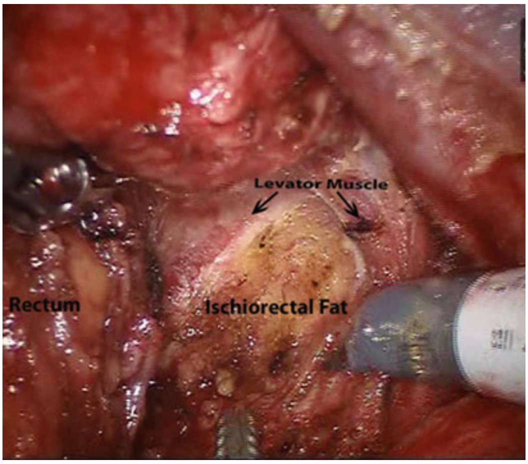

Fig. 3.13 Levator muscle incision

In this surgical approach, the dissection is continued to the levator plane where the muscle is taken widely starting at its origin along the bony structures of the deep pelvis. The fibers of the levator muscle are then divided at their origin in the right and left lateral positions. Afterward, the circumferential division of the muscles is carried out posteriorly and anteriorly. The dissection is continued in the ischiorectal space as far distally as possible utilizing the robotic arms just before encountering the perineal skin.

A circumferential incision is made around the anus. Since the levator muscles have been divided, the ischiorectal fat is encountered and the prior dissection plane quickly found.

4.4 Tips and Tricks Abdominoperineal Resection

- In the TME phase, the key maneuver is going from "known to unknown," usually meaning posterior to lateral, and remembering to avoid dissecting into the vagina or through Denonvilliers' fascia unless the tumor is infiltrative there (Milsom et al. 2006c).
- In the hybrid robotic-laparoscopic approach, the robot is used only to perform total mesorectal excision; the mobilization of the left colon and upper rectum and ligation of the inferior mesenteric vessels are performed laparoscopically.
- Currently, the indications for a robotic assisted approach for the treatment of distal rectal cancer are mainly male and obese patients who have been treated with preoperative chemoradiotherapy. The excellent quality of the mesorectal dissection, associated with excellent vision and pelvic surgical field exposure, may translate into lower rates of conversion and positive circumferential resection margin, and perhaps a lower incidence of anastomotic leak, even without conducting routine ileostomy.

5 Cross-References

► Surgical Anatomy of the Colon, Rectum, and Anus

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Topographic Anatomy in Proctologic Surgery

4

Ezio Falletto

There is no considerable muscle in the body whose form and function are more difficult to understand than those of the levator ani, and about which such nebulous impressions prevail. (Dickinson R: Studies of the levator ani muscle 1889)

Contents

1 Introduction

The pelvic floor's shape is usually described as a boat's hull, but the most similar comparison is to a funnel with a wide large upper part (the pelvic floor in the strictest sense of the word) and two lower bottlenecks: the urethral and anal sphincters.

The topographic anatomy of the human perineum is defined by four palpable points of bony pelvis: the symphysis pubis anteriorly, the left and right ischial tuberosities laterally, and the coccyx posteriorly.

The transverse perineal muscles (left and right) set in the middle of the perineum and with a horizontal transverse position divide the perineum in an anterior (with urological and genital apparatus) and posterior (with anal canal) region. Just anteriorly to the transverse perineal muscle on the midline, the central point of the perineum or perineal body is present.

2 The Anus: Macroscopic Aspects

The internal surface of anal canal is characterized by the pectinate or dentate line (from his peculiar shape) at about 2 cm from anal verge. It divides

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the anal canal in an upper part lined by mucosa and a lower part with the skin. At this level, the embryological junction between postallantoic gut (from endodermic sheet) and the proctodeum (from ectodermic sheet) is placed. At the dentate line, there are little pockets known as crypts of Morgagni in which ducts of anal glands flow out. The internal opening of anal fistula is usually considered to be set in this place.

Above the dentate line, mucosa is thrown in longitudinal folds called Morgagni's columns. Under the mucosa, the internal hemorrhoidal venous reticulum is often visible. Theoretically in this zone, sensitive touch receptors are missing, so it can be considered an insensitive epithelium to cutting or other trauma (patient feels pain if the rectal wall is dilated, for the presence of distension receptors).

Near 0.5–1 cm below the dentate line, the anocutaneous line could be found. The lining between these two lines is called anal pecten or transitional zone, and it is formed by bright modified keratinized skin but without hair or sebaceous glands and closely adherent to underlying tissues.

Below anocutaneous line, a normal, hyperpigmented skin with hair follicles and glands is present.

3 Anal Sphincters

Anal sphincter system is a double cylindrical muscular structure shaped by an internal cylinder (the internal sphincter) and the external sphincter around it.

– The Internal Anal Sphincter: The more internal cylinder of sphincter apparatus is a thickening of the last part of circular muscle coat of the distal rectum. For this reason, it is difficult to measure a precise length, but usually it is defined from 1.5 to 2 cm of length with many sex- and constitution-related differences.

It is composed by smooth muscular involuntary fibers, and it is mainly responsible of anal resting pressure.

– The External Anal Sphincter: It is usually considered formed by three bundles of striated voluntary muscle fibers, and it is responsible of anal voluntary contraction. The superficial bundle is subcutaneous, wrapping the internal sphincter, and it is the first muscular structure we run into.

The superior bundle can be divided by puborectalis muscle fibers with difficulty.

Sphincter fibers have a strong link with the central perineal tendon (or body) anteriorly and with the anococcygeal ligament posteriorly.

Its length is 2.5–3.5 cm with many sex- and constitution-related differences. In particular in women it is usually shorter, especially anteriorly.

– Intersphincteric Longitudinal Muscle Fibers: They lie between internal and external sphincters. They consist of nonstriated muscle fibers mixed with elastic tissue, and they are the distal continuation of longitudinal muscle layer of the rectal wall. Some of these fibers go through the internal sphincter, and inferiorly they pass over the end of the internal sphincter and are attached to the anal skin forming the anocutaneous line that represents the intersphincteric groove, the macroscopic endoanal border between the internal sphincter and external sphincter.
– Musculus Submucosa Ani or Parks' Ligament: Smooth muscle fibers and elastic connective tissue lying above the internal sphincter and under mucosa and anal skin. It has connections with intersphincteric longitudinal muscle fibers, the superficial part of the external sphincter and anal skin, also called corrugator cutis ani. It was considered an important pathogenic factor (if damaged) in the onset of hemorrhoidal prolapse.

In 1976 Shafik proposed an interesting mechanical theory (the three-loop system) in which the three muscular components of anal sphincters work with different force's vectors: deep and superficial loop pulling anteriorly toward the pubis and the central loop pulling posteriorly toward the coccyx. In this way, a complete closure of the anus can be obtained.

4 Blood Supply of Anal Canal and Sphincters

– Arteries: The anal canal above the pectinate line is supplied by the terminal branches of the superior rectal (hemorrhoidal) artery, which is the terminal branch of the inferior mesenteric artery. The middle rectal artery (a branch of the internal iliac artery) and the inferior rectal artery (a branch of the internal pudendal artery) supply the lower anal canal. The terminal branches of all these arteries have anastomoses with branches of opposite site and near arteries.

Nevertheless, recent studies have demonstrated that superior rectal artery play a predominant role in blood supply of the distal rectum and anal canal, and the middle rectal artery is missing in near half of cases. Besides above 4 cm from the anorectal junction, majority of rectal branches are located outside the rectal wall, and then they pass thorough the rectal wall and reach anal submucosa at near 2 cm from pectinate line.

– Veins: venous drainage of the anal canal occurs by two plexuses of veins; below the pectinate line, beneath the anal skin, lies the external hemorrhoidal venous plexus which drains inferior hemorrhoidal vein into systemic veins via internal iliac and internal pudendal veins.

Above the pectinate line, submucosally, lies the internal hemorrhoidal venous plexus which drains into portal system via middle (inconstant) and superior hemorrhoidal vein and inferior mesenteric vein.

5 Lymphatics of Anal Canal

Following blood vessel supply, lymphatic drainage of the anal canal takes place via three routes:

– Upwards: through lymphatics and glands accompanying superior hemorrhoidal vessels to inferior paramesenteric glands
– Laterally: following medial hemorrhoidal vessels to pelvic side wall
– Downwards: following inferior hemorrhoidal vessels to parailiac glands and inguinal nodes

6 Nerve Supply of Sphincter

– *Motor Innervation*:
  - Internal Anal Sphincter: It is a smooth muscle; consequently it is under involuntary control; it operates in an automatic (reflex) manner. It is controlled by:
    – Parasympathetic nerves (from the S2, S3, and S4 levels of the spinal cord) cause relaxation of the internal anal sphincter.
    – Sympathetic nerves (from the T11-L2 levels of the spinal cord) induce contraction of the internal sphincter.
  - External Anal Sphincter: It is a striated, voluntary controlled muscle. It is controlled by:
    – Pudendal Nerve: formed by spinal roots from the S2, S3, and S4
    – Perineal branch of the fourth sacral nerve (inconstant).

    The pudendal nerve leaves the pelvis via the greater sciatic foramen. It runs under the lower border of pyriformis and crosses the ischial spine and sacrospinous ligament to enter Alcock's canal in the lateral aspect of the ischiorectal fossa. The first major branch of the pudendal nerve is the inferior rectal nerve, which gives motor supply to the external sphincter and sensation to the perianal skin. After giving off the inferior rectal nerve, the pudendal nerve continues as the perineal nerve. It gives motor supply to the anterior perineal muscles and the sphincter urethra and continues then as the dorsal nerve of the penis/clitoris.

    The puborectalis muscle gets some motor supply from the pudendal nerve, but most of the motor supply comes from a direct branch of S4.

– *Sensory innervation*: The cutaneous sensation of the anal and perianal skin up to the pectinate line is conveyed by afferent fibers of the inferior rectal nerve, the first branch of the pudendal nerve.

Sensation of anal mucosa above dentate line is mediated via the parasympathetic nerves.

7 Levator Ani Muscle

If you consider the pelvic floor's shape as a funnel, the levator ani consists in the wide upper part of funnel. It is a broad, thin muscle attached peripherally to the inner surface of the side of the pelvis and united medially with muscle fibers of the opposite side.

It consists of three parts:

– Puborectalis Muscle: It is the most medial muscle and forms the upper part of external anal sphincter apparatus. It arises from the lower part of the back of the symphysis pubis and runs backward alongside the anorectal junction to join his fellow behind the bowel at the level of the anorectal junction, forming a strong U-shape loop which slings the rectum to the pubis. Its contraction approaches bowel to pubis, forming the anorectal angle.
– Pubococcygeus M: It forms the median part of the levator ani. It arises from the back of the pubis and the anterior part of obturator fascia and is directed horizontally backward. Its fibers lie between puborectalis and iliococcygeus muscles, posteriorly to the bowel they fuse with fibers of the opposite side to constitute a broad fibrous band lying on the anococcygeal raphe.
– Iliococcygeus M: This lateral and very thin muscle arises from the ischial spine and posterior part of pelvic fascia covering the obturator internus muscle. Fibers run backward, downward, and medially to the coccyx forming the anococcygeal raphe.

8 Blood and Nerve Supply of Levator Ani Muscle

Arteries: Branches of the internal pudendal artery ramify to supply levator ani muscles.

Veins: Internal pudendal veins drain blood from these muscles to internal iliac veins.

Nerves: The contraction of levator ani muscle is supplied by a double innervation, from inferior rectal nerve deriving from the pudendal nerve and directly by twigs from fourth sacral nerves.

9 Lateral Spaces of Pelvic Floor

– Perianal Space: It is a subcutaneous space, lateral to superficial part of external sphincter. It contains finely lobulated, subcutaneous fat, and here hematoma or perianal abscesses can occur.
– Ischiorectal Fossa: This is a pyramid-shaped space located below levator ani muscle, above perianal space, and laterally limited by the fascia of obturator internus. A thin nonmuscular, fascial partition divides it from underlying perianal space.
– Pelvirectal or Supralevator Space: This is a deep space between peritoneal floor, above levator ani muscle, and limited medially by rectal wall.

Anatomy of Alcock's Canal: The space within the obturator fascia lining the lateral wall of the ischiorectal fossa that transmits the pudendal vessels and nerves is called Alcock's canal. This fascial compartment is located in the inferior border of the obturator internus fascia. It extends from the ischial spine to the posterior edge of the ischiopubic ramus. Opening the canal, the internal pudendal vessels and the three branches of the pudendal nerve can be identified.

The pudendal nerve comes down from sacral nerve roots 2, 3, and 4, runs underneath the piriformis muscle, goes between the sacrospinous and sacrotuberous ligaments at the ischial spine, travels through Alcock's canal between the obturator internus and levator ani muscles, and divides into three terminal branches.

In this condition, the nerve can be compressed at:

• The space between sacrotuberous and sacrospinous ligaments (~70 % cases)

- Within the pudendal canal of Alcock (~20 % cases) – while straddling of the falciform process of the sacrotuberal ligament by the pudendal nerve and its branches
- Under an hypertrophic piriformis muscle

10 Anatomy of Rectum

The average length of the human rectum may range between 10 and 15 cm, depending on sex and body type differences. Its caliber can be compared to that of the sigmoid colon at its onset. It begins at the rectosigmoid junction, 3–5 cm above the peritoneal reflection (at the level of third sacral vertebra) and ends at the anorectal junction in front of the coccyx tip.

However, it gets dilated near the anus, where it forms the rectal ampulla. The internal cavity of the rectum is divided into three or four chambers; each chamber is partly segmented from the others by permanent transverse folds: the valves of Houston, namely, the superior valve at about 11–13 cm from the anal verge, the middle one at about 8–9 cm from anal verge, and the inferior valve at 5–6 cm.

It has two anteroposterior flexures: the sacral flexure that follows the curve of sacrum and coccyx and the anorectal or perineal flexure that follows the anorectal angle.

In the upper third of the rectum, the front and sides are covered by the peritoneum; in its middle third, the front only; its lower third is subperitoneal and devoid of the peritoneum.

The distal subperitoneal rectum is surrounded especially posteriorly by the mesorectum, an adipose tissue surrounded by its own fascia. The posterior face of mesorectum looks like a “bilobate lipoma” and reach the presacral or Waldeyer’s fascia. Anteriorly the mesorectum is thin and it stretches up to the Denonvillier’s fascia. The fascia that circumscribes the rectum offers a relative avascular plane and it is called “the holy plane” by Heald.

In male the Denonvillier’s fascia divides the rectum from the prostate, seminal vesicles, vasa deferentia, ureters, and bladder wall. In female the extraperitoneal rectum lies immediately behind the posterior vaginal wall.

The mesorectum is very rich of lymphatic vessel and nodes; for this reason it is the first field of rectal cancer spreading.

11 Blood Supply of the Rectum

– Arteries: The rectum, which is part of the distal portion of the hindgut, is mainly supplied by the superior rectal artery, arising as a main branch from the inferior mesenteric artery (IMA).

  The *superior rectal artery* typically continues in the same downward course as the IMA to reach the back of the upper third of the rectum. At this point, it bifurcates into two vessels. The larger right branch supplies the posterior and lateral surface of the rectum. It divides into two main branches, which run down to the right anterior and posterior aspects of the rectum. The smaller left branch supplies the anterior surface of the rectum and continues undivided down the left lateral aspect of the rectum. These branches generally break up into smaller vessels that finally penetrate the muscle layer to reach the submucosa. Here they proceed downward as straight vessels, which run in the columns of Morgagni and terminate usually above the anal valves as a capillary plexus.

  The *middle rectal arteries* originate from the anterior divisions of the internal iliac arteries or from their inferior vesical branches. They proceed medially and forward below the pelvic peritoneum, in the tissue of the lateral ligaments, to reach the rectal wall. Here they anastomose with the branches of the superior and inferior rectal arteries. However, their arrangement is variable and the middle rectal artery may be absent.

  The *inferior rectal arteries* spring from the internal pudendal arteries in Alcock’s canal in the fascia of the outer walls of the ischiorectal fossa. They run medially and slightly forward, dividing into branches that penetrate the external and internal anal sphincters, and finally reach the submucosa and subcutaneous tissue of the anal canal.

– Veins: The venous drainage of the rectum is to the inferior mesenteric vein into the portal system. Veins from the upper two-thirds of the rectum are drained by the superior rectal vein.

  Veins from the lower third of the rectum are drained by the middle and inferior rectal veins into the internal iliac veins.

12 Lymphatics of the Rectum

– *Intramural Lymphatics*: In the submucous and subserous layer of the rectal wall, there are continuous lymphatic plexuses, which drain into the extramural lymphatics.
– *Extramural Lymphatics*: The extramural lymphatics follow the blood vessels supplying the rectum and anal canal. Lymph from the parts of the rectum that receive blood supply from the superior rectal artery is drained to pararectal and to superior rectal nodes. From these nodes, lymph passes to inferior mesenteric nodes.

The lymphatic drainage of the remainder of the distal rectum and anal canal is dependent on its relationship to the mucocutaneous junction. The area proximal to the mucocutaneous junction can drain superiorly (parallel to the middle rectal arteries on the corresponding pelvic side wall) or inferiorly to follow the inferior rectal arteries. These two possible pathways lead to internal iliac nodes, common iliac nodes, and the lumbar trunk. Lymphatic drainage below the mucocutaneous junction does not parallel blood vessels.

In the perineum, the collecting lymphatic ducts pass together with lymphatic channels from the perianal skin to superficial inguinal nodes.

Part II

Physiology

Physiology of the Colon

5

Gabrio Bassotti and Edda Battaglia

Contents

Abstract

The colon (or large bowel), the gut portion comprised between the ileocecal valve and the anal sphincter, is the distal part of the intestine. Physiologically, the colon has important functions, such as absorption of water and electrolytes and storage, transport, and expulsion into the external environment of the contents residual from the previous digestive processes, i.e., the feces. These processes are based on biochemical and physical pathways and are controlled by both extrinsic and intrinsic nerve supplies. This chapter will take into consideration such physiological functions, particularly those related to secretion/absorption and motility.

Abbreviations

HAPC	High-amplitude propagated contractions
LAPC	Low-amplitude propagated contractions
MMC	Migrating motor complex
RMC	Rectal motor complex

1 Introduction

The colon (or large bowel) is the most distal part of the gut; it is connected to the small intestine by the ileocecal valve (that represents the proximal anatomic boundary) and after developing in several segments (cecum, ascending, transverse,

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descending, sigmoid, and rectum) communicates with the external environment by means of the anal sphincter. Although there are several, even substantial, anatomic and physiological differences between the colons of different animal species, in humans the large bowel is basically conformed as that of omnivores, with the most strict resemblance (apart from apes) to that of pigs (Bassotti 1997).

The main physiological functions of the colon are those related to the processing of the contents arriving daily from the terminal ileum; this processing includes storage, extraction of all the useful contents, packing, distal transport, and eventually external expulsion of the residual contents that represent, as a final result, the fecal matter. To this purpose, the colon possesses absorptive, secretory, and motor activities that integrate each other to carry on the above functions (Bassotti 1996).

2 Absorptive and Secretory Functions of the Large Bowel

The colon is the final area for recapturing electrolytes and water prior to excretion, and in order to maintain this electrolyte homeostasis, a complex interaction between secretory and absorptive processes is necessary (Geibel 2005). On average, about 9 L of water are absorbed daily by the small intestine and colon; of these, about 80 % is absorbed by the small intestine with isotonic mechanisms and the remaining 20 % by the large intestine against an osmotic gradient (Powell 1987).

Water transport across gut epithelia follows two pathways (Laforenza 2012): (1) a paracellular route, through the spaces between cell junctions, and (2) a transcellular route, through apical and basolateral cell membranes (Field 2003; Fischbarg 2010). Lateral intercellular spaces also provide a compartment in which both pathways may communicate (Kaissling and Kriz 1985). Although the paracellular pathway has been traditionally considered as the main route for water movements coupled to nutrient absorption, to date this pathway for water transport has been shown to be limited (Masyuk et al. 2002). The transcellular pathway is based on different mechanisms, such as passive diffusion through the phospholipid bilayer, cotransport with ions and nutrients, and diffusion through water channels called aquaporins (Benga 1989; Verkman 2011; Zeuthen 2010). The latter are responsible for osmotically driven transmembrane water movements (Mollajew et al. 2010).

Every day, about 1,500 mL of semisolid contents cross the ileocecal valve; of these, the colon absorbs most of its water and electrolytes, leaving less than 100 mL of liquids, that are eventually eliminated with the feces. Most of this absorption is carried out in the proximal colon; the colonic mucosa actively absorbs sodium, whose electric gradient in turn also causes absorption of chloride. In normal conditions, there is a net colonic absorption of Na^+ and Cl^-, the two quantitatively most important inorganic ions transported in the large intestine. To be secreted, Cl^- ions are accumulated above electrochemical equilibrium by a Na^+–K^+–$2Cl^-$ cotransporter in the basolateral membrane, the prominent Cl^- loading transporter in this membrane beside a Cl^-–HCO_3^- exchanger at the same location (Rajendran et al. 2000; Schultheiss et al. 1998). When Cl^- channels open, Cl^- penetrates into the colonic lumen, and it is followed by a paracellular flux of Na^+ due to electroneutrality and by a flux of water due to osmotic processes. Colonic absorption and secretion is controlled by neurotransmitters, mainly released from the submucosal plexus, hormones, and paracrine substances, acting on membrane-bound receptors in order to modify intracellular second messenger systems regulating ionic transport across the epithelium. In addition, small gaseous molecules, the so-called gasotransmitters (nitric oxide, hydrogen sulfide, and carbon monoxide), can act as signaling molecules to affect intestinal transport (Pouokam et al. 2011).

An important agent secreted by colonic cells is mucus, constituted by large, highly glycosylated proteins called mucins (McGuckin et al. 2011). The mucus layer is important to protect the colonic wall from both direct damages and from bacterial aggressions, to aggregate and lubricate the feces and, also thanks to its alkalinity (pH 8, due to a great amount of sodium bicarbonate), to constitute a mechanical barrier protecting the mucosa from the acids present in the feces (Johansson et al. 2013). Moreover, this layer serves as a partial food source for intestinal bacteria that synthesize short-chain fatty acids from the degraded material and, depending on the bacterial species, deliver acetate, propionate, and butyrate as energy sources for the host epithelium (Wong et al. 2006).

3 Motor Functions of the Large Bowel

Colonic motor activity mainly serves to transport, and eventually excrete into the external environment, the final result of the digestive processes, represented by the feces. This function is carried out by the mechanism of defecation through a process that, in physiological conditions, involves coordination of ileal, colonic, and anorectal motility and is regulated by both central and peripheral pathways (Brookes et al. 2009). It is worth noting that, differently than in segments of the upper gastrointestinal tract, the movement of contents through the large bowel is measured in hours or days, instead of seconds or minutes, so that prolonged observations (such as those obtained by means of pancolonic manometry) are needed (O'Brien and Phillips 1996).

The contractile activity of the large bowel is represented by waves of different amplitude, occurring both as single events or grouped in bursts (Bassotti et al. 1993), and it may be subdivided in segmental and propagated activity, as also described by radiological techniques several years ago (Ritchie 1986); the first mainly serves as a means to favor absorption (also helped by the peculiar anatomic conformation, the haustrations, in the proximal segments of the viscus) whereas the second has mainly a transport function.

Segmental activity. Segmental contractions, the contractile equivalent of myoelectrical short spike bursts (Bueno et al. 1980), represent most (probably more than 80 %) of the overall daily colonic motor activity and are generally characterized by relatively low-amplitude (on average, less than 60 mmHg) waves, even though sporadic single nonpropagated waves of amplitude >100 mmHg may be observed (Narducci et al. 1987). These contractions, by means of to-and-fro movements, delay the transit of contents allowing the viscus to maximize its absorptive function that is also enhanced by the *haustrae* (Ritchie 1971). By means of combined scintigraphic and manometric techniques, it has been demonstrated that a bolus injected at the splenic flexure level may spread in an oral or aboral direction, in relation to the differences between the motility index (mostly due to segmental contractile activity) at the injection site and at adjacent segments (Moreno-Osset et al. 1989). Indeed, segmental contractions may generate forward propulsion of contents, provided that an adequate distally directed pressure gradient develops (Garcia-Olmo et al. 1994), and experimental in vitro models postulate that a zone of muscular relaxation preceding the contraction is an important element for transport (Sinnott et al. 2012).

The segmental activity occurs apparently in a random fashion and is represented by single contractions of variable shape and dimensions appearing at different colonic segments in the time course, often without apparent relationship between each other; however, especially in the distal segments (descending to rectum), segmental contractions may appear arranged in bursts having a more or less regular frequency (Bassotti et al. 1995; Dinoso et al. 1983). These groups of waves are generally arrhythmic, but in some instances (that represent less than 6 % of the overall daily motility of the large bowel), a rhythmic frequency may be observed; these harmonic patterns, in which the three cycles per minute predominate, are recorded especially in the sigmoid colon (Bassotti et al. 1989b) (Fig. 5.1). This fact has led to hypothesize the presence of a functional sphincter at the rectosigmoid junction (Ballantyne 1986), where a high-pressure zone has been repeatedly described (Shafik et al. 1999; Wadhwa et al. 1996).

Differently than in the small bowel and in some animal species (e.g., the dog (Sarna et al. 1984)),

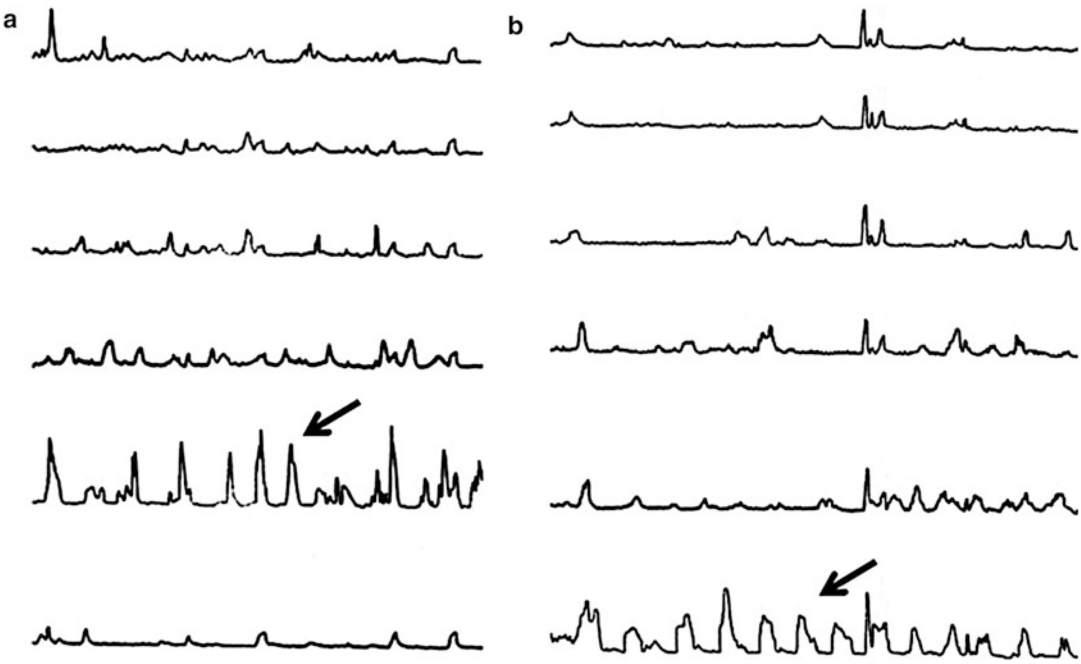

Fig. 5.1 (**a**, **b**) Representative manometric tracings of colonic segmental contractions. The arrows show the presence of regular (about 3 cycles per minute) contractile activity in the sigmoid colon (**a**) and in the rectosigmoid junction (**b**)

however, the bursts of contractions do not appear regularly spaced in the time course following a periodic pattern with specific aborally propagated contractile events such as that described in the small bowel (i.e., the migrating motor complex, MMC (Deloose et al. 2012)). In the human large bowel, propagation of the contractile bursts may be observed (Bampton et al. 2001; Hagger et al. 2002), even though most of them are nonpropagated or migrate orally (Dinning et al. 2014; Rao et al. 2001). The rectum, also, displays a peculiar motor pattern, the so-called rectal motor complex (RMC), that occurs independently from the MMC of the small bowel (Kumar et al. 1990), and it is characterized by a series of contractions featuring a frequency of 2–3 per minute, a burst duration of more than 3 min, and amplitude more than 5 mmHg (Enck et al. 1989; Orkin et al. 1989; Prior et al. 1991). The function of the RMC is unknown; however, it has been speculated that these complexes might act as a sort of braking mechanism, since they are often seen to propagate in a retrograde manner (Rao and Welcher 1996) and are accompanied by a rise in anal canal pressure (Ferrara et al. 1993) but are not related to anal events such as passage of flatus (Rønholt et al. 1999).

Propagated activity. In man, colonic propagated activity may be basically subdivided into two types of propagating contractions, the so-called low-amplitude propagated contractions (LAPC) (Bassotti et al. 2001) (Fig. 5.2) and the high-amplitude propagated contractions (HAPC) (Bassotti and Gaburri 1988; Bharucha 2012; Crowell et al. 1991; De Schryver et al. 2002) (Fig. 5.3); both events are probably the manometric equivalent of the migrating long spike bursts described by electromyographic techniques (Bueno et al. 1980; Garcia et al. 1991) and are the main factors responsible for the transport of contents within the colon.

LAPC represent the most frequent type of propagated events, occurring up to more than 120 times per day, with amplitude less than 50 mmHg and propagation over relatively short (about 20 cm) distances (Bampton et al. 2001;

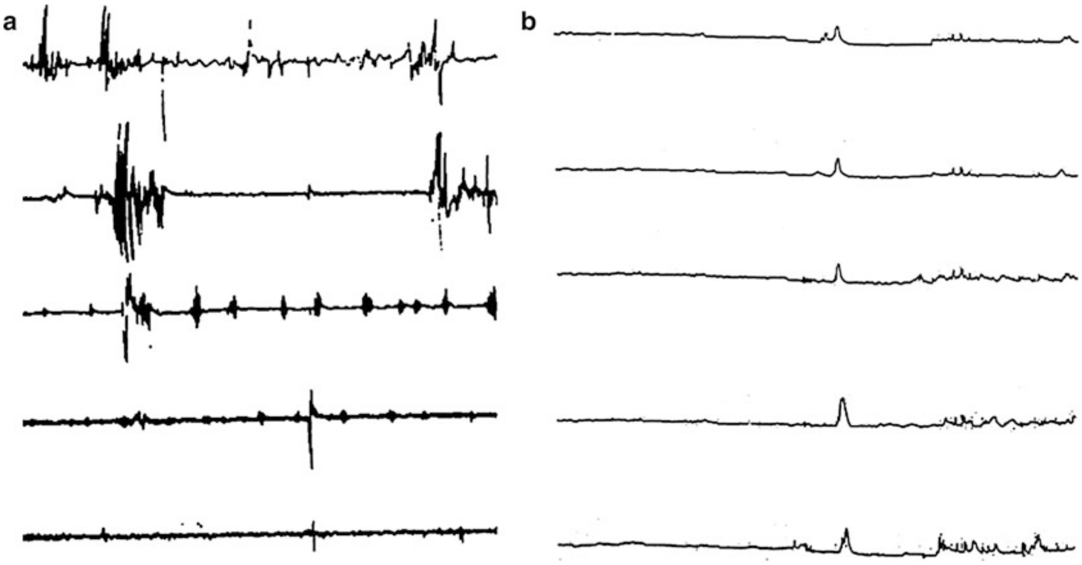

Fig. 5.2 Colonic low-amplitude propagated contractions. (**a**) Electromyographic recording. (**b**) Manometric recording

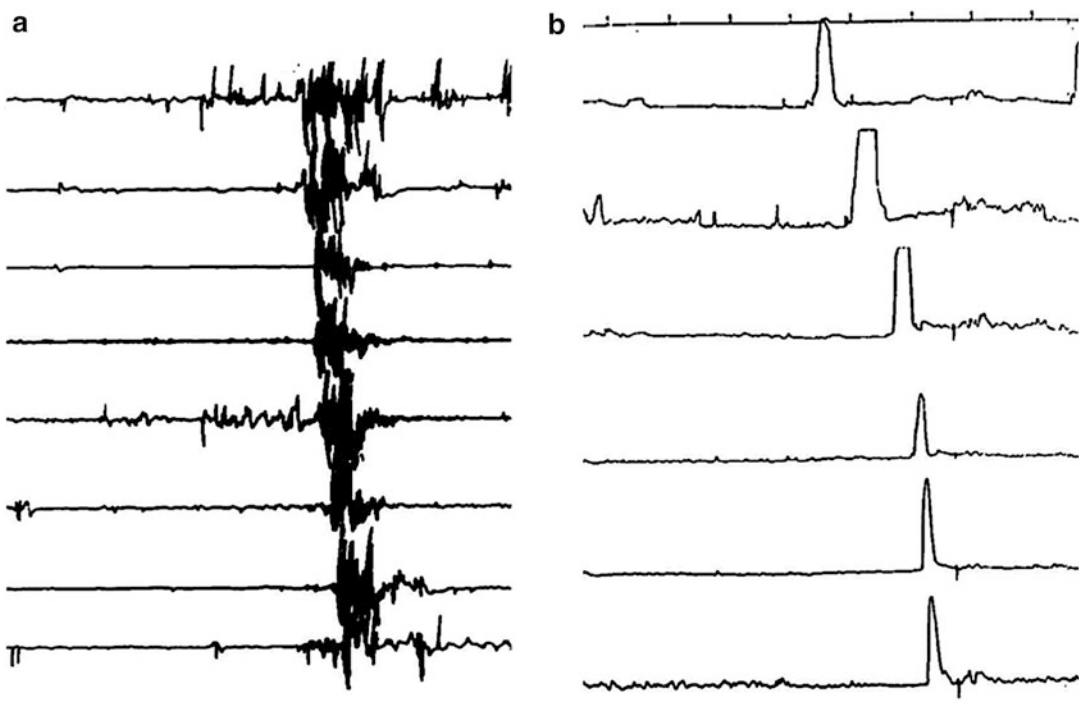

Fig. 5.3 Colonic high-amplitude propagated contractions. (**a**) Electromyographic recording. (**b**) Manometric recording

Bassotti et al. 2001; Rao et al. 2001). These propagated sequences have also been shown to display a spatiotemporal linkage, in that two consecutive events originating from different colonic segments overlap (Dinning et al. 2009; Dinning et al. 2010b); thus, a series of regionally linked LAPC may span the entire length of the colon (Dinning et al. 2010a). The exact physiological function of LAPC is unknown, but some studies suggest that these events may be associated with the transport of liquid colonic contents (Chauve et al. 1976; Cook et al. 2000) and of gas (Bassotti et al. 1996).

The presence of infrequent, vigorous propulsive contractions able to shift colonic contents over long tracts of the large bowel had been documented in man with radiological methods at the beginning of 1900 (Hertz 1907; Holtzkhnecht 1909) and then called mass movements (Holdstock et al. 1970); subsequently, combined radiological and manometric techniques demonstrated that the manometric equivalent of mass movements were represented by the HAPC (Torsoli et al. 1971). These contractions are less frequent compared to LAPC, have higher average amplitude (about 100 mmHg) compared to the latter, and represent a constant event in healthy subjects (Bassotti et al. 1992). The main physiological function of HAPC, that often starts in the proximal colon (Dinning et al. 2008), is that of shifting aborally relatively large amounts of contents (Ritchie 1971), creating a right-left pressure gradient able to start the mechanism of defecation and sometimes are associated with the urge to defecate, although only about one third of them travels beyond the rectosigmoid junction (see below), as shown by studies with intracolonic instillation of bisacodyl (Bassotti et al. 1999a; Kamm et al. 1992). HAPC are usually propagated in a caudal direction (although retrograde propagation is present, even in normal subjects, and is usually observed in proximal segments) and appear more frequently in daylight hours, after meals, and after morning awakening. It is also worth noting that the frequency of HAPC is significantly greater in children younger than 4 years, and this is probably correlated to the more frequent bowel movements observed in infants and toddlers (Di Lorenzo et al. 1995).

The basic mechanisms controlling the onset of LAPC and HAPC are poorly known. Some human studies suggest that colonic distension and some chemical stimulation may evoke these contractions (Bampton et al. 2002; Bassotti et al. 1994; Liem et al. 2010), that cholinergic stimulation does not elicit this kind of activity (Bassotti et al. 1991), and that physical exercise is able to stimulate both types of propulsive contractions (Cheskin et al. 1992); HAPC may also be elicited by colonic fermentation of a physiological malabsorbed amount of starch (Jouët et al. 2011).

Circadian trends of colonic motility. In humans, colonic motor activity widely fluctuates around the clock, and it is now clear that both electrical and contractile activity and muscle tone vary according to common physiological events such as eating, sleeping, and morning (or sudden) awakening (Auwerda et al. 2001; Bassotti et al. 1990; Frexinos et al. 1985; Narducci et al. 1987; Steadman et al. 1991). Thus, colonic motility is maximal during the daylight hours and reaches a minimum during night and when sleeping (Furukawa et al. 1994; Narducci et al. 1987). Food ingestion, also, is one of the more powerful physiological stimuli for large bowel motility. The colonic motor response to eating is preceded by a cephalic phase (Rogers et al. 1993), starts within 1–3 min following ingestion of the first mouthfuls of food, and lasts at least 2–3 h (Bassotti et al. 1987); it is mainly composed by segmental contractions (even though HAPC may be observed after meals (Bassotti 1990)), and it is paralleled by increased colonic smooth muscle tone (Steadman et al. 1992). The response to eating is also influenced by the caloric content and the composition of the meal, with stimulation following ingestion of fat and carbohydrates (Levinson et al. 1985; Rao et al. 2000) and inhibition following ingestion of proteins (Battle et al. 1980), and it is mediated by peptides (gastrin, neurotensin, cholecystokinin), prostaglandins, vagal cholinergic pathways, and serotoninergic pathways (Ducrotté et al. 1994). In man, proximal and distal colonic segments exhibit different properties in response to meal ingestion, as shown by the fact that the proximal ones display a relatively rapid – but less sustained in the time course – response compared to the distal ones (Bassotti et al. 1989a); proximal and distal segments of the large bowel also feature quantitatively different tonic activity, likely due to different viscoelastic properties and diameter (Ford et al. 1995). Thus, as also supported by scintigraphic studies (Krevsky et al. 1986; Picon et al. 1992), it has been hypothesized that the colon has different physiological activities, with the proximal segments deputed to the mixing and the storage of contents and the distal ones functioning as conduit apt to propel the feces toward the rectum (Bassotti et al. 1999b). Of interest, intestinal continuity seems to be essential for the elicitation of a colonic motor response to eating (Hallgren et al. 1995).

Defecation. Defecation is a complex physiological event that involves both central (cerebral) and peripheral (colorectal) stimuli (Palit et al. 2012). Thus, in physiological conditions, every individual is able to control how, where, and when to defecate, according to the needs and the social interactions, such as the availability of toileting facilities, and it has been developed in the course of human evolution (Bassotti and Villanacci 2013). Defecation may be subdivided in four different phases, represented by the basal phase, the pre-expulsive phase, the expulsive phase, and termination of defecation (Palit et al. 2012).

Basal phase. This phase is constituted by the above described colorectal motor activity that continuously moves the contents toward the rectum and acts synergically with the puborectal muscle that exerts a resting contractile traction able to maintain the anorectal angle at approximately 90° (Mahieu et al. 1984) and the anal sphincter. The latter is normally contracted, providing an airtight seal, except when the subject consciously wants to defecate or pass flatus. This function is provided by a tonic activity of both the external and the internal (the latter provides about 80 % of the overall activity) anal sphincter, in association with the anal cushions (Frenckner 1975). To allow descent of rectal contents into the upper portion of the sphincter itself, and to perceive their physical nature, the internal sphincter displays intermittent and transient relaxations. This so-called "sampling reflex," in accordance to the nature of the contents (solid, liquid, gas) and the social opportunity, eventually induces the subject to voluntarily relax the sphincter (rectoanal inhibitory reflex) that starts the actual defecation (Miller et al. 1988).

Pre-expulsive phase. Starting with a sensation of "call to stools," this phase shows a strict correlation between appearance of HAPC and the urge to defecate; of interest, HAPC sequences often start before actual defecation, shifting aborally discrete amounts of contents and activating distal colorectal afferents by distension of the viscus wall (Bampton et al. 2000). The progressive rectal distention causes an initial awareness of filling that becomes constant with continued distention and therefore an urge to defecate until the maximum rectal tolerable volume is reached (Broens et al. 1994).

Expulsive phase. If the subject decides that it is opportune to evacuate, the sampling reflex and the presence of a defecatory urge allow the expulsion of variable amounts of colorectal contents. Some other factors, such as voluntary straining and appropriate defecation posture, may influence this phase. The Valsalva maneuver that contracts the diaphragm and the abdominal muscles and relaxes the external anal sphincter helps defecation through an increase of intrarectal and intrapelvic pressures (Petros and Swash 2008). Defecation thus results by a combination of colonic propulsive activity (almost all episodes of defecation are associated with HAPC (Bampton et al. 2000; Herbst et al. 1997; Narducci et al. 1987)), increased intrarectal pressure, of pelvic floor activity inhibition with relaxation of the puborectalis muscle and straightening of the anorectal angle, and relaxation of the anal sphincter (Palit et al. 2012) (Fig. 5.4). The simultaneous occurring of the above phenomena decrease the pressure of the anal canal below that of the rectum, resulting in a gradient toward the external environment; once expulsion starts, anal sensory inputs maintain the propulsion until the rectum is empty (Lynch et al. 2000; McCrea et al. 2008).

Termination of defecation. A sensation of complete rectal emptying with interruption of the maneuvers that increase intrapelvic pressure begins this phase that is partially voluntary, followed by involuntary contraction of the external anal sphincter and of the pelvic floor muscles. Thus, after straining is terminated, there is restoration of the anorectal angle, closure of the anal canal (strengthened by passive distention of the

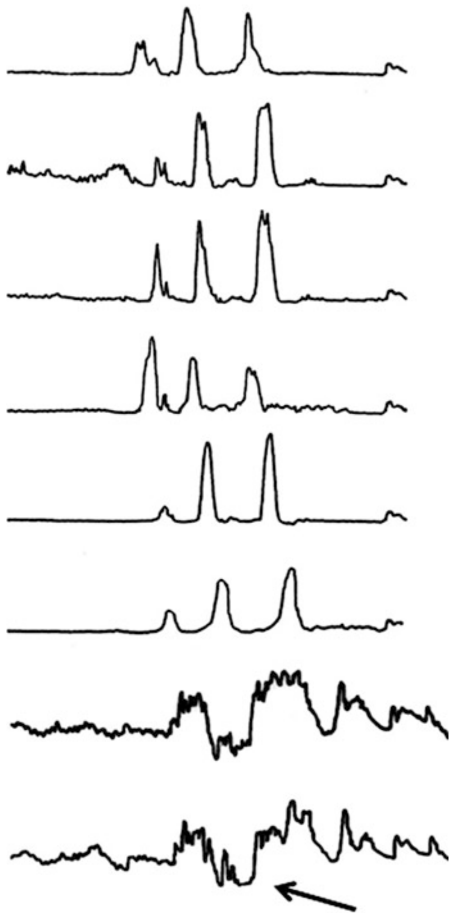

Fig. 5.4 Manometric recording of defecation. Note the propagated high-amplitude sequences in the first six recording points, and the relaxation of the anal sphincter (*arrow*) in the last two recording points

anal cushions), and reversal of the pressure gradient toward the rectal ampulla (Bajwa and Emmanuel 2009).

In conclusion, colonic physiology is based on a series of more or less complex phenomena, all having a common terminal purpose, i.e., that of expelling in an effective manner the residuals of the digestive processes. Although described as separate phenomena, one should always keep in mind that these various functions (and especially those related to the motor activity) integrate each other, and are also under the influence of higher cerebral centers, in order to serve as an effective unit to carry on their physiological meaning.

4 Cross-References

▶ Anorectal Manometry
▶ Factors Affecting Defecation and Anal Continence
▶ Factors Affecting the Intestinal Physiology
▶ Physiology of the Rectum and Anus

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Physiology of the Rectum and Anus

6

Richelle J. F. Felt-Bersma

Contents

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Abstract

The anorectal area is a complex area where autonomic and somatic structures coordinate to maintain continence and to regulate defecation. The fecal bolus entering the rectum is perceived, and by a reflex mechanism, the internal anal sphincter relaxes and allows the content to enter the proximal anal canal, where depending on content and situation conscious relaxation or contraction of the external anal sphincter and puborectal muscle will follow.

With anal endosonography or MRI, the integrity of the sphincters and pelvic floor can be established. The physiology of the anus and rectum can be tested with anal manometry. This allows measuring of the anal resting and squeeze pressure. Furthermore, relaxation during attempted defecation and the distention reflex can be observed. The process of defecation can be observed with defecography.

1 Introduction

The rectum and the anus are at the very end of the digestive tract. Although they do not play a role in digestion and resorption, they have an important function in maintaining fecal continence and regulating defecation. The anatomy of the anorectal area is rather unique; there is coordination between smooth muscle with autonomic innervation and striated muscle with somatic innervation.

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2 Embryology

The primitive gut is formed during the 3rd week of gestation. The anorectal area in humans is derived from four separate embryological structures: the hindgut, the cloaca, the proctodeum, and the anal canal tubercles (Gordon and Nivatvongs 1992; Barucha and Blandon 2007). The hindgut forms the distal third of the transverse colon up to the upper part of the anal canal to the level of the anal valves. The end of the hindgut enters into the cloaca, an entoderm lined cavity what is in direct contact with the surface ectoderm. The cloaca is originally a single tube that is subsequently separated by caudad migration of the urorectal septum into anterior urogenital and posterior intestinal (rectal) outflow. During the 10th week of gestation, the external anal sphincter is formed from the posterior cloaca when the urogenital septum is completed. In the 12th week, the internal anal sphincter is formed from a thickened extension of the circular rectal muscle. The distal portion of the cloacal membrane disintegrates to form the anal tubercules that join posteriorly and migrate ventrally to form a depression, known as the anal dimple or proctodeum. Subsequently, these anal tubercles join the urorectal septum and genital tubercles to form the perineal body, thus separating the rectum and the urogenital tract.

3 Anatomy

The anatomy of the rectum and especially the anus can be viewed perfectly with anal endosonography or MRI (see ▶ Chap. 16, "Colonic and Rectal Endosonography")

3.1 Rectum

The rectum is the terminal portion of the large intestine beginning at the confluence of the three tenia coli of the sigmoid colon and ending at the anal canal. Generally, the rectum is 15–20 cm in length, is intraperitoneal at its proximal and anterior end, and is extraperitoneal at its distal and posterior end. The epithelial lining or mucosa of the rectum is of a simple columnar mucous secreting variety. The rectal wall as a part of the colon, histologically from lumen outward consists of a simple columnar epithelium which forms crypts, lamina propria, muscularis mucosa, submucosa, and muscularis propria formed by an inner circular and outer longitudinal layer of smooth muscle, and serosa.

The volume of a normal rectum varies and lies between 120 and 250 ml (Felt-Bersma et al. 2000)

3.2 Anus

The adult anal canal is approximately 3–5 cm long and begins as the rectum narrows, passing backward between the levator ani muscles. Men have generally a longer anal canal, especially anteriorly. There is also large range of variation in length between individuals of the same sex. The canal has an upper limit at the pelvic floor and a lower limit at the anal opening. The proximal canal is lined by simple columnar epithelium, changing to stratified squamous epithelium lower in the canal via an intermediate transition zone just above the dentate line. Beneath the mucosa is the subepithelial tissue, composed of connective tissue and smooth muscle (submucosal plexus or Meissner's plexus). This layer increases in thickness throughout life and forms the basis of the vascular cushions thought to aid continence (Figs. 6.1 and 6.2).

Lateral to the subepithelial layer, the caudal continuation of the circular smooth muscle of the rectum thickens and forms the internal anal sphincter (with its innervation from the myeneteric plexus or Auerbach plexus), which terminates caudally with a well-defined border at a variable distance from the anal verge. Continuous with the outer layer of the rectum the longitudinal layer of the anal canal lies between the internal and external anal sphincters and forms the medial edge of the intersphincteric space. The longitudinal muscle comprises smooth muscle cells from the rectal wall, augmented with striated muscle from a variety of sources, including the levator ani, puborectalis, and

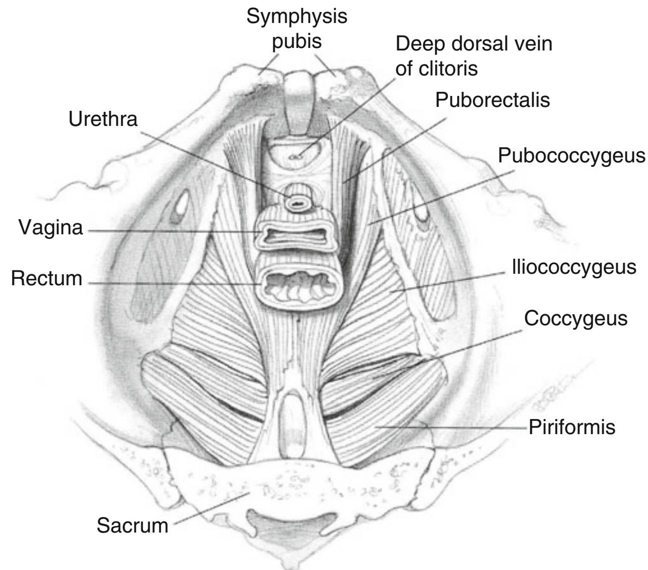

Fig. 6.1 Pelvic view of the levator ani demonstrating its three main components: puborectalis, pubococcygeus, iliococcygeus. Reprinted with permission from (Canda et al. 2010)

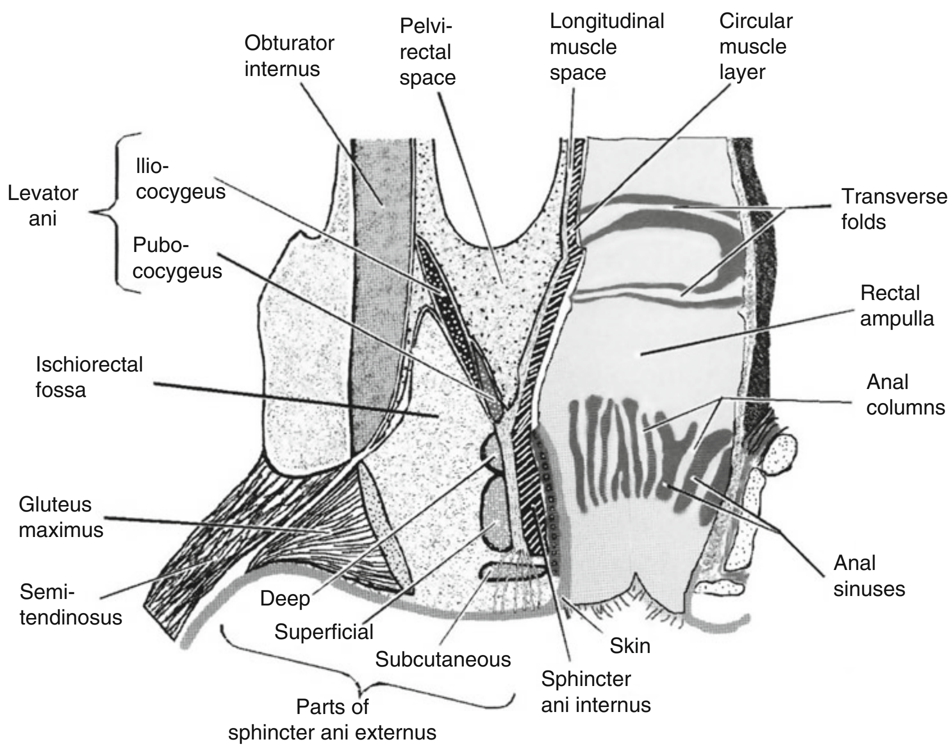

Fig. 6.2 Diagram of a coronal section of the rectum, anal canal, and adjacent structures. The pelvic barrier includes the anal sphincter complex and the pelvic floor muscles. Reprinted with permission from (Canda et al. 2010)

pubococcygeus muscles. Fibers from this layer traverse the external anal sphincter forming septa that insert into the skin of the lower anal canal and adjacent perineum as the corrugator cutis ani muscle.

The striated muscle of the external sphincter surrounds the longitudinal muscle and between these lays the intersphincteric space. The external sphincter is arranged as three supposed structure, originally described by Holl and Thompson and later adopted by Gorsch and by Milligan and Morgan. In this system, the external sphincter is divided into deep, superficial, and subcutaneous portions, with the deep and subcutaneous sphincter forming rings of muscle and, between them, the elliptical fibers of the superficial sphincter running anteriorly from the perineal body to the coccyx posteriorly. Some consider the external sphincter to be a single muscle contiguous with the puborectalis muscle, while others have adopted a two-part model. The latter proposes a deep anal sphincter and a superficial anal sphincter, corresponding to the puborectalis and deep external anal sphincter combined, as well as the fused superficial and subcutaneous sphincter of the tripartite model. Anal endosonography and magnetic resonance imaging have not resolved the dilemma, although most authors report a three-part sphincter where the puborectalis muscle is fused with the deep sphincter.

The blood supply to the anorectal region is rich. The terminal branch of the inferior mesenteric artery is the superior hemorrhoidal (rectal) artery. The superior hemorrhoidal artery branches into right and left branches; the right branch further divides into anterior and posterior branches. The classic hemorrhoidal plexes are then located at the left later, right anterolateral and right posterolateral locations. The middle hemorrhoidal (rectal) arteries are direct branches from the internal iliac arteries. The inferior hemorrhoidal (rectal) arteries are branches off the pudendal arteries which also arise from the internal iliac arteries. The superior, middle, and inferior hemorrhoidal arteries then complete the rich arterial supply to the anorectal region.

The venous drainage of the anorectal region consists of superior hemorrhoidal veins draining into the portal venous system (by way of the inferior mesenteric vein) and the middle and inferior hemorrhoidal veins draining into the caval system (by way of the internal iliac veins). Thus the anorectal region can provide a means of portal decompression when portal hypertension exists.

Lymphatic drainage of the rectum travels along the internal iliac vessels as well as the aorta. Lymphatic drainage of the anal canal can follow the internal iliac vessels but also may travel through channels in the inguinal region.

The nerve supply is complex. The external anal sphincter is innervated by the pudendal nerve (S2–S4), which leaves the pelvis via the lower part of the greater sciatic notch, where it passes under the pyriformis muscle. It then crosses the ischial spine and sacrospinous ligament to enter the ischiorectal fossa through the lesser sciatic notch or foramen via the pudendal (or Alcock's) canal.

The pudendal nerve has two branches: the inferior rectal nerve, which supplies the external anal sphincter and sensation to the perianal skin; and the perineal nerve, which innervates the anterior perineal muscles together with the sphincter urethrae and forms the dorsal nerve of the clitoris (penis). Although the puborectalis receives its main innervation from a direct branch of the fourth sacral nerve root, it may derive some innervation via the pudendal nerve.

The autonomic supply to the anal canal and pelvic floor comes from two sources. The fifth lumbar nerve root sends sympathetic fibers to the superior and inferior hypogastric plexuses, and the parasympathetic supply is from the second to fourth sacral nerve roots via the nervi erigentes. Fibers of both systems pass obliquely across the lateral surface of the lower rectum to reach the region of the perineal body.

The internal anal sphincter has an intrinsic nerve supply from the myenteric plexus together with an additional supply from both the sympathetic and parasympathetic nervous systems. Sympathetic nervous activity is thought to enhance and parasympathetic activity to reduce internal sphincter contraction. Relaxation of the internal anal sphincter may be mediated via non-adrenergic, non-cholinergic nerve activity

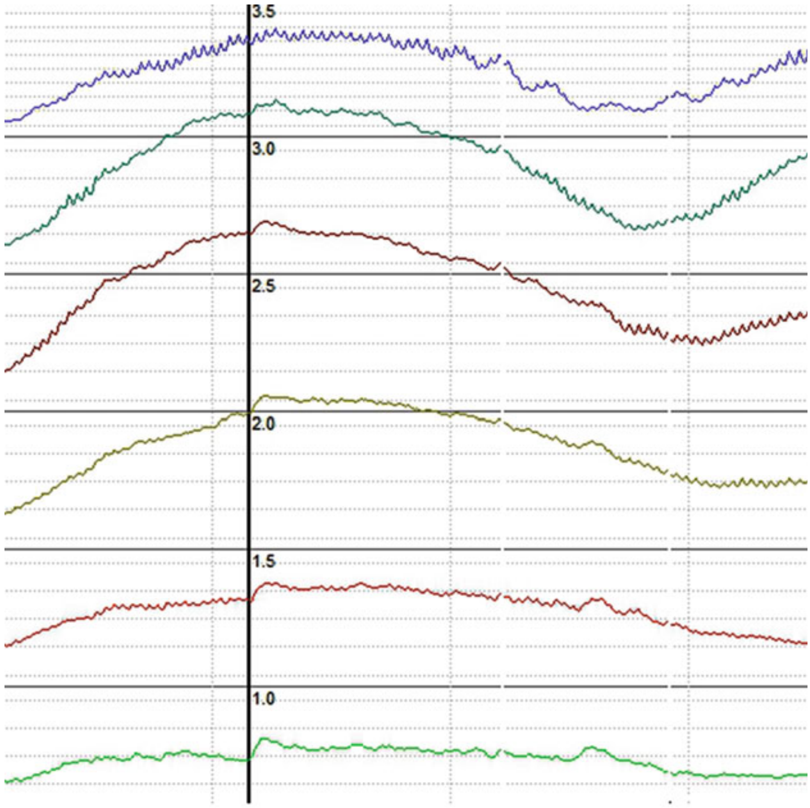

Fig. 6.3 Anal manometry recording of slow waves. Note the high basal pressure

via the neural transmitter nitric oxide. The role of the interstitial cells of Cajal (ICC) is not elucidated yet. Animal studies suggest that intramuscular ICC in the IAS may serve as pacemaker cells rather than as mediators of neuromuscular transmission. A recent study in patients undergoing abdominoperineal resection or proctectomy suggested that the ICC modulate the tone and the spontaneous activity of the internal anal sphincter (Lorenzi et al. 2012).

Anorectal physiological studies alone cannot separate the different structures of the anal canal; instead, they provide measurements of the resting and squeeze pressures along the canal pudendal block demonstrated the basal pressure to be generated for 85 % by the internal and for 15 % by the external anal sphincter (Frenckner and von Euler 1975). Later, it was shown that the basal pressure is composed for 30 % of tonic external sphincter activity, for 45 % of nerve induced internal sphincter activity, for 10 % of pure myogenic internal sphincter activity, and for 15 % of expansion of the hemorroidal plexuses (Lestar et al. 1989). The resting pressure undergoes regular fluctuations. These consist of slow waves (amplitude 5–25 cm H_2O, frequency between 10/min and 20/min) and much larger amplitude, ultra slow waves (amplitude 30–100 cm H_2O, frequency <3/min) (Kerremans 1969). Ultra slow waves (Fig. 6.3) are associated with high resting pressures.

The external anal sphincter and the puborectalis muscle generate the maximal squeeze pressure. Symptoms of passive anal leakage (where the patient is unaware that episodes are happening) are attributed to internal sphincter dysfunction, whereas urge symptoms and frank incontinence of feces are due to external sphincter problems.

Fecal continence is maintained by the complex interaction of many different variables. Stool must be delivered at a suitable rate from the colon into a compliant rectum of adequate volume. The consistency of this stool should be appropriate and accurately sensed by the sampling mechanism.

Sphincters should be intact and able to contract adequately to produce pressures sufficient to prevent leakage of flatus, liquid and solid stool. For effective defecation there needs to be coordinated relaxation of the striated muscle components with an increase in intra-abdominal pressure to expel the rectal contents. The structure of the anorectal region should prevent herniation or prolapse of elements of the anal canal and rectum during defecation.

As a result of the complex interplay between the factors involved in continence and fecal evacuation, a wide range of investigations is needed for full assessment. A defect in any one element of the system in isolation is unlikely to have great functional significance and so in most clinical situations there is more than one contributing factor.

4 Physiology of Continence and Defecation

The function of rectum and anus is an integrated action and coordination of several structures.

Arrival of stool in the rectum causes rectal distension and induces a desire to defecate along with a decrease in anal resting pressure (the rectoanal inhibitory reflex).

This allows the rectal contents to come into contact with the sensitive anoderm, and based on the amount and nature of fecal material "sampled," solid, liquid, or gas, an urge to defecate is induced that can only be resisted by vigorous contractions of the EAS and puborectalis muscle. If social conditions are favorable, the subject sits or squats, holds breath, contracts the diaphragm, abdominal, and rectal muscles, and simultaneously relaxes the EAS and puborectalis muscle. These maneuvers open the anus and move stool. Thus, sensory perception and coordinated movement of stool are important physiologic variables that affect anorectal function.

4.1 Anal Resting Pressure

The resting pressure prevents leaking from fecal material and fluid from the rectum and anus. This can be disturbed by direct damage to the internal sphincter by trauma like forceful dilatation (Speakman et al. 1991; Felt-Bersma et al. 1995), in grade 3c sphincter rupture during childbirth (Visscher et al. 2014), surgery (Felt-Bersma et al. 1995; Johannsson et al. 2013; Hirano et al. 2011; Lindsey et al. 2004), radiation (Canda et al. 2010) as well as scleroderma (Thoua et al. 2012), neuropathy due to autonomic dysfunction like diabetes (Pinna Pintor et al. 1994; Rogers et al. 1988), multiple sclerosis (MS) (Nusrat et al. 2012), Parkinson's disease (Kim et al. 2011; Stocchi et al. 2000), or idiopathic (Ricciardi et al. 2006; Felt-Bersma et al. 1992). Isolated insufficiency of the internal sphincter will rather lead to fecal soiling than overt fecal incontinence, since the pelvic floor and external anal sphincter can compensate.

Anal resting pressure can be established with digital palpation, which correlates well with anal manometry (Orkin et al. 2010; Felt-Bersma et al. 1988) (see ► Chap. 19, "Anorectal Manometry"). With high resolution anal manometry (HRAM), a more detailed image can be obtained throughout the sphincter (Fig. 6.4).

4.2 Rectoanal Inhibitory Reflex

Increasing rectal distension is associated with transient reflex relaxation of the internal anal sphincter and contraction of the external anal sphincter, known as the rectoanal inhibitory or distention reflex. This can be demonstrated with anorectal manometry and was originally described by Schuster (Schuster et al. 1965). With (high resolution) anal manometry (HRAM), it can be shown that the more air is inflated in the balloon, the more profound the anal pressure drops. Furthermore, the level of the largest drop is in the middle of the anal canal, where the internal sphincter is at its maximum, can be seen (Fig. 6.5). This correlates very well with the anatomical image, which can be seen with anal ultrasound (Fig. 6.6)

The exact neurological pathway for this reflex is unknown, although it may be mediated via intrinsic nerves of the myenteric plexus and stretch receptors in the pelvic floor. Its existence

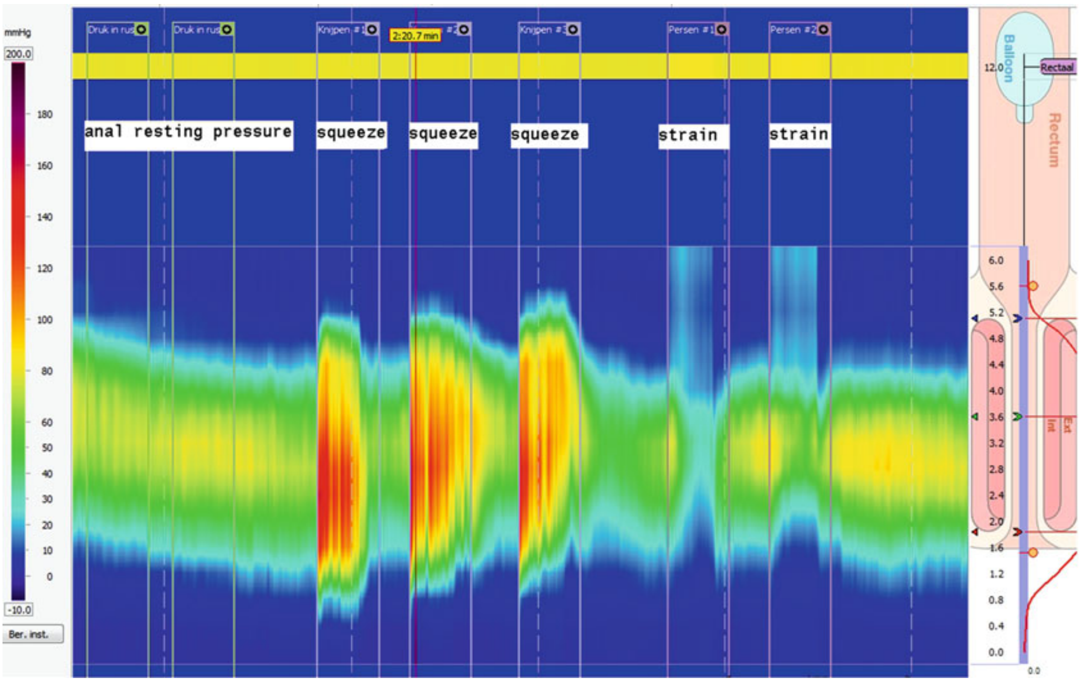

Fig. 6.4 Normal 3D HRAM with anal resting pressure, increase during squeeze and relaxation during strain

depends on intramural autonomic ganglions and its modulation on the integrity of the autonomic nervous system. Patients with rectal hyposensitivity have higher thresholds for rectoanal inhibitory reflex; it is absent in patients with Hirschsprung's disease, progressive systemic sclerosis, Chagas' disease, and initially absent after a coloanal anastomosis, although it often recovers. When anal pressures are low, the reflex can also not be elucidated.

4.3 Contraction of the External Anal Sphincter and Pelvic Floor

Fecal continence is maintained by an adequate basic tone of the pelvic floor and anal sphincter. When this is insufficient, pressure of the stool in the rectum can easily overcome the pressures generated by the internal and external anal sphincter. Causes for external anal sphincter dysfunction can be both muscular and neurological. Trauma of the anal sphincters during childbirth, even unnoticed, can be an important reason. Anorectal surgery, especially surgery for perianal fistula, is a notorious cause (Visscher et al. 2014). The most frequent (peripheral) neurological cause is stretch damage of the pudendal nerve resulting in atrophy of the external sphincter and to a lesser extend of the puborectal muscle. This can occur during chronic straining with childbirth (Snooks et al. 1986) and chronic constipation (Snooks et al. 1985), as was demonstrated in the eighties of the previous century. Another cause of peripheral neuropathy is diabetes (Pinna Pintor et al. 1994; Rogers et al. 1988).

In (central) neurological diseases, like MS (Nusrat et al. 2012), Parkinson's disease (Kim et al. 2011; Stocchi et al. 2000), and spinal cord injury (Valles et al. 2006), the external anal sphincter and pelvic floor may also be involved. A good impression of the contraction of the external anal sphincter can be obtained with digital rectal examination compared to anal manometry and has been confirmed throughout the years (Felt-Bersma et al. 1988, 1992). With anal manometry, the increase in pressure above the anal basal pressure is generally defined as the

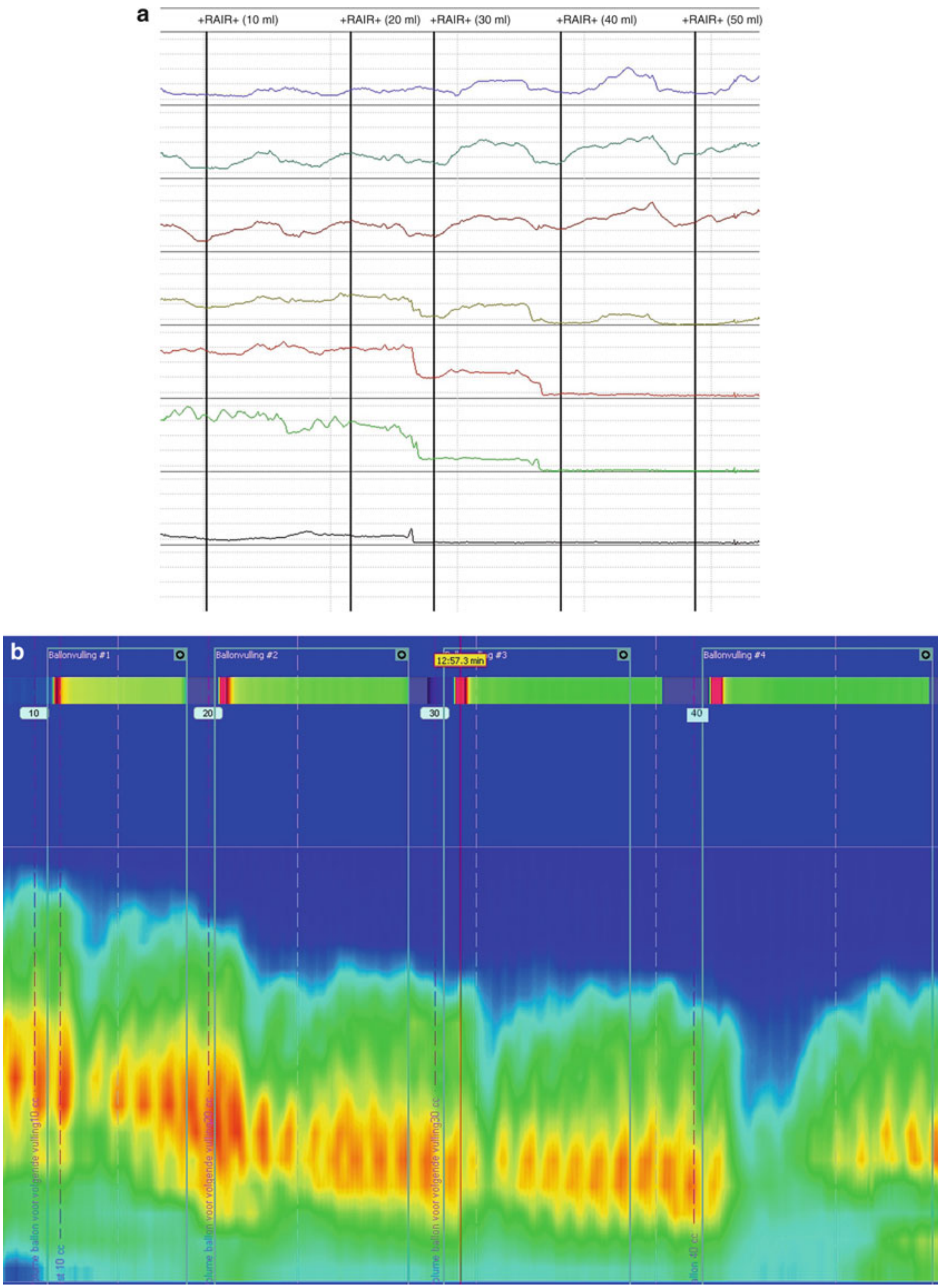

Fig. 6.5 (**a**) Conventional and (**b**) 3D-HR anal manometry. Inhibition reflex. A fast drop in resting pressure, followed by a slow recovery. Increasing the volume of distention from 10 to 40 ml gives a pore profound relaxation

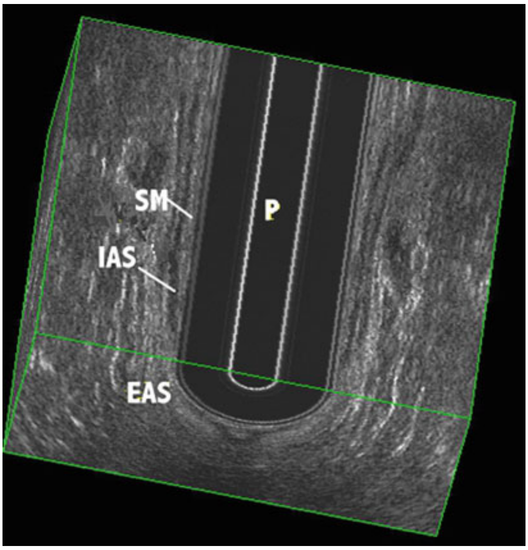

Fig. 6.6 3D anal ultrasound of the anal sphincter, frontal view. The external anal sphincter (*EAS*) is longer and runs more distal then the internal anal sphincter (*IAS*)

squeeze pressure. With HRAM, the functional anatomy becomes clear, since the more distal, where the bulk of the external sphincter is present, the highest pressures are found (Fig. 6.4). A problem is the large range of normal values of all anal pressures and the large overlap in patients with fecal incontinence (Felt-Bersma et al. 1990; Gundling et al. 2010). Generally, normal values are established in healthy persons without anorectal complaints or previous surgery in that area. The problem comes that for instance fecal incontinence has several causes, like thin stools, small rectal capacity, and incompetent sphincters. But complaints for instance of fecal incontinence can be masked by relatively hard stools, thus including lower pressures as normal values. Lam (Lam et al. 2012) et al. suggested a model for predicting fecal incontinence with introducing these factors as well as sex and age. With the HRAM (Lee et al. 2014) and the 3D HRAM (Li et al. 2013), new studies with new normal values are published. Depending on the device and technique, normal values may differ. All studies agree that men have higher pressures then women and aging decrease pressures. The effect of parity differs largely and is due to differences is methodology and sample size.

4.4 Relaxation of the External Anal Sphincter

For defecation, the external anal sphincter and pelvic floor need to relax. Generally, this will be occurring automatically, but under certain circumstances this can be difficult or sometimes impossible. This phenomenon is called dyssynergia. Originally this was thought to be a variant of vaginismus, where psychological factors play a role. Indeed sexual abuse and psychological factors play often a role, but also wrong habits can induce that. For instance, if a patient has poor rectosigmoid motility with hard stools or a large rectocele, secondary contra productive maneuvres during defecation may become a wrong habit. Furthermore, when passage of stools causes pain due to a fissure, relaxation can be difficult. Relaxation of the pelvic floor can be established during rectal examination (Tantiphlachiva et al. 2010) (Fig. 6.7). With HRAM or manometry, this can be confirmed (Ratuapli et al. 2013) (Fig. 6.8a, b). Instead of a drop in resting pressure, no change or even an increase can be seen (Fig. 6.8c). Another means of testing is the rectal balloon expulsion test, where the inability to expel the balloon within 2 min is considered to be abnormal (Chiarioni et al. 2014). Defecography has to demonstrate the inability to relax has given great insight, but should not be used anymore to confirming this defecation disorder (Videlock et al. 2013; Minguez et al. 2004).

4.5 Rectal Compliance, Capacity, and Sensitivity

The rectal compliance and rectal capacity to store the stools is an important factor to maintain continence. A certain reservoir needs to be present to avoid frequent defecation and incontinence. Several techniques to measure rectal capacity, compliance, and sensitivity are available. Generally, a rectal latex balloon is slowly filled with air or water, while the patient indicates the first sensation, the urge to defecate and the maximum tolerance volume (MTV). If the rectal balloon catheter contains a pressure tip, the rectal

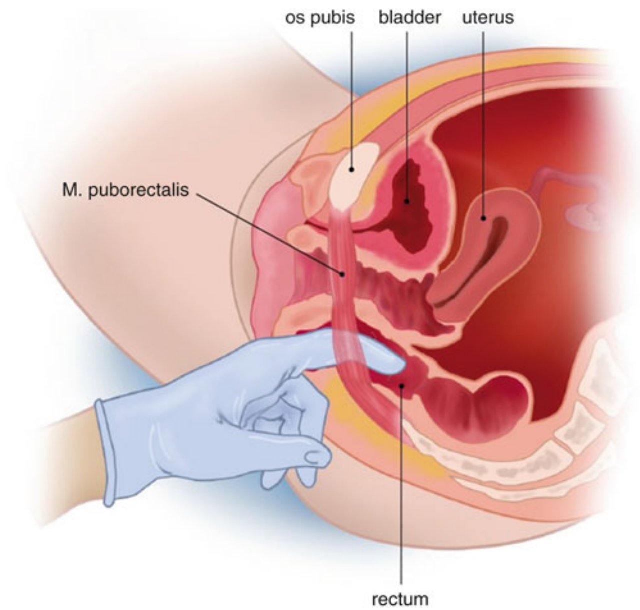

Fig. 6.7 Examination of the puborectal muscle

compliance can be calculated (dV/dp) (Fig. 6.9). Another means of measuring the characteristics of the rectum is the barostat. With this technique, a polyethylene, non-compliant bag is introduced into the rectum, and subsequent inflation with air and registering pressures and pain scores can obtain detailed information. The advantage of the barostat procedure is that the characteristics of the rectal bag do not interfere with the measurements. Rectal volumes measured with the latex balloon are smaller than with the polyethylene balloon (Sloots et al. 2000; Gladman et al. 2005).

A small MTV leads to fecal incontinence, a MTV between 60 and 100 ml leads to fecal incontinence in 63 % and a MTV <60 ml inevitably renders patients incontinent (Felt-Bersma et al. 2000). Again, large overlap between controls and patients with fecal incontinence or constipation exists (Felt-Bersma et al. 1990; Gundling et al. 2010).

Patients with proctitis regardless of its cause, inflammatory bowel disease or radiation, have a smaller rectal capacity (Felt-Bersma et al. 2000). In male patients after radiation for prostate cancer maximum tolerable volume decreased from 277 to 227 ml ($p < 0.001$) (Krol et al. 2012), another study in male patients with prostate or bladder cancer the maximum tolerable volume decreased from 245 ml to 200 ml ($p < 0.05$) (Kushwaha et al. 2003). In patients with irritable bowel syndrome (IBS), rectal sensation and MTV may also be smaller. This is not so much due to a decreased compliance of the bowel but an increased sensitivity for stimuli (Ludidi et al. 2012).

In patients with hyposensitivity of the rectum, the urge to defecate will comes only after larger amounts of feces have entered the rectum, so that large amounts of stool are necessary to obtain that urge, thus increasing the size of the rectum and thus decreasing the hyposensitivity further. Probably the most important cause of hyposensitivity is constipation and evacuation problems with dyssynergia, thus by stretching the rectum further deteriorating rectal sensitivity (Lee et al. 2013; Lam and Felt-Bersma 2013). Other causes of hyposensibility are the same neurological disorders as mentioned previously with internal and external sphincter dysfunction.

Regular emptying of the rectum can reverse a large rectum that partly as was shown in a study in elderly receiving daily enema's to remove the stools (Read et al. 1985). Large rectums are solely found in patients with constipation (Felt-Bersma et al. 2000).

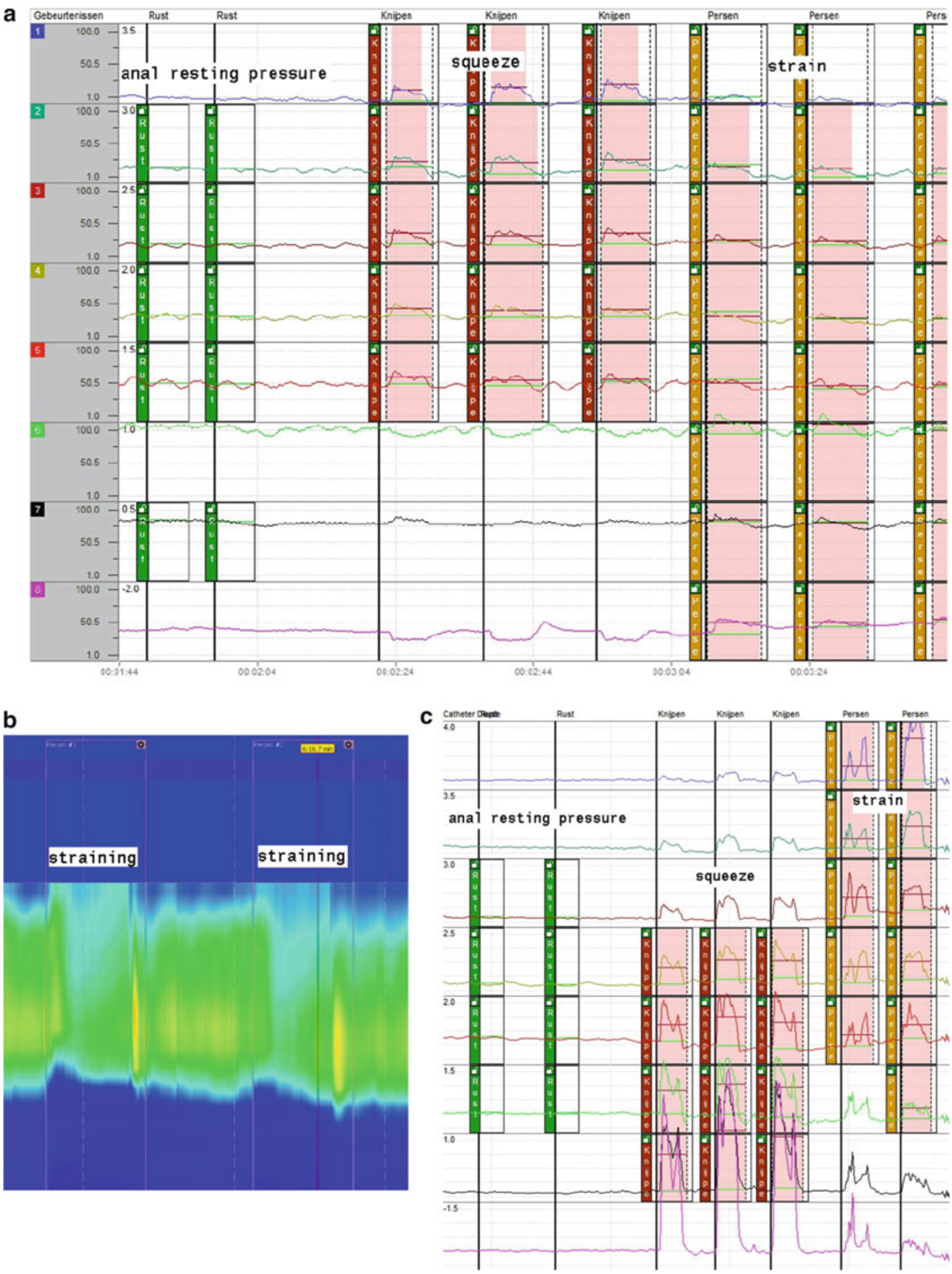

Fig. 6.8 (**a**) Normal relaxation during straining conventional manometry and (**b**) 3D HRAM. (**c**) paradoxal increase during straining

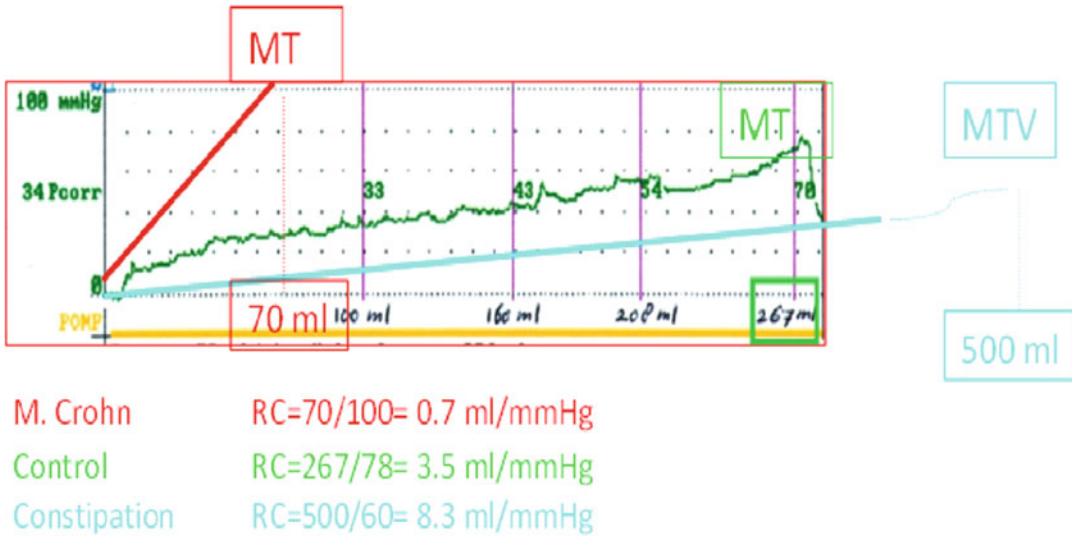

Fig. 6.9 Rectal compliance. On the x-axis the volume and on the y axis the balloon pressure. *MT* maximal tolerable point. Patients with a stiff rectum have a low compliance (dp/dV), controls a normal compliance and constipated patient a high compliance

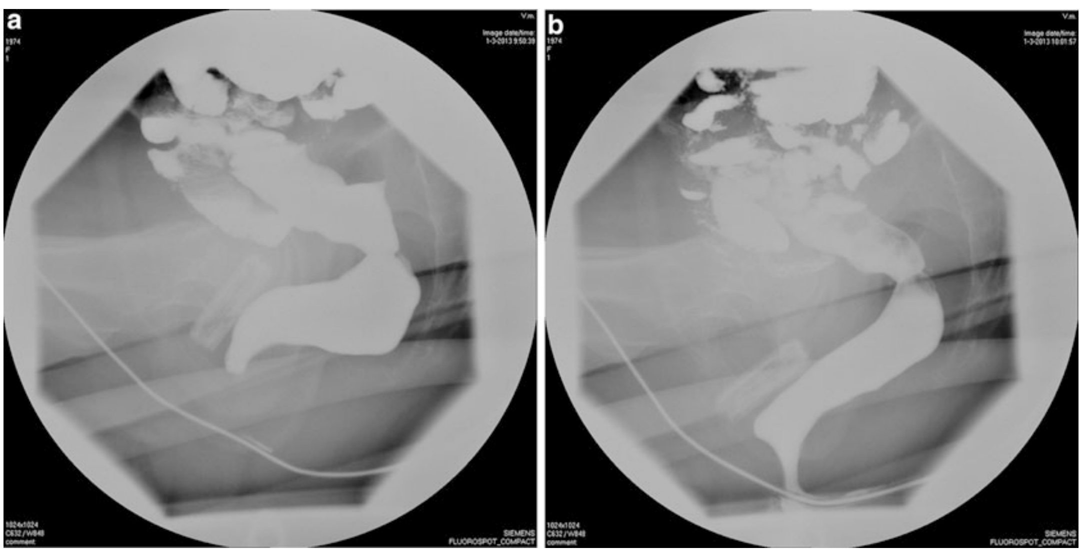

Fig. 6.10 Defecography, normal image in (**a**) rest and (**b**) during straining

4.6 Defecography: Visualization of Defecation

The physiology of defecation has been studied by means of defecography, the technique is described elsewhere (see ► Chap. 17, "Radiologic Imaging of Colo-Recto-Anal Dysfunctions: Procedures and Indications"). With this technique the contour of the rectum, the rectovaginal septum and the small bowel and their movements during contraction and attempted defecation can be seen. This technique has provided great insight in the process of normal and abnormal defecation. Many contour abnormalities can be found in healthy volunteers (Palit et al. 2014; Shorvon et al. 1989), which questions the clinical relevance of these findings in patients. In normal defecation, the puborectal sling relaxes, the anorectal angle obliterates and the rectum empties in 2 min with hardly any contrast remaining (Fig. 6.10). Small rectoceles are considered normal in parous women. With forceful straining, a rectal intussusception can be seen, which does not necessarily is abnormal. However, less straining is a better way to defecate, since longstanding straining is associated with pudendal dysfunction, leading to fecal incontinence (Snooks et al. 1985). In patients with dyssynergia, the puborectal sling does not relax and the anorectal angle does not obliterate. Sometimes rectal intussusceptions can be found.

The inability to defecate and rectoceles can be established during physical examination and confirmed with anal manometry, so the indication for defecography is reduced to suspicion of enteroceles and internal descent with intussusception and should therefore be performed only if surgery is a potential option in patients. Studies are emerging using perineal ultrasound defecograpy to demonstrate abnormalities; prescreening for conventional defecography could be an option (Martellucci and Naldini 2011).

Since many abnormalities during defecation are related to excessive training, defecation rehabilitation, and training should be the first choice and surgery should be reserved for selected cases, with over abnormalities, like large descent of the pelvic floor.

5 Cross-References

► Anorectal Manometry
► Colonic and Rectal Endosonography
► Radiologic Imaging of Colo-Recto-Anal Dysfunctions: Procedures and Indications

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Factors Affecting the Intestinal Physiology

7

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Contents

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Abstract

The main functions of the colorectum are transport and storage of feces and absorption of water, electrolytes, and short-chain fatty acids. The main functions of the anal canal are to prevent involuntary defecation while opening during defecation at an appropriate time and place. Colorectal or anal function is affected by several physiological factors including diet, exercise, sleep, age, hormonal status, and childbirth. Abnormal colorectal or anal function is caused by or associated with neurological disease, connective tissue disease, diabetes, bile acid malabsorption, surgery, irradiation therapy, inflammation, various types of commonly used medication, or psychiatric disease. In a large proportion of patients, symptoms of colorectal and anal dysfunction remain idiopathic.

1 Introduction

The colorectum has three main functions: (a) transport and storage of feces, (b) absorption of water and electrolytes, and (c) absorption of short-chain fatty acids. The two main functions of the anal canal are (a) to remain closed between voluntary defecation and (b) to open at defecation. A number of conditions may interfere with these basic functions and thereby cause symptoms.

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2 Basic Colorectal Physiology

The smooth muscle cells of the colorectum are arranged in bundles connected by gap junctions making each muscle layer function as a syncytium. The resting membrane potential of −50 to −60 mV is not constant but undergoes undulating changes called slow waves generated by the interstitial cells of Cajal. **Spike potentials** are action potentials occurring when the resting membrane potential becomes more positive than −40 mV. During spike potentials, contractions occur because calcium enters the smooth muscle cells. Several factors influence the occurrence of spike potentials, either by depolarization making the membrane potential more positive and thus the cells more excitable or by hyperpolarization making it more negative and the cells less excitable. Depolarization of the membrane potential makes the cells more excitable and thus contractions more likely. Depolarization is stimulated by stretch of the muscle cells, acetylcholine, and several gastrointestinal hormones. Hyperpolarization making the cells less excitable is caused by epinephrine and norepinephrine.

Colorectal contractions are generated by coordinated contractions of the smooth muscle cells within the bowel wall. Two main types of colorectal contractions exist: Single nonpropagating (or haustral) contractions and **high-amplitude propagating contractions**. **Haustral contractions** are located in a short segment of the colonic wall, and their main function is to mix colonic contents thereby promoting the absorption of water, salts, and short-chain fatty acids. High-amplitude propagating contractions are the physiological basis for colonic mass movements and appear a few times per day. They span large parts of the colon, thereby moving stools toward the anorectum. **Colonic mass movements** usually appear in the morning or after meals as part of the gastrocolonic response (Dinning et al. 2014).

Bowel contents passing the ileocecal valve are semifluid. Usually, 1,500–2,000 ml passes to the colon every day. During the passage of the colon, most of the water is absorbed typically leaving 100–150 mg of stools to be passed per day. Furthermore, the colon has the capacity to absorb additionally 6,000 ml per day. The colon absorbs sodium and chloride while it secretes potassium and bicarbonate. Most absorption of water takes place in the right colon while the main function of the left colon is storage of feces.

Short-chain fatty acids (SCFAs) are produced by anaerobic bacterial fermentation of dietary fiber, mainly in the right colon. SCFAs are absorbed by the mucosa and provide a major source of energy for the colonic mucosa cells.

3 Basic Anorectal Physiology

Between defecations or flatus, the rectum is empty and the anus closed. Most of the time, the anal canal is kept closed by the tone of the internal and, to a lesser extent, the external anal sphincter muscles. The tone of puborectalis muscle creates the rectoanal angle which acts as a valve blocking passage of rectal contents to the anal canal.

The mucosa of the anal canal is densely innervated by mechanoreceptors. Stool or air within the lumen of the anal canal is sensed by these, allowing the subject to contract the external anal sphincter muscle if appropriate.

Defecation is usually preceded by a colonic mass movement moving stools into the rectum. Stretch of the rectal wall will cause reflex contraction of the rectosigmoid and relaxation of both the puborectalis and internal anal sphincter muscle. The latter is termed the **rectoanal inhibitory response**. Rectal distension is sensed by the subject who may support defecation by assuming an appropriate sitting position and perform a Valsalva manoeuver. Under normal circumstances, defecation includes emptying of almost the whole rectosigmoid.

4 Diurnal and Postprandial Changes

Colorectal motility is generally suppressed during **sleep** and motility is increasingly suppressed with increasing depth of sleep (Furukawa et al. 1994). Thus, colonic mass movements are almost absent during the night but reappear upon awakening. Rhythmic rectal contractions, termed the **rectal motor complex**, are usually localized or propagating aborally. They are most common during the night and probably serve to keep the rectum empty and avoid fecal incontinence during sleep.

Within few minutes after a meal, colorectal tone increases, and colonic mass movements increase in number in the postprandial period. This is termed the ***gastrocolic respon***se. Changes are most prominent in the left colon and rectum and mediated by both sympathetic nerve fibers and by the release of cholecystokinin and, perhaps, gastrin. Meals rich in lipids are especially strong stimulants of the gastrocolic response which usually lasts 30–60 min.

5 Diet and Colorectal Function

Diet and fluid intake have significant effects on colorectal function. **Dietary fibers** are resistant to hydrolysis by small intestinal enzymes. They pass unabsorbed to the colon, where they retain water and add bulk to the stools. Increased intake of fiber, either as part of the diet or as supplement, can reduce colonic transit time and alleviate symptoms of constipation. Side effects from dietary fiber mainly include formation of gas causing abdominal discomfort and flatulence. Production of gas is mainly observed with short water-soluble fibers. Unless the subject is dehydrated, moderate increase of water intake does not per se affect colorectal function or reduce symptoms of constipation. An intake of 2 l of water will however enhance the positive effects of dietary fiber on constipation-related symptoms (Anti et al. 1998).

In recent years, poorly absorbed short-chain carbonhydrates have received increasing attention. These low-fermentable oligosaccharides, disaccharides, monosaccharides, and polyols (**FODMAPs**) are part of the modern daily diet of many people. In some subjects, especially patients with irritable bowel syndrome, they may worsen diarrhea, abdominal pain, and bloating (Halmos et al. 2014). The role of low-FODMAP diet as treatment of irritable bowel syndrome is still not established.

6 Physical Activity and Colorectal Function

Constipation is strongly associated with immobility. In healthy subjects, **physical activity** stimulates colorectal motility thereby reducing colonic transit time (Oettlé 1991). In patients with chronic idiopathic constipation, moderate physical activity 30–60 min per day will also improve stool consistency (De Schryver et al. 2005). Extreme sports often cause diarrhea and, in rare cases, intestinal hypoperfusion with abdominal pain and rectal bleeding.

7 Age and Colorectal Function

The prevalence of constipation increases significantly with **age**. It is however disputed to what extent this is caused by age per se or whether the main reasons are comorbidity and increased use of medication associated with age. There is evidence of age-related degeneration of the enteric nervous system, but a study comparing healthy young and healthy elderly men found no differences in rectosigmoid motility. Likewise, most colorectal transit times are within normal range in healthy elderly people.

The prevalence of **fecal incontinence** increases with age from approximately 3 % in those aged from 20 to 29 years to 16 % in persons aged 70 or more (Ditah et al. 2014). In nursing home residents, the prevalence may be as high as 47 %. Idiopathic fecal incontinence is associated with a weakened pelvic floor as indicated by reduced anal pressures and increased distensibility of the anal canal.

8 Hormonal Status and Colorectal Function

Abdominal pain and diarrhea are significantly more common in the days just before and during menses than in between (Bernstein et al. 2014). Constipation is very common during pregnancy, mainly because of relaxation of smooth muscle cells. Anal resting and squeeze pressures are significantly lower in post- than in premenopausal women, but it is unknown whether this is due to lower levels of estrogen.

Thyrotoxicosis may cause diarrhea while both hypothyrosis and hypercalcemia in hyperparathyroidism may cause constipation. Rare syndromes with excessive production of hormones or neurotransmitters from neuroendocrine tumours may cause diarrhea. The least rare of these is diarrhea as part of the carcinoid syndrome caused by large amounts of serotonin. Histamine acts on intestinal H_2 receptors to stimulate water, electrolyte, and mucus secretion and to promote strong contractions called power propulsion.

9 Bile Acids and Colorectal Function

Bile acids are synthesized in the liver and flow into the duodenum. In the small intestine, bile acids promote lipid digestion. Thereafter, 95 % of bile acids are reabsorbed in the distal ileum. If reabsorption is compromised due to surgery, inflammation, etc. or saturated because of excessive synthesis, an increased amount will reach the colon where they cause diarrhea by stimulating electrolyte and water secretion as well as colonic propulsive contractions Wingate et al. (1973).

10 Medication and Colorectal Function

An impressive number of **drugs** affect gastrointestinal function causing constipation, diarrhea, discomfort, or pain. The most commonly used are opioids, peroral antidiabetics, diuretics, and antidepressants. A more detailed list is found in Tables 7.1 and 7.2.

Table 7.1 Medication associated with constipation

Analgesics	Bile acid sequestrants
Antacids (aluminum, calcium)	Calcium-channel blockers
Antiarrhythmics	Chemotherapy agents
Anticholinergic agents	Diuretics (potassium wasting)
Anticonvulsants	Iron supplements
Antihistamines	Nonsteroidal anti-inflammatory agents
Antihypertensives	Opioids
Anti-Parkinson Agents	Tricyclic antidepressants
Antipsychotics	5-HT_3 receptor antagonists
Antispasmodics	

Table 7.2 Medication associated with diarrhoea

Abuse of laxatives	Antihypertensives
Antacids (magnesium)	Chemotherapy agents
Antiarrhythmics	Cholesterol lowering agents
Antibiotics	NSAID
Antidiabetics	Proton pump inhibitors

11 Neurology

Normal nerve supply is crucial for colorectal and anal function. Neuronal control of colorectal function has four levels: (a) **the enteric nervous system** within the bowel wall, (b) the prevertebral sympathetic ganglia, (c) the autonomic nervous system within the brain stem and spinal cord, and (d) higher cortical centers.

Disorders of the enteric nervous system may be localized to a short segment as seen in most cases of Hirschsprung's disease or irradiation damage. More general damage to the ENS can be seen in diabetes, Chagas' disease, chronic idiopathic pseudo-obstruction, and as paraneoplastic phenomena.

Damage to the **reflex center** in the sacral spinal cord or to the cauda equina is usually traumatic, iatrogenic (after neurosurgery), or congenital as in spina bifida. An interrupted reflex arch from the sacral spinal cord to the left colon, the rectum, and the anal canal results in a hypotone, flaccid left colon and rectum (Krogh et al. 2002). This is followed by reduced emptying of the rectosigmoid at defecation and severely prolonged transit through the left colon and rectum causing fecal impaction (Krogh et al. 2003).

Lesions above the reflex center in the sacral spinal cord are commonly seen after traumatic injury or spinal surgery. The result is usually increased tone and contractility of the left colon and rectum and prolonged transit throughout the colorectum.

Cerebral lesions, whether congenital, vascular, or traumatic, are associated with constipation. The exact mechanism is not fully explored but may be a combination of immobility, lack of supraspinal reflex control, and deficits.

Lesions of the central nervous system or the peripheral nerves to the anorectum may cause fecal incontinence due to a combination of reduced anorectal sensibility and lack of voluntary control of the external anal sphincter. Hence, most patients with spinal cord lesions and spina bifida as well as many with multiple sclerosis and stroke suffer from severe constipation and fecal incontinence.

Parkinson's disease in not a disease isolated to the substantia nigra. There is also loss of dopamine in the enteric nervous system and autonomic neuropathy. The result is constipation the severity of which is closely related to the severity of the classical neurological symptoms. Dystonia of striated muscle cells is a common feature of Parkinson's disease. Dystonia of the external anal sphincter is the cause of difficult rectal evacuation commonly seen in patients with Parkinson's disease.

12 Diabetes

Diabetes has become one of the most common diseases of the developed world. Diabetes is closely associated with diarrahea, constipation, and fecal incontinence. The pathophysiology of bowel symptoms in diabetes is extremely complex. There is autonomic neuropathy but also signs of degeneration of the enteric nervous system including the pacemaker cells of Cajal (Forster et al. 2005). Today, it is assumed that bowel dysfunction in diabetes is a combination of autonomous neuropathy, enteric neuropathy, dysfunctional cells of Cajal, reduced contractility of intestinal smooth muscle cells, and abnormally high levels of blood glucose.

13 Connective Tissue Disease

Some connective tissue diseases affect colorectal and anal sphincter function. The most well-known is **systemic scleroderma**. The disease may cause enteric neuropathy in early stages, and in more advanced cases there is myopathy of the colorectal wall and especially the internal anal sphincter muscle. Thus, approximately 30 % of patients have constipation, 38 % have diarrhea, and another 38 % suffer from fecal incontinence. Endoanal ultrasound may show fibrosis and atrophy of the internal anal sphincter, and the anal resting and squeeze pressures are lower than normal. The competence of the anal sphincter complex may be severely reduced in systemic scleroderma as illustrated by extremely high distensibility (Fig. 7.1).

14 Irradiation/Surgery

A detailed review of **postsurgical complications** and irradiation damage to the anorectum are beyond the scope of the present chapter. However, symptoms including urge, fractionated defecation, evacuation disorder, and fecal incontinence are common after rectal resection as described for the low anterior resection syndrome (LARS) (Emmertsen and Laurberg 2012). An enhanced postprandial response has been suggested a part of the pathophysiology behind LARS.

Radiation therapy against cancers within the pelvic cavity may cause acute and chronic damage to the rectum and the pelvic floor. Changes include fibrosis, nerve damage, and atrophy of the mucosa.

15 Inflammation

Colorectal function is affected by acute **inflammation** as seen in infectious diarrhea. Furthermore, chronic inflammatory bowel disease affects colorectal function. Thus, active ulcerative colitis increases distal colonic propulsive activity while causing stasis in the proximal colon. Inflammation of the rectal mucosa enhances the sensory response to distention causing hypersensitivity and reduced rectal compliance, thereby contributing to urgency and frequent defecations. Colonic motility has not been studied in Crohn's disease, but it is likely that the changes correspond to those

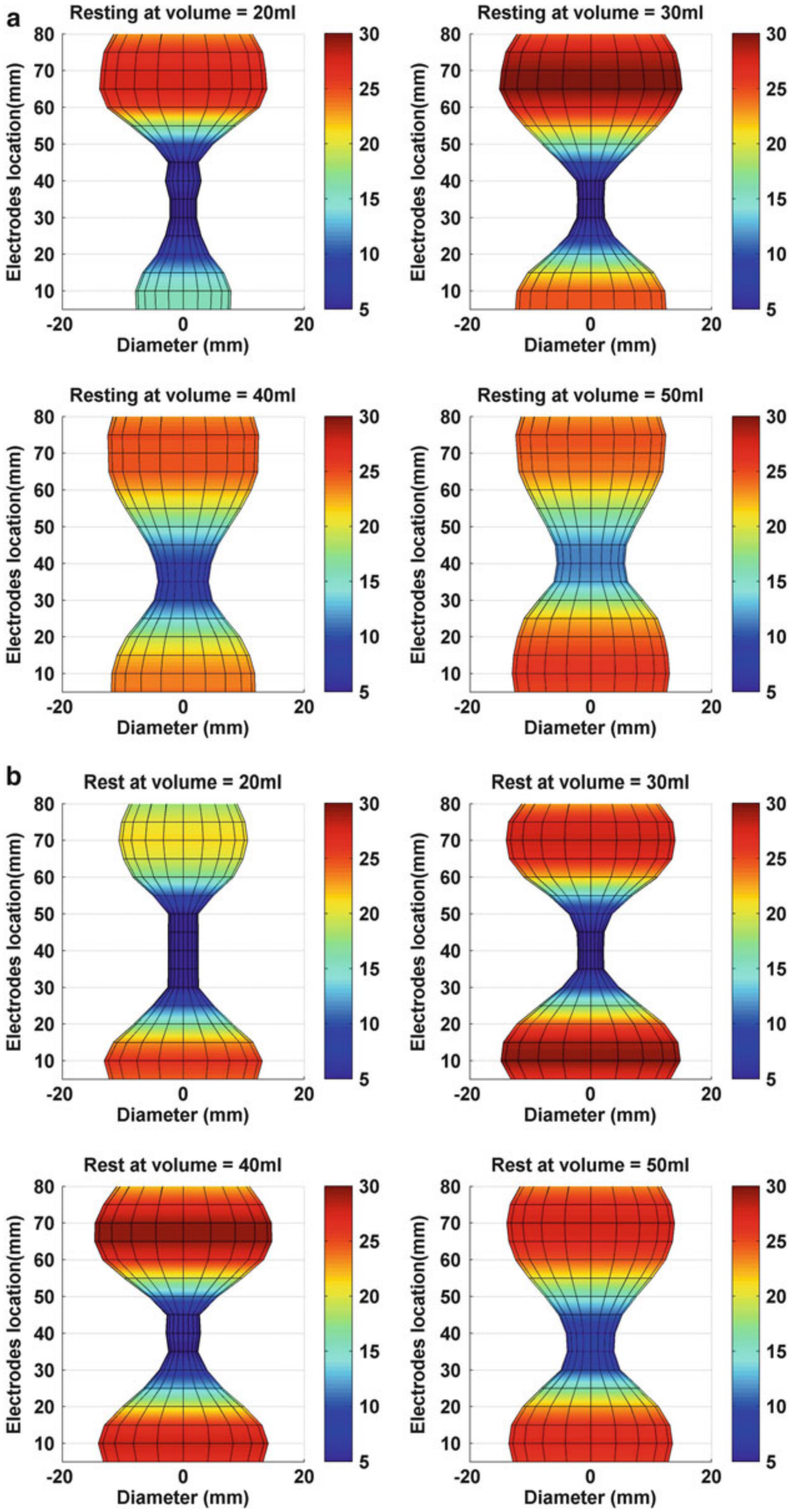

Fig. 7.1 Distensibility of the anal canal assessed with the Functional Lumen Imaging Probe. The anal canal of the patient with atrophy of the internal anal sphincther caused by scleroderma (**a**) reach much higher cross sectional areas during distension than seen in the healthy volunteer (**b**)

seen in ulcerative colitis. There is evidence that Crohn's disease causes damage to the enteric nerve cells not only at the site of inflammation but also in regions without other signs of inflammation.

16 Psychiatric Diseases

Several **psychiatric diseases** are associated with constipation or fecal incontinence. Especially common disorders associated with constipation are mental depression, eating disorders, and dementia. In children, most cases of fecal incontinence are classified as retentive fecal incontinence (formerly known as encopresis) where withholding defecation results in uncontrolled defecation.

17 Idiopathic Constipation and Fecal Incontinence

Even after thorough investigation, many cases of constipation or fecal incontinence remain idiopathic (primary or functional). These conditions are covered in other chapters of this book.

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Factors Affecting Defecation and Anal Continence

8

Simona Giuratrabocchetta and Ivana Giannini

Contents

Abstract

Pelvic floor anatomy and physiology are two of the most complex chapters of the study of the human body; each factor contributing to defecation and anal continence could be comparable to a tile of a puzzle in which the integrity and the perfect interaction of the pieces guarantees the final result. Pelvic floor muscles, anorectum, somatic, autonomic, and enteric innervation, in fact, act together in a "ranking order" essential for the correct function of the pelvic organs.

The muscular part of the pelvic floor is represented by the l*evator ani muscle,* and one of its components, the puborectalis muscle, contributes to the anorectal angle and, together with the internal and the external anal sphincter, is responsible for the anal resting tone. Lastly, a fine and perfect interaction among peripheral reflex (rectoanal inhibitory reflex), somatic, autonomic (sympathetic and parasympathetic), and enteric innervation, and some physical features (rectal compliance, stool consistency) are crucial for the modulation of sensitive and motor anorectal functions ensuring correct continence and defecation.

1 Introduction

Continence and defecation are two physiological functions strictly related to each other. Defecation has been described as the ability to properly

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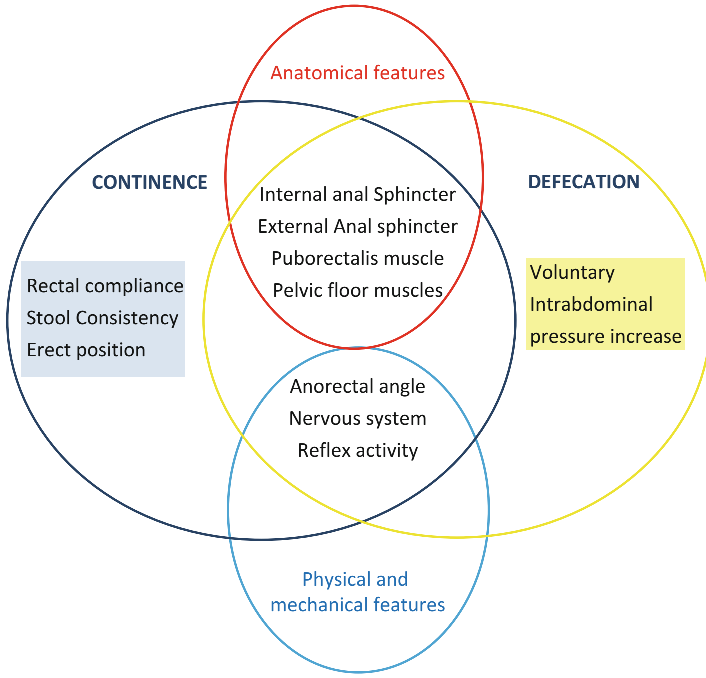

Fig. 8.1 Factors affecting continence and defecation

evacuate stool through the anus in the site and time desired and is controlled by anal continence, which is the voluntary ability to defer defecation to the best moment and site (Altomare et al. 2009).

Several factors are involved in providing anal continence and deferring defecation during different daily activities, including anatomical features, mechanical components, and neuromediated voluntary and involuntary control, each playing a different role both at rest and in squeezing condition (Fig. 8.1).

Pelvic floor is usually described as a complex multifactorial unit (Fritsch and Hötzinger 1995; Kapoor et al. 2008; Altomare et al. 2014); in case of posterior perineum function, this statement is even more sensitive (DeLancey 1999). A complex group of muscles, sensitive and motor nerves, organs, and reflexes are responsible for ensuring continence at rest and in urgency and defecation in pushing. Any deficit occurring in these structures leads to different types and severity of incontinence or obstructed defecation.

Even if the anatomy of the anorectal junction and pelvic floor is well known, the pathophysiology of the defecatory disorders has only recently been fully investigated and is not completely understood yet.

Over the last 50 years, many studies have been carried out to investigate anorectal physiology (Lestar et al. 1989), looking not only to its physical aspects (pressure, volume, and compliance) but also electromechanical (anorectal angle, pudendal nerve terminal latency, peripheral sensory and motor neuron activity) and imaging aspects (dynamic defecography, pelvic floor ultrasound, MRI), to help to understand the etiology of disorder and, as a consequence, the best way to treat patients with pelvic floor dysfunctions.

2 Pelvic Floor Muscles: Scaffold and Sphincters

The perineal floor is composed of several striated muscles, attached on the branches of the pubis, ilio, and ischiatic bones, taking part together in the pelvic scaffold, named *levator ani muscle*. It covers a wide attachment line, from the pubic bone, along the arcus tendineous, to the ischial spine, and it is divided into three components according to its bone insertions: iliococcygeus, ischiococcygeus, and the pubococcygeus. *Levator ani muscle* contributes to continence, as its resting tone makes the lumen of pelvic organs a virtual space. Some fibers of the pubococcygeus component contribute to form the puborectalis muscle that is another important actor in controlling both defecation and continence, as its sling-shaped configuration around anorectal junction is responsible of the amplitude changes of the anorectal angle (ARA).

Although the puborectalis muscle itself seems to play a sphincter-like role, there is a well-defined sphincter apparatus, composed of the internal anal sphincter (IAS) and the external anal sphincter (EAS). IAS is formed by smooth muscle cells out of voluntary control, appearing as a 2–3 mm thick circular muscle layer of the rectum, extended for 2 cm below the dentate line and separated from the EAS by an intersphincteric plane. Its continuous electrical activity, recorded as slow- and ultraslow-pressure waves with low frequency at rest, is responsible for about 70 % of anal resting tone maintenance (Wankling et al. 1968; Kerremans 1969; Lestar et al. 1989); it is also involved in the rectoanal inhibitory reflex (RAIR) evoked by rectal distension (see below). EAS is a striated-fiber voluntary-controlled muscle; it is the distal part of the puborectalis muscle and, according to Shafik, consists of three components arranged around the anal canal as three U-shaped loops (Shafik 1975). Its contribution to the resting anal tone is reported to be around 20 % (Lestar et al. 1989); nevertheless, its main role is the increase of anal tone during voluntary squeeze and during transient increase of intra-abdominal pressure (Shafik 1975; Shafik 1987). Both pelvic floor and sphincter muscle activity are responsible to act as "triggers" or as "targets" of anal pressure changes, at rest and in squeezing. Finally, the hemorrhoid cushions seem to be able to give a modest contribution (5 %) in the resting anal tone, thus playing a role in the maintenance of fine continence (Lestar et al. 1989).

3 Pelvic Floor Reflex Activity and Role of the Nervous System

All neural network controls for every voluntary and involuntary activities work according to a "ranking order" from a simple reflex arch to the higher cortical brain control for more complex activities. Anorectal functions themselves are under this neural "step-by-step" control too. Until about 50 years ago, little was known about the role of the neural pathways and sensitive receptors involved in the control of defecation and continence (Dickinson 1978). Nowadays, it is known that two intrinsic enteric plexus-like nervous systems (Auerbach and Meissner) are located in the wall of the whole gastrointestinal tract and are in contact with smooth muscle cells, modulating their activity by releasing various neurotransmitters; as a matter of fact, they work as a semiautonomous system. Sympathetic and parasympathetic are the extrinsic neural pathways that modulate anorectal functions (sensitive and motor) with opposed roles (parasympathetic acetylcholine stimulates smooth cell muscle activity, adrenergic substances inhibit their contraction). While the sensory pathway for rectal distension is the parasympathetic system to S2, S3, S4 via pelvic plexus, the anal canal sensation pathway follows the pudendal nerve to the same spinal roots (Baeten and Kuijpers 2007). Of great importance, moreover, are the sensory pathways regarding the awareness of rectal filling and the urge to defecate that reach higher CNS control sites (superior frontal gyrus and anterior cingulated gyrus) and evoke the voluntary contraction of puborectalis and EAS as motor response to defer defecation (Porter 1962).

The most important reflex involved in anal continence is the rectoanal inhibitory reflex (RAIR) defined as "the transient decrease in resting anal pressure by ≥25 % basal pressure in response to rapid inflation of a rectal balloon with subsequent return to baseline" (Lowry et al. 2001). Inflating a balloon in the rectum simulates the increasing rectal filling with feces that progressively come from proximal gastrointestinal tract following the coordinated mass movements (rectosigmoid motility).

RAIR is controlled by a neural intrinsic reflex localized in the gut wall, independent of spinal cord involvement (Frenckner 1975), as it has been proven by its absence in Hirschsprung's disease (Lund and Scholefield 1996) and by the pressure fall recorded even when autonomic nerve supply is blocked (Frenckner and Ihre 1976).

4 Physical Features: Anorectal Angle, Rectal Compliance, Stool Consistency

Not only anatomy and physiology have a relevant role in continence and defecation; there are also some physical principles of great importance applied to anorectal functions.

The rectal axis and the anal canal form the anorectal angle (ARA); at rest, it is usually considered to be around 90°–110° (Reiner et al. 2011); this amplitude increases with voluntary anal straining to defecate, helping the stool passage through the anus (Reiner et al. 2011), while it becomes more acute in squeezing, helping maintenance of continence. A recent study also reported that the angle size varies considerably in erectus (<80°) or in squatting position (Altomare et al. 2001): that means that the erect position helps the maintenance of continence, whereas the pelvic floor relaxation and the sitting position straighten the ARA, allowing defecation (Baeten and Kuijpers 2007).

Rectal compliance is the ability of the rectum to work as a reservoir, storing its content and increasing its volume, with minimal increase of pressure, adapting the tone of muscular wall but keeping the internal pressure low. This function is regulated by the baroreceptors sensitive to distension located in the rectal wall, linked to the sensory pathways along the sacral nerves. Several studies have reported that sensory threshold volumes may be influenced by structural and/or rectal biomechanical properties. Patients with intractable constipation, for example, have persistent dilatation of the rectum, so that greater volumes will be required to distend and thus stimulate the rectum (higher compliance). Furthermore, increased rectal compliance results in a failure of generation of forces in the rectal wall during distension that would normally stimulate rectal mechanoreceptors (Scott et al. 2011). This property of storage and adjustment to volume increase is one of the biomechanisms involved to ensure continence.

The consistency and volume of stools are another factor affecting continence. Unlike the rectal wall which is sensitive only to distension, the anal canal is rich in mechano-, proprio-, thermo-, and pain receptors, able to develop a fine discrimination of the stool characteristics, so that each subject always is aware about the stool consistency. Any deficit in these sensitive receptors could lead to incontinence.

With regard to stool consistency, hard and scarce stools can obviously be controlled more easily than watery stools, but they are more difficult to expel; on the contrary, liquid stools or flatus are more difficult to contain and even without organic deficit could lead to minor incontinence (Baeten and Kuijpers 2007).

5 Continence and Defecation: Two Sides of the Same Coin

The interplay of all the aforementioned factors is essential to ensure continence and allow defecation. Most authors agree that the most important element is the puborectalis sling activity, so that the major forms of incontinence come from its damage (Shafik 1987). On the other hand, other authors attribute the most relevant role in preventing incontinence to the overall squeeze action resulting from the internal and external anal sphincter tonic activity; even the closed ARA (*flap valve* theory) in the erect position has been mentioned to be involved in ensuring continence (Altomare et al. 2001).

At rest, the pressure into the rectum is lower than in the anal canal, but the increasing mass in rectum, coming from the proximal gastrointestinal tract, makes that pressure higher, so that the rectal receptors, sensitive to distension, activate the RAIR and the relaxation of IAS. Then, the increased intra-adominal pressure for the Valsalva maneuver and the rectosigmoid peristalsis moves down the stools to the upper part of anal canal, where the highly sensitive receptors discriminate stool consistency, leading to the urgency to defecate; awareness and the reflected contraction of EAS prevents the loss of feces ("*sampling reflex*"). The involuntary RAIR modulated by the IAS relaxation works with the voluntary contraction of EAS and puborectalis muscle, until the rectum with its compliance adapts passively to a new volume and the degree of intrarectal pressure is reduced (Dickinson 1978; Shafik 1987). The stools are then stored back to the high rectum, the urge to defecate disappears, and the anal sphincters restore their basal tone. The puborectalis muscle contraction itself makes the ARA more acute than resting, contributing to maintenance of continence.

When time and place are appropriate, the sitting position and the voluntary increasing of the intra-adominal pressure lead to a straightening of ARA and a voluntary relaxation of puborectalis muscle and EAS, with the final passage of stool through the anus. At the end of defecation, a transient EAS contraction (the "*closing reflex*") closes the anal canal, recovering the basal intraluminal pressures and anal tone (Wexner et al. 2005).

In conclusion, several factors affect the mechanisms of continence and defecation, not only neuroanatomical but also even biomechanical and physical. Several anatomical and physiological studies investigated these issues, reporting different theories about continence and defecation in humans. Despite the wide acceptance of the basic physiology of those, the pathophysiology of some anorectal disorders is still unclear. Knowledge of the different factors involved in the control of continence and defecation mechanisms could help surgeons to adopt the most rational approach to treat their alteration.

6 Cross-References

► Physiology of the Rectum and Anus

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Anal Surgery: Organ and Function Preservation

9

Ann Lowry

Contents

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Abstract

Anorectal disorders such as hemorrhoids, anal fissure, and anal fistulas are common in Western countries. Medical management may resolve symptoms in patients with hemorrhoids and anal fissures, but some patients will require surgery. Intervention is necessary most of the time in patients with anal fistulas. Mortality and significant morbidity are rare after such surgery, but functional disorders particularly incontinence may occur in a significant percentage of patients. Lateral internal sphincterotomy, the current standard procedure for anal fissures, carries a risk of minor incontinence of 15–45 %. Incontinence after hemorrhoidectomy is less common and may be minimized by careful surgical technique. The highest risk of incontinence occurs with fistula surgery. For those patients, preoperative risk factors, clinical symptoms, and the anatomy of the fistula should be carefully assessed. Treatment plans must balance the predicted success against the risk of incontinence.

Anorectal disorders are quite common in Western countries, and while many patients respond to medical management, some will require surgery. Typically, the surgery is relatively minor and most patients recover uneventfully. However, functional disturbances may occur postoperatively. Incontinence is the most frequently reported functional disturbance, but constipation related to anal stenosis may also occur. Depending upon the

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condition and type of surgery, the incidence of postoperative incontinence varies from 0 % to 64 % (Ommer et al. 2008). The most common anorectal conditions are anal fissure, hemorrhoids, and fistula in ano; this chapter will focus on functional changes after surgery for each of these conditions and options for prevention.

1 Anal Fissure

Anal fissures are longitudinal tears in the lining of the anal canal distal to the dentate line. Trauma and hypertonia of the internal sphincter are thought to be part of the etiology, but the exact etiology is unknown (Shawki and Costedio 2013).

Initial treatment of an anal fissure includes bowel management programs and efforts to reduce anal spasm including warm tub baths, topical analgesic ointments, and compounded ointments of either nitrates or calcium channel blockers. Injection of botulinum toxin into the internal sphincter may also be used to reduce the hypertonia of the internal sphincter.

1.1 Lateral Internal Sphincterotomy

Surgery is indicated for patients with persistent, symptomatic fissures who have not responded to medical measures. Lateral internal sphincterotomy (LIS) is generally considered the front-line surgical therapy. The procedure traditionally involves dividing the internal sphincter in either the right or left lateral portion of the anal canal to the level of the dentate line. A recent meta-analysis of studies of LIS for anal fissures with a minimum of 2-year follow-up included 22 studies and 4512 patients (Garg et al. 2013).The success rate ranged from 68 % to 100 %; recurrence was noted in 0–30 % of patients. As many as 47.6 % of patients experienced some change in their continence postoperatively. Incontinence of flatus ranged from 0 % to 36 %, soiling occurred in 0–43 %, and incontinence of liquid stool ranged in 0–21 % and of solid stool 0–4.4 %. One study separated mildly from severely incontinent patients based upon their incontinence scores; 7.5 % of all patients and 67.9 % of incontinent patients experienced mild incontinence, while 3.6 % of all and 32.1 % of incontinent patients had severe symptoms (Kement et al. 2011).

There is evidence that these numbers underestimate the reality. One study compared the results of a chart review to a postal survey of patients postoperatively (Casillas et al. 2005). Chart review revealed incontinence of flatus in 4.4 % and of stool in 2.8 %, while the postal survey of the same patients showed rates of 28.7 % and 31.5 %, respectively. In addition, studies with longer follow-up reveal higher rates (Walker et al. 1985; Nyam and Pemberton 1999; Casillas et al. 2005; Rotholtz et al. 2005; Brown et al. 2007; Hancke et al. 2010; Table 9.1).

One group investigated patients with delayed onset of incontinence after LIS (Levin et al. 2011). The mean interval between surgery and the onset of symptoms was 10 ± 1.2 years. When compared to patients with delayed onset of incontinence after obstetrical trauma, the postoperative patients developed symptoms earlier (mean of 10 years versus mean of 25 years later), but their symptoms were milder.

Table 9.1 Incontinence after lateral internal sphincterotomy with long-term follow-up

Study	Follow-up (mos.)	Response rate (%)	Perfect continence preop (%)	Perfect continence postop (%)
Walker 1985	51	30	NA	85
Nyam 1999	72	83.4	100	89
Casillas 2005	51	62	100	62
Rotholtz 2005	67	n/a	100	89.7
Brown 2007	79	58.5	79	33
Hancke 2010	79	69.4	100	52.4

A number of studies sought to identify risk factors for incontinence after LIS. Age (Khubchandani and Reed 1989; Arroyo et al. 2005), female gender (Hasse et al. 2004), history of vaginal delivery (Sultan et al. 1994; Casillas et al. 2005; Elsebae 2007; Kement et al. 2011), and combination with another anorectal procedure (Kement et al. 2011) were found to increase the risk. Other studies found a correlation of incontinence with preexisting external sphincter defects related to obstetrical injuries (García-Granero et al. 1998; Tjandra et al. 2001). Patients with anterior fissures developed incontinence after partial lateral internal sphincterotomy at a much higher rate than ones with posterior fissures (39 % versus 6 %, $p < 0.003$) (Gandomkar et al. 2015). The length of the sphincterotomy but not resting pressures or width of the defect in the internal sphincter correlated with incontinence (Garcia-Aguilar et al. 1998).

Although the cure and patient satisfaction rates are high, the frequency of incontinence after LIS is concerning. Efforts at prevention include aggressive medical management to avoid surgery, tailoring of the sphincterotomy, and alternative procedures.

1.2 Alternatives: Medical Management

If medical management is successful, then surgery may be avoided completely. Initial therapy includes management of stool consistency and warm baths. Several studies report similar results of approximately 44 % healing rates with 18–27 % recurrence rates in those patients (Shub et al. 1978; Hananel and Gordon 1997). Medical regimens now include the addition of sphincter relaxants including nitrate formulations, calcium channel blockers, and botulinum toxin injections after studies demonstrated decreased anal resting pressures with those treatments (Loder et al. 1994). Reported healing rates with topical nitrates range from 18 % to 85 % (Shawki and Costedio 2013); there is a notable incidence of adverse effects particularly headache (20–90 %), and recurrence rates may be as high as 30 %. A recent systemic review of randomized trials comparing topical glyceryl trinitrate to topical diltiazem (Sajid et al. 2013) demonstrated that the treatments were equally effective, but there were fewer adverse effects with diltiazem.

Injection of botulinum toxin into the anal sphincter muscle also relaxes the anal sphincter and improves circulation (Brisinda et al. 2009; Jost and Schimrigk 1994). The sphincter location and dosages of the injections vary among reported studies. Healing rates range from 41 % to 74 % (Giral et al. 2004; Arroyo et al. 2005; Menteş et al. 2006) with recurrence ranging from 0 % to 53 %. A recent meta-analysis summarized the results of studies comparing botulinum toxin to LIS (Chen et al. 2014). Combination of botulinum toxin injection and topical diltiazem was compared to LIS in a randomized trial (Gandomkar et al. 2015). In patients with a fissure of less than 12-month duration, the healing rate was 100 % in both groups, but in patients with fissures of a longer duration, the healing rate after LIS was significantly higher (86 % vs. 23 %). After LIS, 16 % of patients reported incontinence, while 4 % did after the combination treatment.

A Cochrane review of nonsurgical therapy for anal fissure in 2012 included 5031 patients (Nelson et al. 2012). The review concluded that nitrates resulted in marginally but significantly better healing rates than placebo but the recurrence was common. Botox and calcium channel blockers had similar healing rates but lower incidence of adverse effects. Two studies reviewed the results of a treatment algorithm in which patients progressed from topical ointment to botulinum toxin injections to surgery (Sinha and Kaiser 2012; Farouk 2014). The majority of patients responded to conservative therapy with 1–26 % requiring LIS.

Anal dilatation, fissurectomy, and advancement flaps are among the surgical options to LIS. In addition, partial, tailored, or limited internal sphincterotomy has been suggested to reduce the risk of incontinence.

1.3 Alternatives: Tailored Internal Sphincterotomy

Two methods are described. One involves dividing the internal sphincter only to the apex of the fissure; the other calibrates division of the muscle to the point of release of the anal spasm. In all three randomized trials comparing traditional sphincterotomy to the dentate line and the procedure limited to the apex of the fissure, healing rates were better in the traditional arm (Menteş et al. 2005; Elsebae 2007; Ho and Ho 2005). However, two studies reported higher rates of incontinence in that arm (Menteş et al. 2005; Elsebae 2007). Results of calibrated sphincterotomy are reported in three series; healing rates were comparable to the traditional procedure, but incontinence rates were lower (Cho 2005; Rosa et al. 2005; Menteş et al. 2008). A group of investigators attempted to determine how much of the internal sphincter could be divided without causing incontinence in women (Murad-Regadas et al. 2013). They found the likelihood of maintaining perfect continence after sphincterotomy was significantly greater when 25 % or less of the internal sphincter was divided.

1.4 Alternatives: Anal Dilatation

Anal dilatation, typically manually controlled stretching of the anal sphincter, is performed under anesthesia in the operating room. Published healing rates vary from 40 % to 70 % with a wide range of recurrence (2–55 %) (Nielsen et al. 1993; Farouk et al. 1998; Konsten and Baeten 2000). Most troubling is the up to 40 % incidence of fecal soiling and 16 % fecal incontinence. Anal ultrasound revealed sphincter defects in 50 % of patients after anal dilatation (Nielsen et al. 1993). Because of the reported rates of soiling and/or incontinence, the procedure is not recommended.

Several modifications of anal dilatation are reported. Controlled anal dilatation with anal dilators or a pneumatic balloon dilator has been compared to LIS (Renzi et al. 2008). The procedures resulted in equivalent reduction of mean resting pressures and healing rates. Although early incontinence was reported in both groups, at 24 months, none of the pneumatic balloon dilatation patients were incontinent compared to 16 % of the lateral internal sphincterotomy patients ($p < 0.0001$). In a study comparing anal dilators to lateral internal sphincterotomy, there was no difference in healing rates; no incontinence was reported in either group (Yucel et al. 2009).

1.5 Alternatives: Fissurectomy

Fissurectomy involves resection of the fissure; the rationale is that replacing inflamed tissue with a clean wound will improve healing. A study of 43 patients with a minimum of 5-year follow-up after fissurectomy revealed that all fissures healed initially, but 11.6 % developed a recurrence (Schornagel et al. 2011). The only patient who developed new incontinence postoperatively was the one who had had a prior LIS. Mousavi and colleagues randomized 62 patients with fissures to traditional LIS or fissurectomy (Mousavi et al. 2009). Incontinence of flatus was noted in two patients after fissurectomy; no incontinence was noted after LIS. Healing rates were similar. A retrospective study comparing patients undergoing LIS to fissurectomy found similar rates of healing and incontinence, but the fissurectomy patients were statistically more likely to require additional medical and surgical therapy (Yurko et al. 2014).

1.6 Alternatives: Anoplasty

Anoplasty is performed with the goal of covering the fissure bed with new well-vascularized tissue; dermal flaps are reported most commonly. A randomized trial comparing anal flaps to LIS in 40 patients found similar healing rates (85 % in the flap group and 100 % in the sphincterotomy group) with no incontinence reported in either group (Leong and Seow-Choen 1995). A retrospective study compared 30 patients undergoing LIS to 30 patients undergoing dermal flap closure (Hancke et al. 2010). The groups were comparable and denied preoperative incontinence. In the patients available for a minimum of 72-month follow-up, 71 % of LIS patients and 29 % of the flap patients reported persistent minor anorectal symptoms. No patient reported incontinence of formed stool, but 47 % of the LIS and 5.8 % of

Table 9.2 Results of randomized trial of LIS with and without anoplasty and anoplasty alone (Magdy et al. 2012)

Procedure	Healing rate (1 year) (%)	Recurrence rate (%)	Incontinence (%)
LIS (50)	84	4	14
V-Y flap (50)	48	22	0
Tailored LIS+ V-Y flap (50)	94	2	2

the flap group reported either incontinence to flatus or soiling. One hundred fifty patients with chronic anal fissures were randomized to conventional lateral internal sphincterotomy, V-Y advancement flap, or combined tailored sphincterotomy and V-Y advancement flap (Magdy et al. 2012) (Table 9.2). The use of flaps for recurrent fissures after sphincterotomy or in patients with weak sphincters is reported with good success (100 %) (Nyam et al. 1995).

1.7 Summary

Medical management may help avoid surgery in the majority of patients. The healing rates of botulinum toxin injection are inferior to the surgery, but there is little risk of incontinence. The standard for surgical care is currently LIS, but there is a small but significant risk of incontinence particularly in long-term follow-up. That risk must be balanced against healing rates of any alternative procedure. Tailored sphincterotomy, tailored sphincterotomy with anoplasty, and balloon dilatation show promising results. In patients with fissures without hypertonia or with preexisting incontinence, it would be especially appropriate to consider a flap procedure prior to any division of muscle.

2 Hemorrhoids

Hemorrhoidal symptoms are common, but the incidence is difficult to determine as many people do not seek medical care for these symptoms. Since the denominator is unknown, the percentage of patients who require surgery cannot be reliably calculated. Medical management typically includes improving stool consistency, warm baths, oral medications, and topical treatments. Patients with hemorrhoid symptoms most frequently also complain of constipation although the symptoms may occur in patients with diarrhea. High-fiber diets and fiber supplements are beneficial. A systemic review of seven trials involving 400 patients comparing increased fiber intake to a control group showed benefit in reduction of bleeding and other symptoms (Alonso-Coello et al. 2006). There is little objective data to support the benefit of warm baths, but easing of symptoms is often reported by patients (Tejirian and Abbas 2005). One randomized trial comparing cold to hot baths in patients with anal pain has been reported since that review. There was no statistically significant difference between the two groups although only the patients with warm baths experienced significant improvement in their pain between days 1 and 7 (Maestre et al. 2010). A Cochrane review of phlebotonics, a heterogeneous class of drugs consisting of plant extracts (i.e., flavonoids) and synthetic compounds (i.e., calcium dobesilate), revealed potential benefit for patients with hemorrhoid symptoms including bleeding (Perera et al. 2012). Topical treatments often include creams, ointments, and suppositories. Case series showing benefit have been published, but no recent randomized comparative trials were found (Altomare and Giannini 2013).

Patients with symptoms related only to internal hemorrhoids may improve with office-based treatment. However, patients with symptomatic external hemorrhoids, significant prolapse, or mixed hemorrhoids as well as those who have failed office treatment will require surgery.

2.1 Excisional Hemorrhoidectomy

Excisional hemorrhoidectomy using an open or closed technique is the traditional approach. Both fecal incontinence and constipation secondary to anal stenosis are reported complications. The

Table 9.3 Incontinence after conventional hemorrhoidectomy

Author	N	Follow-up	Incontinence (%)
McConnell 1983	441	1–7 years	0.5
Konsten 2000	35	17 years	20
Johannsson 2002	418	2–11 years	9.6
Smyth et al. 2003	16	3 years	12
Racalbuto 2004	50	4 years	6
Peters et al. 2005	14	3 years	0
Bouchard et al. 2013	488	1 year	8.5[a]

[a]De novo incontinence; 16 % of patients with preoperative incontinence improved

procedure is also reported to impact sexual function in women.

Incontinence may occur in the immediate postoperative period and then resolve (McConnell and Khubchandani 1983). Rates of persistent incontinence range from 0 % to 20 % (Table 9.3). Most commonly, patients complain of incontinence of flatus or fecal soiling, but frank incontinence of stool is also reported. Proposed etiologies of postoperative incontinence include the use of anal retractors (van Tets et al. 1997), inadvertent sphincter injury, and absence of the anal cushions.

One study compared ten patients with anal incontinence after surgery to a matched group (Abbasakoor et al. 1998). Eight of the ten patients had anal sphincter defects on ultrasound; five had internal sphincter defects, two had both internal and external sphincter defects, and one had an external sphincter defect only. No abnormalities were seen on ultrasound in the control group. Within a cohort of 418 patients following Milligan-Morgan hemorrhoidectomy, 40 patients reported fecal incontinence they attributed to the surgery (Johannsson et al. 2013). Nineteen of those patients underwent evaluation and were compared to 15 asymptomatic hemorrhoidectomy patients and 19 people from a matched population-based control group. The study group had higher incontinence scores, more reports of incomplete evacuation, lower resting pressures, and lower threshold volumes during saline infusion test. Four of the nine patients had sphincter defects on anal ultrasound; all defects were in the external sphincter. Both of these studies suggest that anal sphincter injury explains some but not all of the postoperative incontinence. They do make clear the importance of meticulous surgical technique to avoid injury to both the internal and external sphincter muscles.

Thomson postulated that the anal cushions contributed to continence (Thomson 1979); based upon that theory, excision of the hemorrhoids themselves may cause incontinence. Li and colleagues investigated the question of whether removal of the anal cushions alone led to incontinence (Li et al. 2012). Seventy-six patients underwent saline infusion testing before a Milligan-Morgan hemorrhoidectomy. They were sorted into three groups according to their preoperative saline threshold volume. Only the group with the lowest threshold volume experienced a significant difference in pre- and postoperative threshold volumes and Cleveland Clinic incontinence scores; none of the patients reported incontinence. The authors contend that since surgery removed the anal cushions in all of the patients, some other mechanism is necessary to explain the changes.

Constipation related to anal stenosis is a reported complication after hemorrhoid surgery; hemorrhoidectomy accounts for approximately 90 % of cases of anal stenosis (Milsom and Mazier 1986; Brisinda et al. 2009). The incidence after hemorrhoid surgery ranges from 1.5 % to 3.8 % (Eu et al. 1995). Although hard to prove, it is accepted that overzealous excision of the anoderm leads to scarring and ultimately stricture formation. Recent trials report rates of anal stenosis persisting more than 1 year from 0 % to 7.5 % (Boccasanta et al. 2001; Hetzer et al. 2002; Shalaby and Desoky 2001; Palimento et al. 2003; Racalbuto et al. 2004). Some attribute the good results to careful surgical technique to limit the amount of anoderm removed; others credit close postoperative follow-up with rectal examinations and anal dilators if needed (Racalbuto et al. 2004).

One published report suggests the sexual function in women may be affected by hemorrhoidectomy. In Taiwan, Lin and associates surveyed women after hemorrhoidectomy using the Chinese version of the Female Sexual Function Index and compared their results to a control group (Lin et al. 2009). The surgical group had lower average scores (46.38 vs. 65.69, $p < 0.001$) and lower scores in all domains except desire. These findings would need to be replicated to better understand the impact of hemorrhoidectomy on sexual function.

Motivated by the goal of easing the recovery, new methods of surgery including LigaSure hemorrhoidectomy, stapled hemorrhoidectomy, and Doppler-guided arterial ligation have been developed. As opposed to anal fissure surgery, the primary impetus has not been the avoidance of the postoperative functional issues.

2.2 LigaSure Hemorrhoidectomy

The basic principle of LigaSure or other thermal device hemorrhoidectomies is the same as the traditional hemorrhoidectomy, but a thermal device rather than scalpel or scissors is used. Twelve studies with 1142 patients were included in a Cochrane review in 2009 (Nienhuijs and de Hingh 2009). Immediate postoperative pain was less after LigaSure hemorrhoidectomy, but there was no statistical difference in anal stenosis (0–2.3 %) or incontinence (none reported) although follow-up was limited in most studies. A 2-year minimum follow-up study of 666 consecutive patients after LigaSure hemorrhoidectomy revealed that one patient was treated for an anal stricture (0.1 %) and 11 (1.7 %) patients reported incontinence to flatus (Chen et al. 2013). Others have reported a higher incidence of anal stenosis up to 2.5 % (Gravante and Venditti 2007; Wang et al. 2006); caution is warranted to avoid thermal burn to the anoderm left in place.

2.3 Stapled Hemorrhoidectomy

During a stapled hemorrhoidectomy, a cylinder of the rectal mucosa above the internal hemorrhoids is excised with the goal of devascularization and reduction of the anal cushions. The external hemorrhoids are not excised but may be reduced by this technique. A Cochrane review comparing stapled versus conventional surgery for hemorrhoids was updated in 2010 to include 22 studies (Lumb et al. 2010). These studies show that there is reduced immediate postoperative pain with a stapled procedure but a higher rate of recurrent symptoms and need for additional procedures. An additional consideration about the stapled procedure is the rare but severe complications reported including pelvic sepsis, rectovaginal fistula, rectal perforation, and Fournier's gangrene (Molloy and Kingsmore 2000; Ripetti et al. 2002; Wong et al. 2003; Pessaux et al. 2004; Cirocco 2008). Recurrent hemorrhoidal symptoms may be more frequent after stapled hemorrhoidectomy compared to excisional procedures (van de Stadt et al. 2005).

In terms of functional outcomes, there is a nonsignificant trend toward less anal stenosis after a stapled procedure but a nonsignificant trend toward a higher rate of soiling, incontinence, and fecal urgency. Fecal incontinence or soiling after 1 year ranged from 0 % to 10 % (Mehigan et al. 2000; Boccasanta et al. 2001; Correa-Rovelo et al. 2002; Kairaluoma et al. 2003; Racalbuto et al. 2004; de Nardi et al. 2008). Higher rates of urgency and pain were reported (30 %) early after introduction of the technique (Cheetham et al. 2000). Placement of the purse-string suture too close to the dentate line with resulting inclusion of the anoderm and even the anal sphincter muscle in the stapler may be the cause. Anal stenosis occurred in 0–2 % of patients (Boccasanta et al. 2001; Hetzer et al. 2002; Shalaby and Desoky 2001; Palimento et al. 2003; Racalbuto et al. 2004).

2.4 Doppler-Guided Hemorrhoid Artery Ligation

The concept behind this technique is that suture ligation of the terminal branches of the superior rectal arteries will reduce hemorrhoid symptoms. In this technique, the terminal branches are identified about 3 cm above the dentate line using a

Table 9.4 Functional results by type of hemorrhoid surgery

Type of surgery	Incontinence (%)	Anal stenosis (%)	Comments
Conventional	0–20	0–7.5	Postop pain
LigaSure	0	0–2.5	Limited follow-up data
Stapled	0–10	0–2	Higher recurrence rates, rare severe complication Leaves external hemorrhoids

proctoscope equipped with a Doppler probe and a light source (Ratto 2014). Those branches are then ligated. In patients with bleeding but no significant prolapse, ligation is performed alone. In patients with mucosal prolapse, a mucopexy using a continuous suture is added. Most series do not report data on incontinence or anal stenosis. A systemic review of transanal hemorrhoidal dearterialization included 1996 patients; incontinence data reported about only 693 (Giordano et al. 2009a). When incontinence data is reported, the incidence is low (0–0.4 %) (De Nardi et al. 2014; Denoya et al. 2013; Giordano et al. 2009b; Ratto et al. 2010). Anal stenosis is not reported. While postoperative pain and incontinence are less than in conventional hemorrhoidectomy, the rate of recurrent prolapse symptoms ranges from 11 % to 59 % (Giordano et al. 2009a).

2.5 Summary

Treatment of hemorrhoids depends upon the patient's symptoms, presence of symptomatic external hemorrhoids, and the grade of internal hemorrhoids. Medical and office-based treatments should not impair bowel function. The functional impact for each of the surgical options is summarized in Table 9.4. For patients with bleeding and low-grade prolapse, either stapled hemorrhoidectomy or transanal hemorrhoid dearterialization with or without mucopexy poses minimal to no risk of incontinence or anal stenosis. However, external hemorrhoids or tags are not treated, and the rate of recurrent symptoms is higher than after conventional hemorrhoidectomy. For patients with significant external hemorrhoids or failures of other treatments and patients wishing to avoid a higher recurrence rate, then excisional hemorrhoidectomy is the appropriate choice. For those patients, it is important to minimize stretching of the anal sphincter muscles during surgery, use meticulous technique to avoid injury to the sphincter muscles, and limit the amount of anoderm excised. These cautions are pertinent regardless of the technique utilized.

3 Anal Fistulas

The true prevalence of anal fistulas is unknown. One study of a defined population estimated that fistulas occur in 12.3 per 100,000 men and 8.6 per 100,000 women (Sainio 1984). Surgery is frequently necessary to close the fistulas. However, because of the involvement of the sphincter muscle in most anal fistulas, treatment recommendations need to balance the success rate against the risk of postoperative incontinence.

The patient experience information emphasizes the importance of these considerations, and the outcome literature demonstrates that postoperative incontinence occurs frequently enough to influence treatment decisions. When patients and surgeons were asked to list their top five objectives of fistula surgery, both groups included continence in their top three (Wong et al. 2008). Ellis provided 74 patients with 10 scenarios of options for fistula surgery; the scenarios included published success and postoperative incontinence rates (Ellis 2010). The sphincter sparing options were ranked much higher than fistulotomy. The majority (74 %) chose a sphincter sparing procedure as their first choice. Studies vary about whether fistula recurrence or incontinence has more impact on quality of life or patient satisfaction (Sailer et al. 1998; Lunniss et al. 1994; García-Aguilar et al. 2000). One study found that postoperative soiling did not impact quality of life as measured by the Gastrointestinal Quality of Life Index (GIQLI) (Pescatori et al. 2004). Another study with a high rate of incontinence after fistulotomy (50 %) found that 87 % of patients were satisfied with the outcome (Westerterp et al. 2003). However, other investigators found a correlation between postoperative incontinence and quality of life scores (Jarrar and Church 2011). The reasons behind these differences are unclear. Finally another study using GIQLI found significant improvement in scores after successful surgery for anal fistulas (Seneviratne et al. 2009).

Most publications do not include the use of standardized instruments to survey for incontinence or anorectal physiology testing. The results are illuminating when such studies are performed. Roig and colleagues prospectively analyzed 143 patients with pre- and postoperative surveys, manometry, pudendal nerve testing, and anal ultrasounds after fistula surgery (Roig et al. 2009) Preoperative incontinence was noted in 14.2 % of patients and in 49.2 % of patients postoperatively. Incontinence scores worsened in patients after fistulotomy and endorectal advancement flap but improved slightly in patients after fistulotomy and immediate sphincter reconstruction. All surgical approaches produced increased defects in the internal sphincter on ultrasound; increased defects in external sphincter developed only after fistulotomy. The presence of defects correlated significantly with postoperative incontinence and lower manometry results. These findings demonstrate the importance of sphincter sparing surgery to avoidance of new incontinence as well as the frequency with which sphincter injury occurs.

3.1 Fistulotomy

Fistulotomy, division of the tissue overlying the fistula between the internal and external opening including the sphincter muscle, has a reported success rate of 80–100 % (van Tets and Kuijpers 1994; Sangwan et al. 1994; Garcia-Aguilar et al. 1996; Westerterp et al. 2003; van Koperen et al. 2008; Hyman et al. 2009; Roig et al. 2009; Jordán et al. 2010). Recurrence rates range from 0 % to 10 % (van Tets and Kuijpers 1994; Westerterp et al. 2003; van Koperen et al. 2008; Jordán et al. 2010).

Minor postoperative incontinence ranges from 18 % to 52 %, while soiling has been reported in up to 45 % of patients (van Tets and Kuijpers 1994; Sangwan et al. 1994; Garcia-Aguilar et al. 1996; Westerterp et al. 2003; van Koperen et al. 2008; Hyman et al. 2009; Roig et al. 2009; Jordán et al. 2010). Incontinence of solid stool is much less frequently reported. Perfect continence is rarely documented, but in one study only 29 % of patients had perfect continence after fistulotomy (van Koperen et al. 2008). Several studies report that the incontinence rate varies with the height of the internal opening (van Tets and Kuijpers 1994; Cavanaugh et al. 2002; Westerterp et al. 2003; Jordán et al. 2010). Fistulotomy results in impaired incontinence in 8–44 % of low fistulas and 34–82 % of high fistulas (van Tets and Kuijpers 1994; Westerterp et al. 2003; Jordán et al. 2010). Preoperative incontinence predicts postoperative incontinence (Jordán et al. 2010).

Fistulotomy for an intersphincteric fistula is frequently considered to be a low-risk procedure, but the LIS data suggests that incontinence may occur in a significant proportion of patients. In a study of 148 patients undergoing fistulotomy for intersphincteric fistulas, postoperative incontinence was noted in 20.3 % of patients (Toyonaga et al. 2007). Six patients reported soiling, 27 patients incontinence of flatus, and four patients incontinence of liquid stool. There was a significant decrease in resting pressures and anal canal length but no effect on squeeze pressures.

4 Alternatives

So while the recurrence rate after fistulotomy is low, the price may be high in terms of impaired continence. Because of that, alternative procedures with the goal of maximizing success and minimizing postoperative incontinence are of interest. Those procedures with at least a moderate published experience include cutting setons and setons with second stage fistulotomy, fistulotomy with immediate sphincter repair, insertion of fibrin glue, insertion of a fistula plug, advancement flaps, and the ligation of the intersphincteric fistula tract (LIFT).

Novel techniques such as laser probe treatment to destroy the fistula epithelium and obliterate the tract, video-assisted anal fistula treatment utilizing a fistuloscope, and the use of stem cells are all being tested (Giamundo et al. 2014; Meinero and Mori 2011; Herreros et al. 2012; Meinero et al. 2014). However, the data about the results is limited and therefore the procedures are not included in the following discussion.

4.1 Setons

A seton is a foreign material inserted in a fistulous tract. The variety of materials and technical variations were recently reviewed (Subhas et al. 2012). In general, they are used as chronic loose setons to promote drainage, as cutting setons to gradually divide the intervening muscle, and as the first stage of a two-stage fistulotomy. The goal for chronic loose setons is not healing of the fistula; they are utilized primarily in patients with inflammatory bowel disease. Theoretically, cutting setons result in gradual division of the muscle with the resulting fibrosis limiting separation of the divided ends and therefore the risk of incontinence. When used as a first stage, fistulotomies are typically performed 6–8 weeks after insertion of the seton.

Incontinence rates of cutting setons were evaluated in a meta-analysis in 2009 (Ritchie et al. 2009). The literature was heterogeneous in terms of material and technique making pooling of the data difficult. The average reported incontinence rate was 12.3 %. Only one third of the studies reported the type and degree of incontinence; in those studies, the average incontinence rate was 32 %. Among the incontinent patients, 46 % were incontinence of flatus, 69 % of liquid stool, and 18 % of solid stool. It is possible that more detailed questioning of patients resulted in more reliable information. Because of the quality of the literature, it is difficult to identify whether patient or technique factors produce the difference in rates. However, in a systemic review, Vial and colleagues compared studies in which the internal sphincter was divided surgically at the time of placement of the seton to studies in which it was left intact (Vial et al. 2010). The evidence was deemed low quality but showed that when the internal sphincter was left intact, the fistula recurrence rate was 5 % with a 5.6 % rate of incontinence. When the muscle was divided, the recurrence rate was 2.9 %, but the incontinence rate was 25.2 %. The data suggests that preservation of the internal sphincter reduces the rate of incontinence with minimal increase in recurrence rates.

Most of the literature on two-stage fistulotomies is older (Pearl et al. 1993; Van Tets and Kuijpers 1995; Graf et al. 1995; Garcia-Aguilar et al. 1996; Rosa et al. 2006). The recurrence rates ranged from 3 % to 9 %. A wide range (5.4–65 %) of postoperative incontinence is reported. One of the studies with a high rate included only patients with high anal or rectal internal openings; the authors concluded that two-stage fistulotomy is not recommended for this group of patients (Van Tets and Kuijpers 1995).

Two studies compared the results of cutting setons to two-stage fistulotomy (Graf et al. 1995; Garcia-Aguilar et al. 1998); in both studies, the rates of incontinence were high (44–66 %). Graf and colleagues found significantly less incontinence after the two-stage fistulotomy (25 % versus 78 %), while Garcia-Aguilar and colleagues found no difference between the two options.

A final alternative is laying open the subcutaneous tract with the placement of a seton and removal of the seton after 6–12 weeks. One study reported that 86 % of 42 fistulas healed although 19 % recurred (Fung et al. 2013) which is similar to data reported in earlier studies (Thomson and Ross 1989; Eitan et al. 2009). No patient reported incontinence although standardized instruments were not used. A study with longer follow-up reported 65 % early success rate which decreased to 20 % over time (Buchanan et al. 2004).

4.2 Fistulotomy with Sphincter Reconstruction

Concern about incontinence led to trial of fistulotomy with immediate sphincter reconstruction. The technique was first described in 1985 (Parkash et al. 1985) with scattered reports until more recently (Perez et al. 2005; Jivapaisarnpong 2009; Roig et al. 2010; Arroyo et al. 2012; Ratto et al. 2013; Hirschburger et al. 2014). In addition, a randomized trial comparing this technique to advancement flap has been published (Perez et al. 2006). Recent series include a notable number of patients who present with incontinence suggesting a bias in patient selection (Table 9.5) (Perez et al. 2005, 2006; Jivapaisarnpong 2009; Roig et al. 2010; Arroyo et al. 2012; Ratto et al. 2013; Hirschburger et al. 2014). Recurrence rates range from 4.2 % to 12 %. With the exception of one study reporting no incontinence (Jivapaisarnpong 2009), the range is 12–35 % with the majority of patients reporting soiling or incontinence of flatus (Perez et al. 2005, 2006; Roig et al. 2010; Arroyo et al. 2012; Ratto et al. 2013; Hirschburger et al. 2014). The randomized trial found no difference in recurrence or incontinence between advancement flaps and this technique (Perez et al. 2006). One concern about this technique is whether the long-term functional results will mirror the deterioration of function seen in long-term follow-up of sphincteroplasty for obstetrical injury (Glasgow and Lowry 2012).

4.3 Fibrin Glue

Fibrin glue is inserted in the fistula tract to seal the fistula while avoiding any division of the sphincter muscle. Successful fistula closure varies from 14 % (Buchanan et al. 2003) to 74 % (Tinay and El-Bakry 2003). Studies with longer follow-up report high rates of recurrence with time (Cintron et al. 2000; Buchanan et al. 2003; Sentovich 2003). Investigators rarely report data on continence, but a randomized trial found no postoperative incontinence in patients treated with fibrin glue (Lindsey et al. 2002). This option is rarely utilized since other procedures have been introduced.

4.4 Fistula Plugs

Two fistula plugs are commercially available: the Cook Surgisis® AFP™ plug and the Gore BIO-A® plug. Much more data is available for the Cook plug. The reported success rates for the Cook plug vary considerably from 14 % (Safar et al. 2009) to 83 % (Champagne et al. 2006). A similar variation in success is reported for the Gore plug with a range of 16 % (de la Portilla et al. 2011) to 73 % (Ratto et al. 2012). No change in continence is reported in the few studies providing continence data (Christoforidis et al. 2009; Garg 2009; Schwandner et al. 2009 Chung et al. 2010; McGee et al. 2010; Schwandner

Table 9.5 Results of fistulotomy with sphincter reconstruction

	N	Recurrence (%)	Preop FI (%)	Postop FI (%)	
Perez 2005	35	5.7	31.4	14.2	Nine of 11 incontinent patients improved; three continent patients worsened
Perez 2006	30	7.1	18	32	
Jivapaisarnpong 2009	33	12	0	0	
Roig 2010	75	10.6	20	21.3	No change in FIQL
Arroyo 2012	70	8.5	31	21.4	Fifteen of 22 incontinent patients improved; eight of 40 continent patients worsened
Ratto 2013	72	4.2	4.2	15.2	Primarily soiling
Hirschburger 2014	50	10	8	12	Incontinence of flatus only

et al. 2008; van Koperen et al. 2011; Ommer et al. 2012; Madbouly et al. 2014).

4.5 Advancement Flaps

Endorectal advancement flaps are frequently performed as sphincter sparing procedures for complex or high transsphincteric anal fistulas. The flaps may be mucosal, partial thickness or full thickness.

A review article published in 2010 included 35 studies over a 30-year interval involving 1654 patients (Soltani and Kaiser 2010). The weighted average success was 79 % with an average follow-up of 29 months; success rates ranged from 36 % to 98 %. Correlation of the type of flap with success rate was inconclusive. The weighted average for new incontinence was 13.3 % with a range of 0–35 %. The authors did note that only half of the 35 studies provided data about continence. In addition, the data reported generally did not include the use of standardized surveys, scoring systems, or anal manometry. Data from representative series is presented in Table 9.6.

One group evaluated changes in anorectal anatomy and manometry after fistula surgery including advancement flaps (Roig et al. 2009). The flaps were accompanied by fistulectomy, so the results may not be completely transferrable to advancement flaps alone. New incontinence occurred in 52.4 % of 45 patients. Twelve patients developed new internal anal sphincter defects, and four patients had new external sphincter defects visible on anal ultrasound. Mean resting pressures but not mean squeeze pressures were significantly decreased. The authors suggest either the use of anal retractors or division of the internal sphincter for full thickness flaps as the reason for the new internal sphincter defects. A number of previous surgeries (Mizrahi et al. 2002) and postoperative ectropions (Soltani and Kaiser 2010) have been also proposed as contributing factors.

Since the review article in 2010, several other relevant reports have been published. One meta-analysis included six studies of 408 patients comparing anal fistula plugs to mucosal advancement flaps (Leng and Jin 2012). The success rates were statistically similar. Three of the six studies reported data on continence. In the combined groups, 1 of the 83 patients undergoing fistula plug insertion and 13 of the advancement flap patients developed new incontinence for a risk difference of −0.08. Two randomized trials compared mucosal flaps to the LIFT procedure (Mushaya et al. 2012; Madbouly et al. 2014). In

Table 9.6 Incontinence after advancement flaps

Study	Flap type	N	New incontinence	% new soiling
Mizrahi 2002	Partial thickness	106	9 %	NR
Dubsky et al. 2008	Mucosal Full thickness	25 20	16 % minor, 16 % major 20 % minor, 5 % major	NR
Christoforidis 2009	Partial thickness	23	35 % minor, 17 % major	NR
Roig 2009	Type not specified/with fistulectomy	45	52.4 %	NR
Khafagy et al. 2010	Partial thickness Mucosal	20 20	10 % 0 %	NR NR
Abbas 2011	Partial thickness	19	27.8 %	NR
van Koperen 2011	Mucosal	29	No change in mean FI scores	12 % (NS)
Jarrar 2011	Horizontal partial thickness	77	No new FI Ten FI patients improved	No new soiling 3/17 soiling resolved
Mushaya 2012	Partial thickness	14	7 %	
Madbouly 2014	Partial thickness	35	8.6 % minor	0

the first study, five patients (14 %) of the flap patients reported new incontinence at 4 weeks, and two (6 %) patients had persistent incontinence at 12 weeks (Madbouly et al. 2014). In the second study, one patient reported minor incontinence after a flap procedure. None of the LIFT patients developed new incontinence in either study. In a retrospective review of the outcomes of 75 patients with a mixture of types and etiologies of fistulas undergoing advancement flaps, an overall success rate of 93 % was reported; primary success after one flap was 72 % and the remainder healed after two or more flaps or fistulotomy (Jarrar and Church 2011). Only 43 % of patients reported normal continence preoperatively; after surgery, 57 % reported normal continence. However, 34 % of patients were incontinent after one repair and 56 % of patients after two or more repairs. Incontinence was associated with decreased quality of life scores.

In general, the data regarding continence and advancement flaps is limited and low in quality. It is clear that preoperative assessment is important as a significant percentage have impaired continence before the procedure (Jarrar and Church 2011). While most studies report some worsening of continence after an advancement flap, the study with the most detail reported improvement at least after the first repair (Jarrar and Church 2011). Further prospective evaluation with detailed information utilizing standardized instruments and anatomic evaluation of the anal sphincter are necessary to better identify the risk related to the procedure and whether the risk is related to technical differences, preoperative status, number of repairs, or other factors.

4.6 Ligation of Intersphincteric Fistula Tract Procedure

Ligation of intersphincteric fistula tract (LIFT) is another sphincter sparing technique to treat fistulas. Through an incision in the intersphincteric groove, the tract is identified in the intersphincteric space. It is then isolated, divided, and suture ligated on both sides. Five meta-analyses or systemic reviews have been published in recent years (Vergara-Fernandez and Espino-Urbina 2013; Yassin et al. 2013; Alasari and Kim 2014; Hong et al. 2014; Murugesan et al. 2014). Four of the five publications report mean healing rates which vary between 71 % and 81.37 % (Vergara-Fernandez and Espino-Urbina 2013; Yassin et al. 2013; Alasari and Kim 2014; Hong et al. 2014). Murugesan and colleagues argued that the data was too heterogeneous to be pooled and reported a range of 40–94 % success. Neither of the studies that documented continence reported any change (Vergara-Fernandez and Espino-Urbina 2013). As noted in the section above, neither of the two randomized studies of advancement flaps and LIFT reported any incontinence in the LIFT patients.

4.7 Summary

Fistulotomy has the highest reported healing rates when various surgical options are compared (Garcia-Aguilar et al. 1996; van Koperen et al. 2008; Roig et al. 2009). However, incontinence is reported after fistulotomy in 18–52 %. That complication is more common in complex fistula patients but occurs after treatment of simple fistulas as well. Patients with higher risk of incontinence include those with baseline incontinence (Roig et al. 2009; Jordán et al. 2010) or a high transsphincteric fistulas (Abbas et al. 2011; Koehler et al. 2004; Pezim 1994) and anterior fistulas, older patients (Abbas et al. 2011), and women with previous vaginal deliveries (Hasegawa et al. 2000). Particularly in those patients, other options need to be considered.

Any fistula surgery requires a thorough discussion with the patient about the expected healing rates and risk of incontinence. Some patients will accept the risk of incontinence to avoid repeated surgery. Others are accepting the need for multiple procedures in order to avoid incontinence. Overall, there is a trend away from procedures that involve division of the sphincter muscle, but they still may represent half of the fistula surgery performed (Blumetti et al. 2012).

For low-risk patients with intersphincteric fistulas, fistulotomy offers a high healing rate but up to 20 % risk of incontinence. In higher-risk patients, the risk of incontinence even for these “simple” fistulas may not be acceptable. Patients may choose to live with their symptoms or undergo an advancement flap. For most patients with transsphincteric fistulas, a LIFT procedure offers reasonable success rates with no documented risk of impaired continence. For patients with evidence of an internal sphincter defect at the fistula site or a supra-sphincteric fistula, insertion of a fistula plug does not affect continence but in many studies has a relatively low success rate. Perhaps new plug designs will improve those outcomes. Fistulotomy with immediate sphincter repair and endorectal advancement flap are alternative options. In some countries, the medical legal environment limits acceptance of the former. While cutting setons and two-stage fistulotomies are alternatives, they are less commonly used given the rates of incontinence and availability of alternatives. Novel techniques being evaluated now may provide an answer for the more complex situations and presumably further the trend toward sphincter sparing procedures. In the meantime, the choice of the procedure must be tailored to the anatomy of the fistula, the functional and anatomic status of the sphincter, and patient preference.

5 Conclusion

The literature clearly demonstrates that functional issues may be a significant issue after anorectal surgery. Treatment plans must be carefully tailored to the patient. Medical management and Botox are sphincter-saving options for patients with anal fissures. LIS provides a high cure rate but should be avoided in patients with low resting pressures or previous sphincter injury. For patients requiring hemorrhoid surgery, careful surgical technique is critical. Decision making about patients with anal fistulas may be complicated. Treatment plans must be based upon patient symptoms and anatomy as well as careful informed consent about the possible need for multiple procedures versus the risk of incontinence.

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Part III

Diagnosis

Patient's History Data Collection in Coloproctology

10

Lilli Lundby

Contents

Abstract

For experienced specialists including coloproctologists, it may seem trivial and basic to study the optimal way to take a good history from a patient. However, a detailed description of the patient's symptoms is the key to reach an accurate medical history and a correct diagnosis, which is essential to quality patient care and treatment. Accurate and standardized data collection is fundamental for quantifying symptoms and for measuring the outcome of medical or surgical treatment. Generally, questionnaires and data collection forms are used, but new technological developments in the collection of patient-reported outcome such as tablet computers and smartphone applications linked to electronic charts are currently evolved.

1 History Taking

The definition of history taking is the complex process of interviewing a patient to determine symptoms and to assess factors affecting health status. Obtaining an accurate patient's history is absolutely essential to reach the underlying cause of the problem, thereby establishing a correct diagnosis with the aim of providing the right medical care. The basis of an accurate medical history is good communication between doctor and patient, and that means the patient allows the doctor to ask the necessary questions in order to

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obtain open and honest answers. The science in history taking is asking the right questions and then interpreting the answers from the patient.

A much quoted citation is:"Listen to your patient and she will tell you the diagnosis." The doctor should be cautious not to dominate the history taking with a series of detailed questions, which may limit the patient to answer instead of speaking from the heart. The gold standard in history taking is open questions that encourage the patient to tell what are of concern to her/him, e.g., "How are you"? Open questions can also provide more specific information by, e.g., asking "Tell me about your bowel problems." Questions with options are sometimes needed to clarify the problem such as "what was the consistency of your stools?". Leading questions directing the patient toward an answer or suggesting possible answers may be necessary, but the risk is that the doctor will bias the answers from the patient and include his own interpretation, and this should be avoided. Plain language or language that is appropriate to the patient's education should be used and only one question asked at a time in order not to confuse the patient.

At the end of history taking, it is advisable to summarize what the patient has told about the present complaint and the main symptoms and furthermore have statements approved by the patient so that incorrect assumptions can be avoided. This allows you to correct any wrong information and to further expand on important aspects. Finally, to confirm that shared understanding has been obtained, the patient should be encouraged to consider whether there are more questions that are not answered.

Taking history from a patient is a very important skill for a doctor to master no matter which area of specialization you represent. Within the field of coloproctology, many of the bowel symptoms that patients present are taboo, and they find it embarrassing to consult a healthcare professional and talk about bowel problems. When a topic is considered taboo, it is not discussed openly, and this is particularly true regarding bowel dysfunction. Conditions such as fecal incontinence and urgency are especially distressing and socially debilitating so the doctor should be respectful and aware of the patient's comfort level. In general, patients are reluctant to discuss bowel habits, and the doctor should be extra aware and compassionate to make the patient feel comfortable about sharing her problems and breaking the taboo.

In coloproctology the number of symptoms that the patient presents is limited. Changes in bowel habits, including diarrhea or constipation or a change in stool consistency, are common symptoms. Anal or rectal bleeding is common and may occur for many reasons. Loss of appetite, unexplained weight loss, and feeling tired or unwell at the same time as noticing any other abdominal symptoms are more unspecified symptoms. First of all it is essential to exclude that the patient suffers from a malignant disease. On suspicion of malignancy, the cancer investigation program should be initiated as soon as possible. Any inflammatory bowel disease, diverticular disease, or functional bowel disorders such as fecal incontinence, constipation, obstructed defecation, or irritable bowel syndrome should be considered. Anal mucosa prolapse or rectal prolapse must be excluded by a thorough questioning about symptoms of prolapse associated with passing stool or with physical exertion and by performing a physical examination with the patient placed on a toilet chair. Anorectal pain, perianal swelling, discharge, or bright red bleeding indicates a proctologic condition, which could be fissures, anal fistulas, or hemorrhoids.

For each patient, the diagnosis is made after a thorough analysis of the symptoms presented in the history. Differential diagnosis must be considered, and a physical examination together with special investigations should support the tentative diagnosis (Table 10.1).

Table 10.1 History – crucial issues

Let the patient tell her story
Open questions
Keep it simple
Keep an open mind
Reconsider tentative diagnosis

2 Structured History Taking

History taking in coloproctology requires a systematic approach. Following a general framework ensures that you structure the history and maintain an overview during history taking and that you do not miss any important information.

2.1 Present Complain

The site and time of onset of a symptom and the evolvement to current presentation are essential to determine. When did the symptom start and was it a sudden or gradual, constant, or intermittent onset? A thorough description of the character and severity of the symptom is important to present together with information on periodicity and frequency. Furthermore factors that predispose, provoke, or alleviate symptoms should be described. The doctor may define symptoms differently than the patient; therefore, it is important to check that terms such as diarrhea, incontinence, constipation, pain, and discharge are interpreted in the same way by the patient.

2.2 Past Medical History

Recording of any previous hospital admissions, anorectal diseases, cancer or polyps, abdominal operations, and possible medical treatments including radiotherapy are essential. Past medical history should ascertain whether former abdominal surgical procedures have been performed and should report the outcome of the surgery. General medical conditions and comorbidities such as diabetes and metabolic disorders can cause abdominal symptoms and these should be recorded. If major surgery is considered, patients should be questioned about symptoms of respiratory and cardiac diseases.

2.3 Family History

Patients with a family history of large bowel cancer among first-degree relatives have a three- to four-time greater risk of developing cancer than in families without a history of cancer. Taking a family history can be used as a tool to identify these individuals and to decide who should be referred for genetic counseling. Furthermore information on inflammatory bowel conditions, hereditary bowel diseases, and other significant genetic conditions should be recorded.

2.4 Obstetric History

The past obstetric history is relevant for patients with fecal incontinence and other disorders of the pelvic floor. Number of births and reports on tears of the perineum or the anal sphincter are significant.

2.5 Drug History

Drug history is to establish which regular medication the patient is taking including dosage and frequency. A thorough drug history provides an opportunity to review the need for the patient to take medication and to find out whether the patient is actually taking them. It is essential to know whether a patient is taking anticoagulants if a biopsy or surgical treatment is contemplated, and patients on oral contraceptives should be identified. It is important to note that several different drugs may affect gastrointestinal motility and lead to functional gastrointestinal disorders. Any allergy should be recorded.

2.6 Personal and Social History

This section documents factors in the patient's lifestyle, personal habits, and environment, which may reveal a risk of developing or having an illness. Information to be gathered could include: smoking and alcohol intake. How many cigarettes a day has the patient been smoking and for how many years. Which type of alcohol and how many units a week has the patient been drinking? Activities of daily living, family relationship, and occupation should be explored. Has the patient been exposed to any HIV risk factors?

2.7 Review of Systems

Systemic enquiry consists of a screen for symptoms in other body systems that are not covered in the present complaint and may be missed. It is essential to be selective and focus on the system relating to the actual problem or to a differential diagnose. The main symptoms that should be covered are cardiovascular, respiratory, and gastrointestinal, which is covered in the present complain, urinary, gynecological, CNS, musculoskeletal, and dermatology.

Reviewing what the patient has told should complete history taking. Important points should be repeated so that the patient can correct if there are any misunderstanding or errors.

Keynote: A well-taken history will often provide more clues to the diagnosis than the physical examination. It provides a basis for confidence and trust between the patient and the doctor (Table 10.2).

3 Data Collection

Generation of a diagnosis is the basis for medical decisions and for the opportunity to solve a clinical problem. The diagnosis is based on the collection of information about symptoms, history, and objective findings on the physical examination, and these data must be gathered and organized in patient records. Maintaining complete and detailed medical records for each patient is essential for the continuity of the further treatment.

Table 10.2 Structure of history taking

Chief complaint: Patient's presenting symptoms and present illness
Past medical history: Patient's previous hospital admissions, surgical, and general medical history
Family history: Cancer, inflammatory bowel diseases, hereditary bowel diseases, other genetic conditions
Obstetric history: Number of births, any perineal or anal sphincter lesions
Drug history: Patient's medications including allergies
Personal and social history: Patient's lifestyle, personal habits, and social environments. Use of tobacco and alcohol. Exposed to any risk factors.
Review of systems: Cardiovascular, respiratory, gastrointestinal, urinary, gynecological, CNS
Review of patient's history

Collection of clinical data for the patient record can be achieved through several different patient-reported outcome measures that include: paper forms completed by the patient at the first visit, local electronic medical data records filled in at site, or central Web-based systems.

To standardize accurate data collection of patient history and physical findings, questionnaires or data collection forms should be used. A wide selection of scores has been constructed and they allow grading of severity of symptoms and co-assessment of quality of life. The value of the data and subsequently of the patient's record relies on the quality and integrity of that instrument. If significant data points are not registered, a meaningful analysis and diagnosis may not be possible.

Collecting data in coloproctology from patients with functional disorders such as fecal incontinence questionnaires including well-defined scoring systems like the St Mark's Incontinence Grading System, the Wexner Continence Grading Score, or the ICIQ-B Score should be used routinely(Vaizey et al. 1999; Jorge and Wexner 1993; Cotterill et al. 2011). For patients with constipation or obstructed defecation, the Cleveland Clinic Constipation Score, Patient Assessment of Constipation Symptoms (PAC-SYM), and the Obstructed Defecation Score are recommended (Agachan et al. 1996; Frank et al. 1999; Altomare et al. 2008). Ideally symptom-specific questionnaires evaluating quality of life such as the Fecal Incontinence Quality of Life (FIQL) scale or the Patient Assessment of Constipation Quality of Life (PAC-QOL) should also be incorporated (Rockwood et al. 2000; Marquis et al. 2005).

These instruments can be used to quantify the symptoms that the patient presents at the first visit and to measure the outcome or the changes of symptoms following medical or surgical treatment and furthermore provide a disease specific evaluation of the quality of life. Additionally they can facilitate comparison between different treatment modalities and different units.

While symptom questionnaires provide a snapshot of bowel habits, they may not adequately assess the variability and alterations in bowel function over time. Furthermore questionnaires are filled in retrospectively with the risk of recall bias. Bowel habit diaries are prospectively filled in often during a 3-week period and measure the actual number of bowel accidents, urgency, and defecations, and the calculated variables often serve as an endpoint for clinical decision making.

A technological advancement during the recent years is collection of patient-reported symptoms with a tablet linked to an electronic chart. The increased use of smartphone application within the clinical environment provides opportunity to integrate this new technology into data collection in coloproctology. Medical smartphone apps can be a particularly suitable measuring tool for conditions with frequent registration and with symptoms that fluctuates over time.

Despite the rapid technological developments in smartphone apps and the increasing trend to self-monitoring all sorts of health parameters, only a single review from 2012 in colorectal themed apps could be detected. O'Niell and Brady identified 68 individual apps, among which five were duplicates. Only 29 % were rated by customer satisfaction rating, and only one third of the apps had documented medical professional involvement in their design and content, and only three of these were free to be downloaded (O'Niell and Brady 2012). Major colorectal diseases, such as bowel cancer, were poorly represented, whereas apps for irritable bowel syndrome were more common.

The development and introduction of smartphone applications are currently in progress, and when the legal aspects on data security are solved, there is no doubt that smartphone apps will be a future tool in data collecting in coloproctology.

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Visiting a Coloproctology Patient: Rules and Suggestions

11

Carlo Ratto, Francesco Litta, Lorenza Donisi, and Angelo Parello

Contents

Abstract

A careful clinical examination remains of crucial importance in a patient affected by coloproctological diseases. These conditions have a significant impact on the patients' quality of life and, moreover, are often associated to personality disorders or real psychiatric conditions: some measures are therefore needed to minimize the impact of the visit and to establish immediately an effective relationship of mutual trust.

It is therefore clear that the choice of where and how to visit a coloproctology patient is very important to properly address the diagnostic-therapeutic approach. A full coloproctologic examination should always provide a series of well-defined phases, with the evaluation of several different aspects of the same disease. For many common diseases treated in a proctology clinic, a proper clinical examination can address, or not, the patient to a subsequent assessment by radiology or anorectal physiology testing.

Growing *literature* evidences support the surgeon in the diagnosis of colorectal diseases and have highlighted the importance of treating these conditions in a specialist colorectal context.

First outpatient visit of a coloproctology patient is often a decisive phase, which inevitably affects all subsequent stages of his treatment. In this context, physical evaluation in a patient affected by

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coloproctological disease requires specific skills and procedures. In fact, a careful clinical examination, including a correct psychological approach to the patients, remains of crucial importance. Moreover, an expert colorectal surgeon should be able to perform physiology testing and make a diagnostic integration of physical and imaging examinations.

This chapter will be focused on how to perform a complete clinical examination in a coloproctology patient, with the support of the literature available on this topic. A detailed description of clinical findings in each disease has been already given elsewhere in this book.

1 Psychological Aspects

In the office is established the first contact between the patient and the specialist: at that time the patient provides basic information, and it is important to establish immediately a relationship of mutual trust. To achieve this, it is often necessary to detect, and overcome, certain patient's states of mind: anxiety and fear or, conversely, reluctance and superficiality could make it harder for an objective assessment of the real clinical conditions. These difficulties may emerge as both medical questions and the visit may be embarrassing for the patient. Also, there is a clear disparity between the physician and the patient with regard to the knowledge about his/her disorder/disease.

Coloproctological diseases have a significant impact on the patients' quality of life (QoL), and this can obviously cause in them quite obvious strong emotions. Nevertheless, it is a common experience for a colorectal surgeon to visit patients affected by real psychiatric or emotional disorders, and *literature* supports this empiric finding. Recently, Sit et al. described that about a third of patients affected by hemorrhoids or anal fissure had a "type D (distressed) personality," and this had a great impact on their QoL, because "type D personality" is characterized by tendency to experience social inhibition and negative emotions, such as hostility, depression, and anger (Sit et al. 2014a, b). Similarly, in patients undergoing defecography for evacuation disorders, there was a high rate of emotional disturbances and psychiatric illness, when the examination showed negative findings (Kashyap et al. 2013). Other clinical conditions commonly managed in a coloproctology outpatient clinic, such as Crohn's disease, ostomy, or colorectal cancer, were strongly associated with psychiatric comorbidity (Sprangers et al. 1995; Maconi et al. 2014).

It is also interesting to underline the notable impact on the lives of those who cure these patients: a study investigating psychological conditions of colorectal surgeons and nurses of the British National Health Service showed that stress, burnout, and psychiatric disorders have a prevalence of about 30 %, and this was not directly related to the treatment of neoplastic diseases (Sharma et al. 2008).

It is questionable how to minimize the impact of the clinical examination in these patients already sorely tried by their condition (Table 11.1). Some interesting tips could be suggested:

- Avoid anticipations about possible negative scenarios about the disease, even before visiting the patient.
- Explain to the patient the steps of the visit which is going to be subjected.
- Do not fully undress the patient: this can reduce his embarrassment.
- After the visit, talk to the patient "face to face and eye to eye;" this strengthens the relationship of mutual trust.
- Provide the patient information brochures on its pathology; this reduces the knowledge gap that exists between the surgeon and the patient.

Table 11.1 How to minimize the impact of the clinical examination in a coloproctology patient

Tip	Effect
Avoid anticipations about negative scenarios	It reduces anxiety and fear
Explain the patient every step of the visit	It reduces the fear of pain
Do not fully undress the patient	It reduces the embarrassment
Talk to the patient "face to face and eye to eye"	It strengthens the relationship of mutual trust
Provide the patient information brochures	It allows the patient to better understand his condition

2 Where to Visit a Coloproctology Patient

A coloproctology clinic should have some specific characteristics and, above all, should have some essential equipment. A room too small and packed with all the equipment (Fig. 11.1a) or, conversely, any aspecific general surgery office (Fig. 11.1b) is not suitable. Everything a colorectal surgeon should have is clearly described elsewhere in this book; however, specific examination tables, instrumentation for the anoscopy and proctoscopy, fistula probes, and, if possible, an ultrasound scanner equipped with an endoanal/endorectal probe could be particularly useful (Fig. 11.2) (García-Olmo and Pascual Migueláñez 2010).

With regard to the positioning the patient for proctologic examination, some practical considerations are needed. To obtain a correct lithotomy or prone (jackknife) positions, specific examination beds are needed; however, they are not always possible to have in an outpatient clinic, mostly due to their costs; on the other hand, the knee-chest position could result very embarrassing for the patient. On the contrary, the left lateral Sims' position could be obtained easily and quickly on any medical bed and is usually well tolerated by the patient. In a specifically designed study, a questionnaire about medical positioning was administered to 178 patients evaluated for a coloproctological disease: in about two thirds of the cases, the Sims' position was the preferred choice (Gebbensleben et al. 2009). However, the same research group stated that the knee-chest position may allow a better and detailed patient evaluation (Kuehn et al. 2009).

In our opinion, the Sims' position is comfortable for both the patient and the coloproctologist and could fit the in-office diagnostic necessity in the majority of patients; however, if any doubt should arise, different position should be suggested.

3 How to Perform a Coloproctologic Examination

Today, the diagnostic value of the complete abdominal examination is more limited than in the past mainly because the more frequent use of imaging tests and screening campaigns have made the diagnosis of colorectal diseases increasingly accurate and early. Furthermore, studies have shown that a complete abdominal examination may fail in the correct diagnosis also in acute clinical conditions or in cases of malignancies

Fig. 11.1 Where to visit a coloproctology patient: a room small and too packed with instruments (**a**) or a generic general surgery (**b**) office is not ideal

Fig. 11.2 Where to visit a coloproctology patient: a specific table, anoscopy/proctoscopy, probes, and, if possible, an endoanal scanner are needed

(Laurell et al. 2006; Gans et al. 2015). In fact, it is increasingly rare today to diagnose a colorectal cancer, by detecting a marked hepatomegaly, an abdominal mass, or malignant ascites during the physical examination. Nevertheless, a careful and targeted abdominal examination could be very useful: with the abdominal inspection, the presence of existing abdominal scars (Pfannenstiel, median laparotomy, etc.) or abdominal hernias (umbilical or incisional hernia) should be detected. In this way, it will be possible to better plan any abdominal surgery with regard to the choice of the surgical incision, the positioning of trocars, or the choice of the type and position of a potential stoma.

On the other hand, the complete proctologic clinical examination provides, first, a careful inspection of the perineal area and, then, the digital rectal examination (Table 11.2) (Talley 2008). Following the patient's correct positioning, the buttocks are moved gently, and it is therefore possible to observe the perianal skin and anal mucosa, searching for the presence of lumps or swellings (Fig. 11.3), openings of an anal fistula (Fig. 11.4), the features of a hidradenitis suppurativa (Fig. 11.5), perineal scars, and skin tags (Fig. 11.6). Then, the patient is asked to strain and to observe hemorrhoidal piles prolapse (radial lines on the prolapsing tissue) or a true rectal prolapse (concentric lines) (Fig. 11.7). Finally, it is also possible to evaluate the neuromuscular integrity of the perianal region eliciting a reflex, the so-called anal wink (see below in the section on Fecal Incontinence).

After a gel lubrication of the finger and the perianal skin (Fig. 11.8), the digital rectal examination should ensure:

Table 11.2 A complete proctologic physical examination

Phase	What to observe
Inspection: (a) At rest, with the buttocks moved	Perineal scars, skin tags, openings, lumps, swellings, dermatological problems, warts, fissures
(b) Straining	Rectal or hemorrhoidal prolapse, perineal descent, prolapsing polyps
(c) "Anal wink" reflex	Contraction of the sphincter complex
Digital rectal examination: (a) Evaluation of anal pressures	Resting anal pressure, squeeze pressure, sphincter complex relaxation
(b) Evaluation of the anal and rectal mucosa	Indurations, fibrosis, thickening, ulcerations, protrusions, irregularities, evocated pain
(c) Evaluation of the secretions on the finger	Blood, pus, serum, mucus, feces color

- Evaluation of the resting anal pressures, squeeze anal pressure, and relaxation of the sphincter anorectal complex. At this stage, it may already be possible to make the diagnosis by correlations between different information already acquired; for example, the palpation of a discontinuity of the internal anal sphincter covered by a non-ulcerated anal mucosa, together with a low resting pressure, could be related to a previous anal surgery described by the patient; the presence of an anterior perineal scar in a woman with low squeeze pressure may be related to a history of difficult vaginal delivery occurred many years before; an absent or incomplete relaxation at the straining, together with the absence of a rectocele or a rectal intussusception, may suggest the clinician a functional, rather than anatomic, constipation.

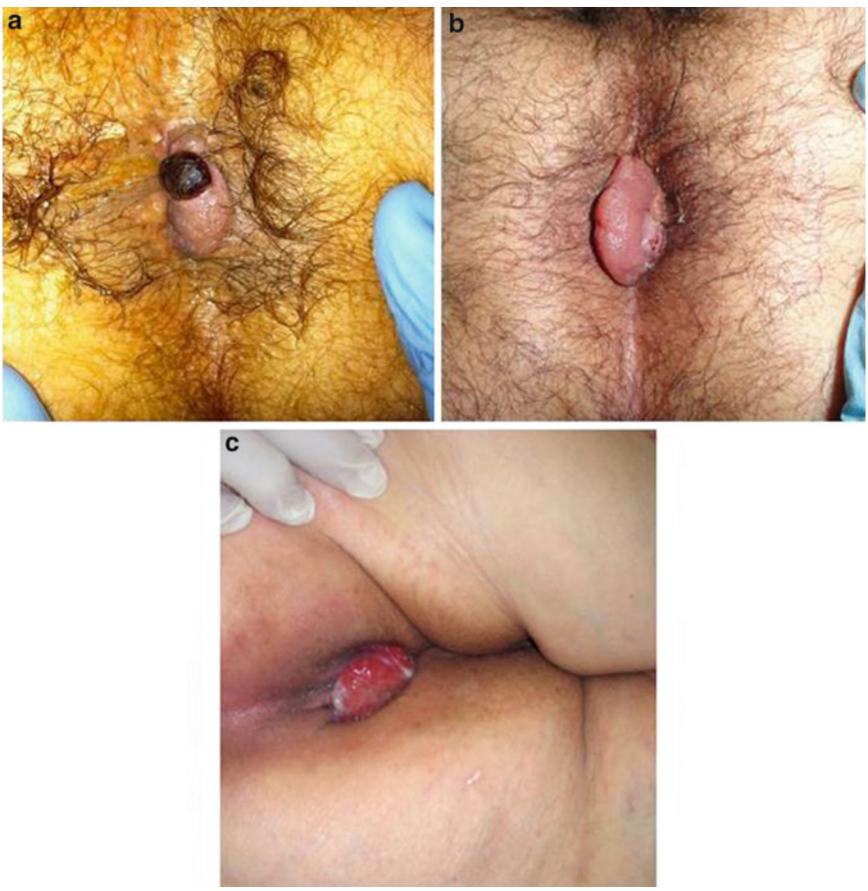

Fig. 11.3 Anal lumps or swellings: hemorrhoidal thrombosis (**a**, **b**), anal cancer (**c**)

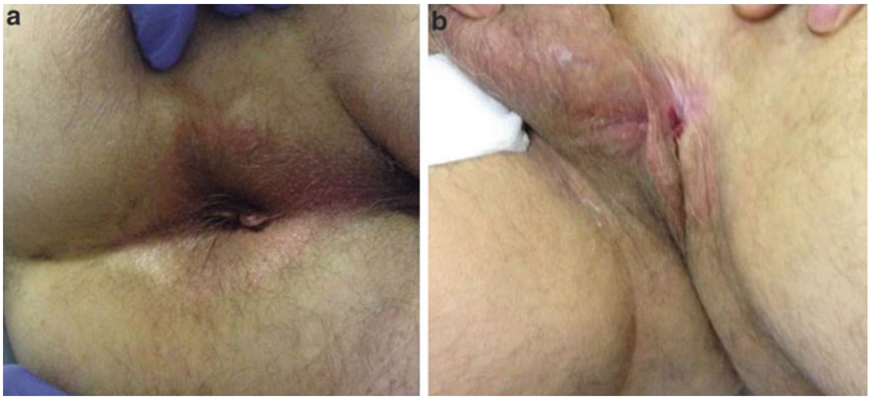

Fig. 11.4 Complex anterior anal fistula in a male patient: no external anal opening is indentified near the anal verge (**a**); a large orifice is located very far from the anal verge, near to the scrotum (**b**)

Fig. 11.5 Features of a hidradenitis suppurativa

- Evaluation of the anal and rectal mucosa to assess the presence of induration, fibrosis, thickening, ulceration, protrusions, and irregularities.
- Evaluation of the finger after his removal, to assess the presence of blood, pus, mucus, and serum secretions and to observe the feces color.

A colorectal surgeon or endoscopist should never forget to evaluate the prostate in a male patient: any alteration of gland consistency, irregularity, the size, and the presence of nodules should be reported to the patient, and a urological examination should be recommended (Deshpande et al. 2009). In women it is useful to know the status of their menstrual cycle before performing a digital rectal examination: mainly in the immediate proximity of menstruation, the uterine cervix could be distinctly palpated, which should therefore not be considered a malignancy.

4 Literature Evidence on the Role of the Clinical Examination

A detailed description of clinical findings associated to each colorectal disease is given in another chapter. In this section, the available literature on the role of the clinical examination in some common proctologic disease is summarized, trying to report its potentials, but also its limitations.

- *Anal Fissure*. This condition can be usually diagnosed, thanks to a simple anal inspection and careful digital examination of the anal canal (Fig. 11.9). From the literature, however, it emerges as the physical examination could be imprecise in the evaluation of the sphincter tone. In a study by Jones et al., 40 patients affected by anal fissure were prospectively evaluated by digital anal canal examination and anorectal manometry. A comparison between the clinical and manometric assessment of maximum resting pressure showed that in only 15 out of 35 patients considered to be affected by sphincter hypertone, the diagnosis of anal fissure was confirmed after the physiology testing, so the specificity for

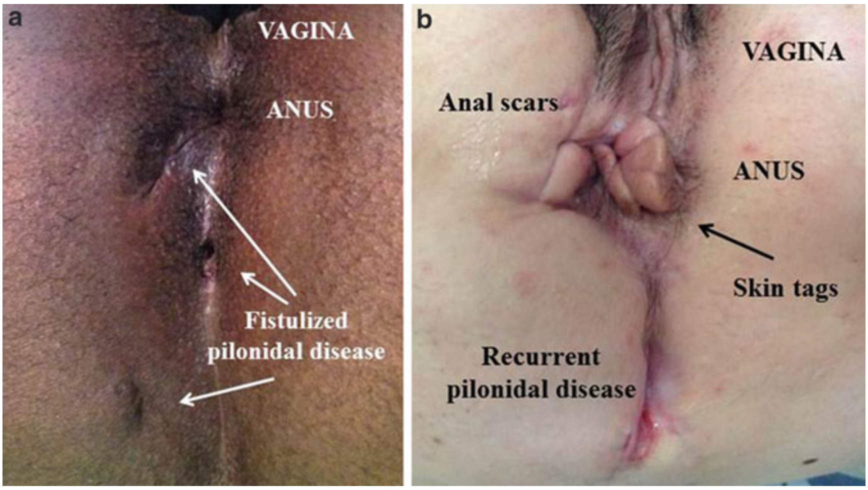

Fig. 11.6 Fistulized pilonidal disease: an orifice is close to the anal verge; an anal fistula should be excluded (**a**); recurrent pilonidal disease and a recurrent anal fistula in a patient submitted to multiple failed fistulotomies (**b**)

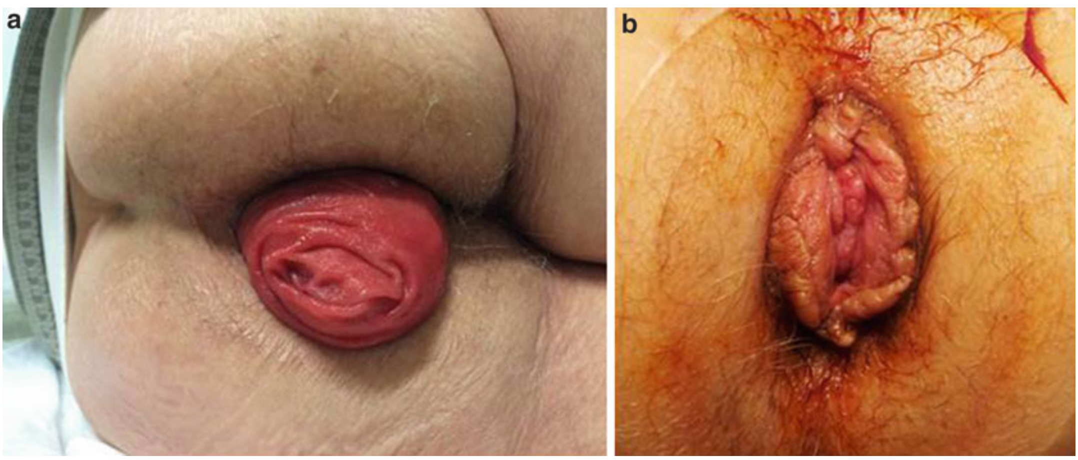

Fig. 11.7 Difference between hemorrhoidal and rectal prolapse: concentric (**a**) or radial (**b**) lines on the prolapsing tissue

hypertone was only 16 % (Jones et al. 2005). Moreover, a larger number of patients than it normally is assumed had a normal or low resting pressure; this could have a very important value in the planning of fissure management, in particular concerning decision-making process toward surgery because in these patients the risk of postoperative fecal incontinence is not negligible. Similarly, other findings showed that the reliability of digital anal canal examination in the identification of sphincter tone was low (Eckardt and Kanzler 1993). For these reasons, performing an anorectal manometry in a patient that should undergo an anal sphincterotomy could be of great interest.

- *Anal Fistula*. A careful collection of the clinical history of the patient, together with an accurate physical examination, may have a decisive role in the treatment of anal fistulas. Objectives are to identify an acute sepsis (Fig. 11.10), to identify the internal and external anal openings, and to define the potential course of the fistula tract. However, some difficulties could emerge: the digital anorectal examination, and, even more, the fistula probing, could be painful for the patient, thus limiting the accuracy; moreover, the fistula probing could be dangerous, given the risk of a false fistula tract creation (Fig. 11.11).

The identification of the internal anal opening is of great importance in anal fistula surgery, as showed by many authors (Garcia-Aguilar et al. 1996; Sygut et al. 2010; Jordán et al. 2010). Since 1887, the Goodsall's rule supports the proctologist during the identification of the internal anal opening and the fistula tract (Zbar 2009) (Fig. 11.12). However, recent studies have questioned the predictive value of this rule. Cirocco et al. conducted a study on 216 patients, showing that 90 % of the cases with an external opening posterior to the transverse anal line had an anal fistula tracking to the midline (in accordance with the rule); however, the rule was observed in only one patient out of two when the external opening was anterior (Cirocco and Reilly 1992). Similar quite disappointing results were showed by Gunawardhana and Deen in 2001: the predictive accuracy of the Goodsall's rule was low (59 %), and, in this study, it was lower when the external opening was posterior (Gunawardhana and Deen 2001). Finally, it seems that the Goodsall's rule fails in the same way in patients suffering from a Crohn's or non-Crohn's related fistula, even if the failure is higher in women with an anterior external opening (Coremans et al. 2003).

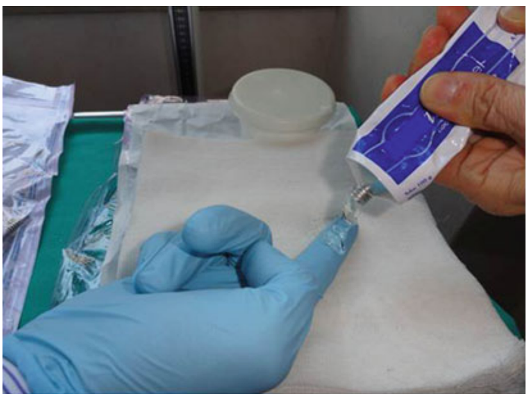

Fig. 11.8 Generous gel lubrication of the finger and of the perineal skin before a digital rectal examination

Other useful clinical data emerged from a study by Becker et al. in 2006: "complex fistula patients" were older than "simple fistula ones" (44 vs. 37.6 years), a posterior external anal opening was often associated to a posterior curved tract leading to the posterior midline of the anal canal, complex fistulas were associated to a higher number of operations (3.38 vs. 1.25), and, more interestingly, the distance between the external opening and the anal verge was longer in complex fistulas (4.4 vs. 2.8 cm), the latter evidence confirming a common observation in a proctology clinic (Becker et al. 2006). For this reason, the

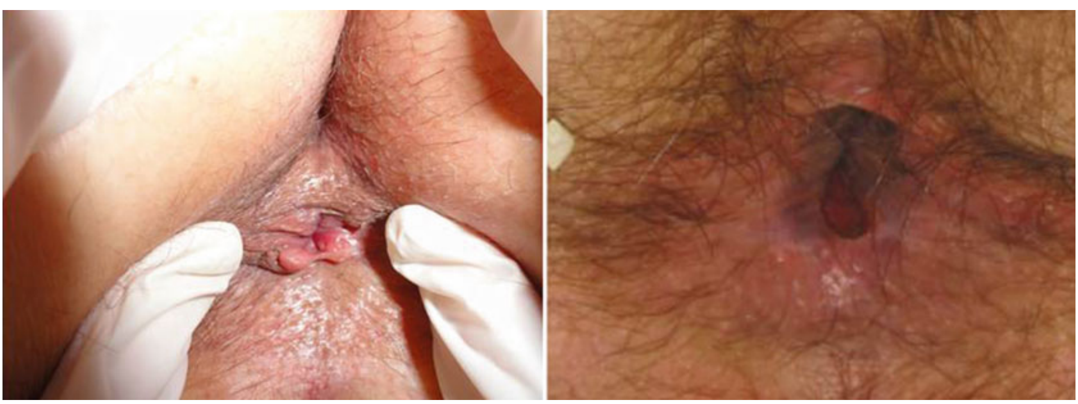

Fig. 11.9 At perineal inspection, after the buttocks are gently moved, an anal fissure can be observed

Fig. 11.10 A small abscess very close to the verge, associated to an intersphincteric anal fistula

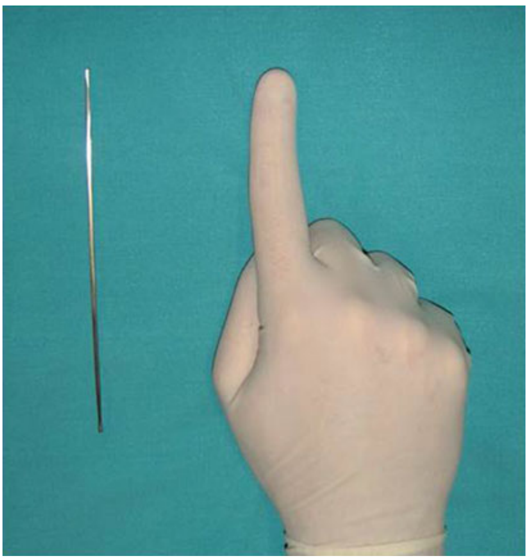

Fig. 11.11 A complete clinical examination, together with a careful probing, could be very useful in the diagnosis of anal fistulas. However, the last maneuver, if difficult or too painful for the patient, should be stopped to avoid the risk of a false fistula tract creation

authors concluded that a careful clinical examination could be sufficient to identify complex anal fistulas, thus avoiding expensive and unuseful radiological examinations.

However, several studies have stressed the role of the modern 3D endoanal ultrasound (EAUS) or magnetic resonance imaging (MRI) in the management of anal fistulas (Fig. 11.13). Toyonaga et al. have shown in a prospective study that a simple, painless, quick, cheap, and easy-to-perform EAUS was superior to physical examination in the identification of the internal anal opening, the primary tract, and the secondary horseshoe extension (Toyonaga et al. 2008). In our institution, since many years, 3D-EAUS is performed preoperatively in patients affected by anal fistula, with good results (Ratto et al. 2005) (Table 11.3); for this reason, given the importance to identify not only the internal anal opening but all features related to this condition (primary tract, secondary tracts, high intersphincteric extension, residual abscess cavity, etc.), addition of a 3D-EAUS to a careful clinical evaluation is strongly suggested whenever possible and available.

- *Defecatory Disorders*. The most common defecatory disorders evaluated during a proctology visit are rectocele, rectal intussusception and prolapse, enterocele, and anismus. The first clinical examination is crucial because the surgeon should establish the need (or not) of physiology and radiological testing. The accuracy of physical examination in diagnosing defecatory disorder is still questionable.

In a study by Siproudhis et al., the global agreement between clinical and radiological evaluation was 78 %, 85 %, and 56 % in the diagnosis of rectocele, intussusception, and perineal descent, respectively (Siproudhis et al. 1993). Another study focused on the evaluation of rectal intussusception stated that the clinical examination was accurate mainly for cases in which the internal prolapse was longer than 3 cm; the overall sensitivity rate was 87 %, but the specificity rate was only 51 %, so the authors suggested further evaluation by defecography in case of positive findings at the clinical examination (Karlbom et al. 2004). Nevertheless, these authors have given a simple but useful tip in the clinical evaluation of these patients, the test of “fast expulsion of rectoscope”: the expulsion of the rectoscope during the last defecation attempt should be faster in patients with rectal intussusception (Karlbom et al. 2004).

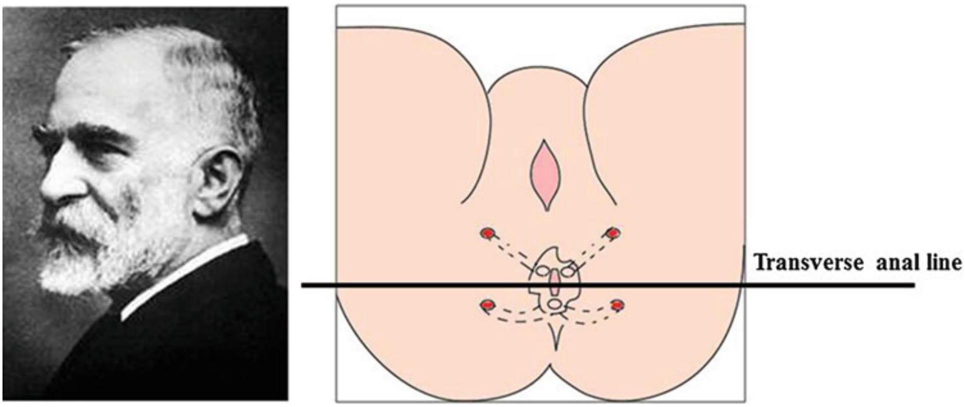

Fig. 11.12 The Goodsall's rule: an external anal opening posterior to the transverse anal line should be related to a curvilinear fistula tract, which it will open in the midline into the posterior anal canal; an anterior opening should be related to a radial and linear fistula tract

Another condition frequently associated to obstructed defecation is the anismus, an anorectal dyssynergia whose treatment is simply a pelvic floor rehabilitation. Its identification is of crucial importance to avoid performing unnecessary and inappropriate therapy. A recent study by Rao et al. stated that a simply digital anorectal examination was able to identify dyssynergia in patients with chronic constipation, with a sensitivity and specificity of 75 % and 87 %, respectively. A digital anorectal examination could, therefore, facilitate the selection of patients which should be submitted to anorectal manometry (Tantiphlachiva et al. 2010).

- *Fecal Incontinence*. An accurate clinical examination of patients with fecal incontinence should not be limited to the evaluation of resting and squeeze pressures by digital anorectal examination, because perineal inspection could reveal many useful data.
- A simple test that should always be performed in these patients is the so-called anal wink test: rubbing the perineal skin with a cotton pad, it stimulates the reflex contraction of the external anal sphincter, eliciting a reflex that evaluates the integrity of the pudendal pathway. If absent, it can be hypothesized a neurological etiology and an electromyography should be prescribed to obtain a better evaluation (Talley 2008).

Some useful findings at anal inspection emerged in a prospective study on 312 patients evaluated by digital anorectal examination and anorectal physiology tests. The presence of a perineal scar was associated to a low squeeze pressure, while a patulous anus to a low resting pressure. However, the accuracy of digital anorectal examination in detecting a sphincter lesion was low, particularly when the sphincter lesion was smaller than 90° (Dobbene et al. 2007).

Similar quite disappointing results in the identification of a sphincter lesion by digital anorectal examination has been recently reported, with a sensitivity of 67 % and a specificity of 55 % (Roos et al. 2012). At the perineal inspection were also evaluated the length of the perineal body, measuring the distance from the posterior fourchette to the anterior midline of the anal verge, and the pelvic floor muscle strength, using a validated score. Although very useful, these parameters were not able to reveal statistically significant differences between patients with and without sphincter injury. For this reason, at least an endoanal ultrasound should be included in the diagnostic workup of a patient with fecal incontinence.

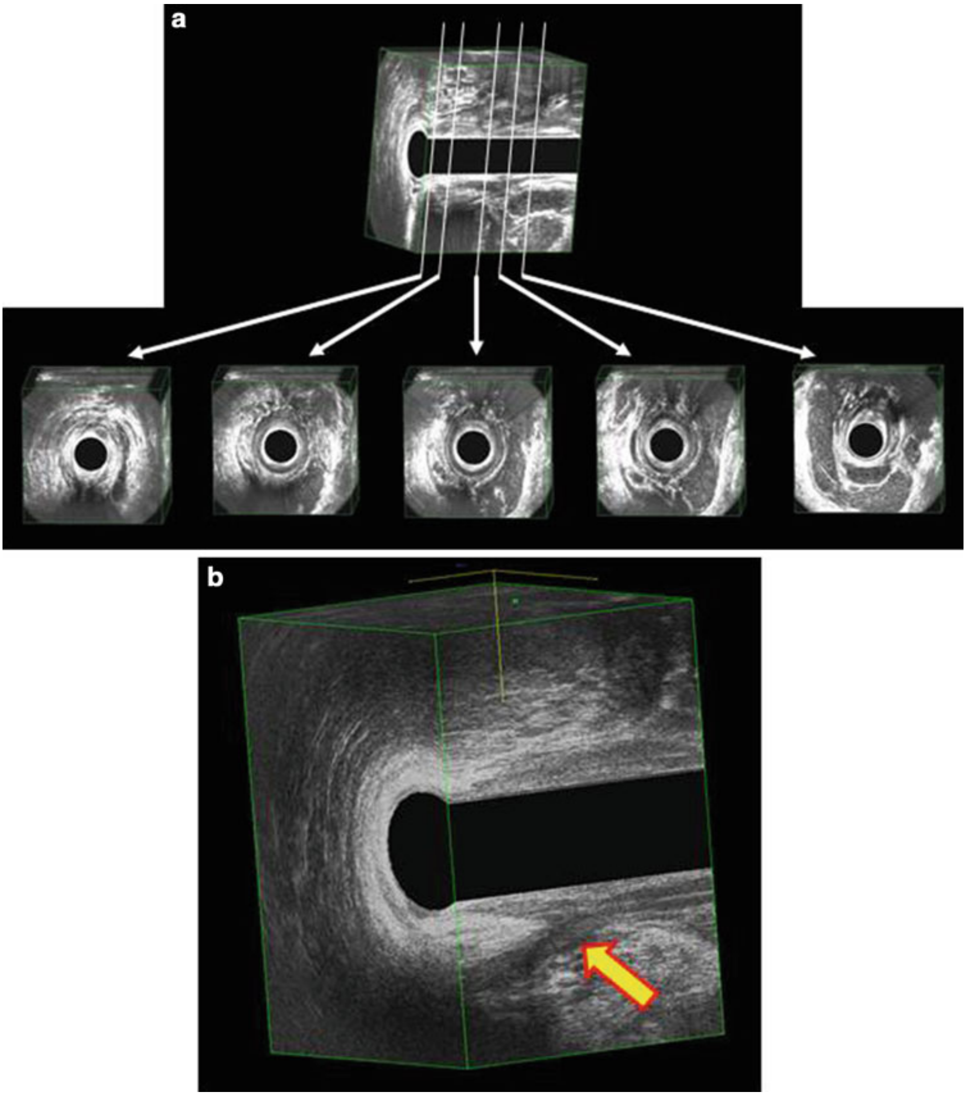

Fig. 11.13 3D endoanal ultrasound: anal abscess with horseshoe extension (**a**), posterior transsphincteric anal fistula (**b**)

Table 11.3 Correspondence between preoperative ultrasound and intraoperative findings (From Ratto et al. 2005)

3D-EAUS + H_2O_2 injection	Results (%)
Primary tract	94.1
Secondary tract	96.1
Internal anal opening	91.2
Abscess	100
Horseshoe extension	96.1

5 Physicians and the Clinical Assessment of Coloproctology Patient

Previous sections have stated that a proper clinical evaluation may be difficult even if performed by experienced colorectal surgeons. Given the high prevalence of colorectal diseases in the general population, a complete physical examination should be performed by all physicians correctly, but often it is not so.

A study conducted in the USA has shown that the accuracy in the diagnosis of anorectal disorders greatly varies among specialties and, moreover, was not related to years of experience (Grucela et al. 2010). Very similar results emerged also in a European country (Jimeno et al. 2012).

Usually, coloproctology patients are first evaluated by their general practitioner: however, studies have shown that the concordance rate between the diagnosis made by the general practitioner and the colorectal surgeon was only about 50 % (Springall and Todd 1988; Goldstein 1996).

These results have shown that there are few colorectal surgeons "board certified" who teach in universities, so few students (future doctors) can benefit from this experience. As a consequence, most of the future clinicians will not be able to perform an accurate clinical examination of a coloproctological disease. The presence of a colon and rectal surgeon, and his lessons within the medical students and general surgery residency training programs, is however able to provide clear benefits to students, patients, and universities (Hyman 1999; Longo 2003; Isherwood et al. 2013).

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Signs and Symptoms in Coloproctology: Data Collection and Scores

12

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Abstract

Many patients with colorectal disorders experience complex symptoms that are difficult to quantify, making measurement of these symptoms challenging. The subjective perception of the patient must be the foundation of any symptom evaluation and requires consideration of two different yet related components: symptom severity and measurement of impact.

1 Background

1.1 Symptom Severity Scores

Symptom severity scores are useful when evaluating subjective, complex, and multifactorial symptoms. Furthermore, such tools are increasingly relevant when there are varying levels of severity of symptom experience. In turn, these tools are equally helpful to assess treatment outcomes when the goals of the interventions involve varying degrees of improvement as opposed to an “all or none” response.

Symptom scores may be simple or intricate. Such scoring tools may be merely descriptive, a severity grading scale, or a severity summary score. In addition to a summary score, some colorectal scoring tools include a threshold score, below or above which the presence of a condition is defined. Some colorectal symptom severity scores include a component of measured impact, while other quality of life measures were created

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to solely measure and report the impact of symptoms for a given patient.

1.2 Quality of Life Measures

Impact measures such as quality of life (QoL) scores do not serve the same purpose as severity scores, but rather such tools complement severity scores by assessing what is not directly observable. Impact measures evaluate components which, when taken together, allow clinicians and researchers to make inferences about the quality of life of an individual. Impact measures may be generic scales, specialized scales, or condition-specific scales.

Generic scales assess a wide range of aspects of health-related quality of life. Such measurement tools are applicable to a broad population, yet may be used for detecting changes within a specific population. Due to the wide applicability and broadness of generic QoL scores, these tools may be used to make comparisons across populations. For example, the generic SF-36 QoL score is a well-studied and established score, hence its common use as a comparative when determining construct validity of a new measurement instrument. Generic QoL scales commonly do not have an overall summary score, but rather have a score for each component within the scale that is assessed.

Specialized quality of life scales are developed around a specific condition rather than a specific population, for example, the Gastrointestinal Quality of Life (GIQoL) scale. As these tools are more detailed and specific, they are more responsive to change and can detect more discrete variations. Specialized scales allow for comparison between two populations with different conditions.

Condition-specific scales reflect the dynamic relationship among a condition, its treatment, and quality of life. These scales address specific elements of a condition that affect the lives of persons with this given condition. Thus, condition-specific scales are most sensitive to the effects of a specific health condition.

2 Psychometric Assessment of Symptom Scoring Tools

Symptom scores and quality of life scales endeavor to assess a subjective phenomenon in a reliable and valid manner. Given the lack of objective measures for many complex colorectal symptoms, there remains no adequate criterion standard for comparison (Baxter et al. 2003). Thus, evaluation of the available scoring measures relies on principles established for the assessment of measured clinical and psychological phenomena (Baxter et al. 2003). Effective severity and impact measures should be reliable, valid, and responsive.

Reliability assesses the degree to which a measure reflects the underlying phenomenon rather than measurement error. Thus, a reliable measurement tool should yield reproducible results (reported using a Cronbach's alpha statistic greater than 0.70). Reliability is assessed in two ways: test-retest and internal consistency. Test-retest ascertains the consistency of test results when administered to the same patient in the same condition more than once and is reported using the intra-class correlation coefficient (Streiner and Norman 1994). Internal consistency assesses the extent to which all items within an outcome measure address the same underlying concept or phenomenon. Certainly, internal consistency is reserved for a measurement tool geared to assess one concept (Keszei et al. 2010).

Validity describes the degree to which an instrument measures what it is intended to measure. Validity has four key components (Nunally and Berbstein 1994; Cook and Beckman 2006). Face validity is a subjective assessment of the tool that reflects the extent to which a test appears to measure what it is intended to measure. Content validity is an assessment of the tool that describes the degree to which a test includes all items necessary to represent the concept being measured. Content validity can be determined by item generation based on literature review, expert opinion, and patient input. Criterion validity is an objective measurement of how a tool compares to a gold standard tool, if available. For example, how a fecal incontinence severity symptom score compares to anal manometry testing. Concurrent criterion validity is measured when two tests are simultaneously administered, while predictive criterion validity addresses how the new tool compares to an old tool. When there is no gold standard available, one may defer to construct validity. Construct validity addresses the ability of a test to measure the underlying concept of interest. For example, application of a tool in two different groups should yield the expected different outcomes. Convergent construct validity is demonstrated by correlation to a validated tool for the same outcome measure. Conversely, divergent construct validity refers to a negative correlation with an instrument that measures a different phenomenon.

Responsiveness is the degree to which a tool can accurately detect change when a change has occurred. As such, a highly responsive tool can detect small changes that occur, while an obtuse measure can only detect large changes in the outcome of interest. Internal responsiveness is the ability of the tool to measure change over time, for example, pre- and postoperative assessments. External responsiveness reflects the extent to which changes in a measure relate to changes in other different measures of health status. For example, correlation between changes in Wexner/Cleveland Clinic Florida (Wexner/CCF) fecal incontinence scores and Fecal Incontinence Quality of Life (FIQoL) scores demonstrates external responsiveness.

In this chapter, we will review symptom severity scoring and quality of life (QoL) scores for fecal incontinence and constipation.

3 Measuring Fecal Incontinence

Fecal incontinence is a common, socially embarrassing, and, at times, severely debilitating problem. Due to the complexity of this problem and difficulty explaining and quantifying the symptoms, many patients do not seek care. Major improvements in the diagnosis and management of fecal incontinence have occurred over the past several decades, in large part due to the significant progress that has been made in measuring and reporting incontinence (Baxter et al. 2003). Nonetheless, fecal incontinence is a challenging symptom to measure as it is a complex symptom with quite variable perception and impact on each individual patient. Thus, any measure of fecal incontinence must have the patient at its center. Many scales to measure fecal incontinence have been developed over the last several decades, and these can be categorized as follows: (1) grading scales, (2) summary scores, and (3) QoL scores.

3.1 Grading Scales for Fecal Incontinence

Grading scales for fecal incontinence are simple tools that assign a value to each specific type of incontinence in an ordinal fashion (Baxter et al. 2003). Scoring of fecal incontinence began with such simple grading scales as that described by Sir Alan G. Parks in his 1975 Royal Society of Medicine Section of Coloproctology presidential address. He created a simple grading scale to assess pre- and postoperative outcomes following postanal repairs for "neuropathic" fecal incontinence (Browning and Parks 1983) (Table 12.1). In this four-category scale, grade D denotes the worst degree of fecal incontinence, that is, incontinence to solid stool, while a person with normal continence would receive a grade A. A decade later, Broden and colleagues described another grading scale (Table 12.2) that is also composed of broad categories based on the type of incontinence (Broden et al. 1988). Patients are categorized based on the presence or absence of only one

Table 12.1 Sir Alan Parks' fecal incontinence grading scale (Browning and Parks 1983)

A	B	C	D
Continent to solid and liquid stool and flatus	Continent to solid and liquid stool but not to flatus	Continent to solid but not to liquid stool or flatus	Incontinent to solid and liquid stool and flatus

characteristic – solid, liquid, or flatus incontinence – with no consideration of frequency or impact of incontinence. In essence, all the available fecal incontinence grading scales (Table 12.3) are quite similar and are composed of three to five broad grades, and the spectrum ranges from "excellent" or "perfect" continence to "fully incontinent" or "poor." This broad categorization makes the grading scales easy to use; however, it distils the patient's symptoms into an almost "all or none" phenomenon resulting in a weak discriminatory ability. Thus, these scales do not have much ability to differentiate between patients with slight differences in degree of incontinence or to detect minor but clinically important changes (Baxter et al. 2003). Furthermore, these grading scales are merely descriptive tools; they do not assess the subjective patient experience of fecal incontinence. They solely reflect the subjective physician perspective of incontinence, that is, that incontinence of solid stool is more severe than liquid stool which is more severe than flatus.

Table 12.2 Broden et al.'s fecal incontinence grading scale (Broden et al. 1988)

Degree of incontinence	Definition
None (Baxter et al. 2003) (1)	No episodes of incontinence
Medium (Streiner and Norman 1994) (2)	Episodes of incontinence to solid stool, incontinence to gas, incontinence to liquid stool only or soiling
Severe (Keszei et al. 2010) (3)	Incontinence to solid stool at all times

3.2 Summary Scores for Fecal Incontinence

Summary scores evolved to address some of the weaknesses of grading scales. Summary scores assign values for certain categories of incontinence reflecting the severity of symptoms and create a summary score by summing up these individual values. The number of items in the different summary scores ranges from three to seven (with the exception of the American Medical Systems score that has 39 items), while the range of overall attainable scores is 0 to 31 points (with the exception of the American Medical Systems score that has 120 points) (Table 12.4). Thus, the summary scores are far less obtuse than the grading scales and have a greater ability to discriminate finite differences between patients and small changes in symptoms with treatment. All summary scores are similar in that they evaluate solid stool, liquid stool, and gas incontinence. Furthermore, in all the summary scoring tools, the frequency of incontinence contributes to severity of the overall symptom score. However, the scores differ in the definition of varying frequencies. For example, the most severe frequency may be described as more than one accident per week, daily accidents, or two or more accidents per day (Table 12.4). On the other end of the spectrum, some scores have a category for no

Table 12.3 Fecal incontinence grading scales available in the literature

Author	Year	Grades	Spectrum
Parks (1975)	1975	4	Normal → no control of solid stool
Broden et al. (1988)	1988	3	None → incontinence to stool at all times
Keighley and Fielding (1983)	1983	3	Minor leakage (<1/month) → severe (wear pad on most days)
Hiltunen et al. (1986)	1986	3	Gross FI → continent
Rudd (1979)	1979	5	Perfect continence → totally unsatisfactory, necessitates colostomy
Corman (1985)	1985	4	Excellent → poor (FI to solid stool, requires colostomy)
Williams et al. (1991)	1991	5	Continent → frequent incontinence to solid and liquid stool
Rainey et al. (1990)	1990	3	Continent of solid +/− flatus → incontinent of all stool
Womack et al. (1988)	1988	4	Continent → incontinent to solid and liquid stool and flatus

Table 12.4 Fecal incontinence summary scores available in the literature

Score	Author	Year	Range	Item	Spectrum	Validity tested	Reliability tested
Incontinence score system	Miller et al. (1988)	1988	1–9	3	<1/month → >1/week	+	–
Anal incontinence score	Rothenberger (1989)	1989	0–30	4	<1/month → >1/week	+	–
Pescatori grading and scoring of FI	Pescatori et al. (1992)	1992	0–9	6	<1/week → daily	+	~
Wexner/Cleveland Clinic Florida	Jorge and Wexner (1993)	1993	0–20	5	<1/month → >2/day	+	+
Continence scoring system	Lunnis et al. (1994)	1994	0–13	6	<1/month → most days	+	–
Vaizey/St. Mark's score	Vaizey et al. (1999)	1999	0–24	5	1/month → 1/day	+	+
Fecal Incontinence Severity Index (FISI)	Rockwood et al. (1999)	1999	0–61	6[a]	1–3/month → >2/day	+	+
Mayo FI questionnaire	Reilly et al. (2000)	2000	0–30	6	n/a	+	–
FI questionnaire intended for phone/mail	Malouf et al. (2000)	2000	None	3	<1/month → daily	+	–
American Medical Systems scale	*AMS* ® (O'Brien et al. 2004)	2000	0–120	39	Never → >1/day	+	+
Outcome tool for surgical management of FI	Hull et al. (2001)	2001	0–31	4	<1/month → >1/week	+	–
Clinical bowel function scoring system	Bai et al. (2002)	2002	0–12	6	Variable	+	–
Anal sphincter replacement scoring system	Violi et al. (2002)	2002	0–20	7[a]	Never → always	+	–

[a]Weighted

incontinence, while others define the least severe state of incontinence as less than one accident per month.

Three of the most widely used and cited summary scores for fecal incontinence (in decreasing order of peer-reviewed publication citations) are the Wexner/Cleveland Clinic Florida fecal incontinence score (Wexner/CCF FIS), the Vaizey/St. Mark's fecal incontinence score (Vaizey/St. Mark's FIS), and the Fecal Incontinence Severity Index (FISI). The Wexner/CCF FIS, published in 1993, is composed of 5 items, each scored on a scale of 0 to 4, allowing for a maximum of 20 points for the most severe incontinence (Table 12.5) (Jorge and Wexner 1993). Each of the types of incontinence presentations (solid, liquid, or gas incontinence) is graded equally in this summary score. The Wexner/CCF FIS is by far the most widely cited fecal incontinence score in the peer-reviewed literature, owing to its simplicity of use, availability of a clinically important threshold, exclusive focus on symptoms related to anal sphincter impairment, and its demonstrated validity and reliability. Reflecting its versatility and widespread use, this summary score has been translated and validated to numerous languages.

The Vaizey/St. Mark's FIS, published in 1999, is also commonly used in clinical studies and was based on the Wexner/CCF FIS with two additional items for assessment: the use of constipating medication and the presence of fecal urgency (Table 12.6) (Vaizey et al. 1999). The authors chose to use relative weighting for the items in this score, such that pad use and constipating

Table 12.5 Wexner/Cleveland Clinic Florida fecal incontinence score (Jorge and Wexner 1993)

	Never	Rarely <1/ month	Sometimes <1/week, ≥ 1/month	Usually <1/day, ≥ 1/week	Always ≥1/ day
Solid	0	1	2	3	4
Liquid	0	1	2	3	4
Gas	0	1	2	3	4
Wears a pad	0	1	2	3	4
Lifestyle alteration	0	1	2	3	4

Table 12.6 The Vaizey/St. Mark's fecal incontinence score (Vaizey et al. 1999)

	Never	Rarely 1/4 weeks	Sometimes >1/4 weeks	Weekly ≥1/ week	Daily ≥1/ day
Solid stool incontinence	0	1	2	3	4
Liquid stool incontinence	0	1	2	3	4
Gas incontinence	0	1	2	3	4
Alteration in lifestyle	0	1	2	3	4
	No	**Yes**			
Need to wear a pad or plug	0	2			
Constipating medication	0	2			
Lack of ability to defer defecation for 15 min	0	4			

medications were weighted with 2 points and the remaining items received 4 points, for a total score of 24.

The Fecal Incontinence Severity Index (FISI) is another validated score that is composed of a 20-cell matrix table that addresses type (gas, mucus, liquid, and solid) and frequency (5 categories) of incontinence episodes, which, like the other aforementioned scores, generates a summary score (Rockwood et al. 1999). Developed in 2000 by an American Society of Colon and Rectal Surgeons task force, the FISI is a weighted summary score, created by input from colorectal surgeons and patients. The incontinence categories assessed were determined by the authors. They developed two identical matrices (Table 12.7): one for actual scoring of the individual patient's symptoms and one for weighting. The weighting matrices were completed by 26 colorectal surgeons and 34 patients with fecal incontinence. This resulted in a summary score that ranges from 0 to 61, reflecting frequency of symptoms that range from none to two or more times per day. Though surgeon and patient rankings correlated well, surgeons gave significantly higher weights to incontinence of solid stool, reflecting a physiological interpretation of the event. Conversely, patients rated liquid and solid stool the same and rated gas incontinence significantly higher than surgeons. The authors did not advocate preferential use of the surgeon or the patient weights. Construct validity of the FISI was demonstrated in another study, where patients with worse symptoms had significantly higher FISI scores. Furthermore, severity rankings correlated with 3 out 4 items in the FIQoL scale (Rockwood et al. 1999).

Table 12.7 Fecal Incontinence Severity Index (Rockwood et al. 1999)

	2 or more times/ day	1 time/ day	2 or more times/ week	1 time/ week	1–3 times/ month
Gas					
Mucous					
Liquid					
Solid					

The American Medical Systems ® (AMS) score is another summary score described in the literature but not commonly used (Table 12.4) (American Medical Systems 1996). It is comprised of 39 questions, with a total score ranging from 0 to 120. The AMS score requires a retrospective evaluation of the last 4 weeks and has six levels of severity ranging from never to several times per day. Though this degree of detail and large range of possible values should yield strong distinguishing capacity, this does not seem to be the case. Created to assess the outcomes of the artificial bowel sphincter, the complexity of this score has limited its use, mostly to research purposes.

Vaizey et al. compared the Vaizey/St. Mark's, Pescatori, Wexner/CCF, and the AMS scores. The authors found the highest correlation between the Vaizey/St. Mark's and Wexner/CCF score and the lowest correlation with AMS score. They also found that detection of change was greatest for the Vaizey/St. Mark's and Wexner/CCF scores (Vaizey et al. 1999). Furthermore, Hussain et al. recently assessed intra- and interobserver reliability of the Vaizey/St. Mark's and Wexner/CCF scores (Hussain et al. 2014). To assess intraobserver reliability, each patient was asked to complete both assessments initially at recruitment and then 6 weeks later. No alteration to medications or treatment occurred during this interval. For interobserver reliability, both scores were also completed by a physician and a nurse with the patient. In a group of 39 patients, both scores demonstrated excellent intra- and interobserver reliability.

3.2.1 The Weight Debate

Though all summary scores include incontinence to solid stool, liquid stool, and flatus, some summary scores use equal weights, while others do not. The Wexner/CCF score weighs each item on a scale of 0 (never) to 4 (more than once per day). Thus, in this summary score, there is no judgment by the authors as to which type of incontinence is worse. Other authors chose a different approach and assigned different weights to the same frequencies of different items within the scoring tool. The authors of the Vaizey/St. Mark's score chose lesser weight for pad use and constipating medications that reflected their belief that these items may represent a subjective fear of social embarrassment, rather than actual frequency. However, again, such a method of assigning weights may not reflect the subjective patient experience of incontinence (Baxter et al. 2003). As seen in the FISI, patients and surgeons do not value all types of incontinence in the same way; thus, using surgeon-developed weights may not reflect the patient's experience which is paramount in describing a symptom. Thus, though conceptually intuitive, weighted summary scores add a degree of increased subjectivity. Firstly, who is the most appropriate person to assign the weights: patients or surgeons? If patients, should it be a standardized weighting, or should each individual patient set the relevant weights for themselves, that is, a self-derived weighting scheme? Although weights give additional information, they also add complexity and perhaps subjectivity to the tool, making it less practical for everyday clinical use. The most frequently cited scores are the simple non-weighted scoring systems as they are easy and practical to use.

3.2.2 Inclusion of a Measure of Impact Within the Summary Score

Another approach to including the patient's experience of the symptom, without weighting, is the inclusion of a measure of impact on the patient's life, by including either coping mechanisms, lifestyle alteration, or some other measure of quality of life. The Wexner/CCF score includes two items which address impact: changes in lifestyle and wearing a pad. These items serve as measures of the patient's experience of the symptom and their resultant quality of life. Similarly, the Vaizey/St. Mark's score uses the same items, in addition to constipating medications. These measures of impact have proven to be quite important as patients may limit the severity of their incontinence by altering their lifestyle; that is, a patient might have only infrequent episodes of incontinence by severely restricting their activities so that they can be close to a bathroom at all times. Such a patient's suffering would be appropriately detected by a tool that addresses impact but can be totally missed by a score that solely evaluates severity of incontinence. Thus, measuring impact in addition to severity can enrich our understanding of the patient's symptom.

3.2.3 Inclusion of Items Not Related to Anal Sphincter Function

Some scores include other aspects of incontinence that are not solely related to sphincter function. For example, the Vaizey/St. Mark's FIS (Table 12.6) includes urgency, the Lunniss score includes "difficulty cleaning" (Lunniss et al. 1994), and the Fecal Incontinence Severity Index (Table 12.7) includes mucous discharge. Such inclusions may not be most desirable as these symptoms are not exclusive of compromised anal sphincter function. For example, patients with severe proctitis could have mucous discharge and urgency though their underlying problem is proctitis rather than anal sphincter dysfunction.

3.2.4 Thresholds

Thresholds can be assigned to determine the level at which treatment can be useful or to predict a change in QoL with a change in symptom severity. In addition, thresholds are useful to determine the desired treatment effect and guide sample size calculations in research protocols. Such a useful threshold was determined for the Wexner/CCF score. In 2001, Rothbarth et al. evaluated 35 women with anterior sphincter defects who underwent sphincter repair. The patients completed the Wexner/CCF FIS, Gastrointestinal Quality of Life (GIQoL) index score, and the SF-36 score before and after sphincter repair. The authors found a strong correlation between a Wexner/CCF FIS greater than or equal to 10 and a lower GIQoL index and SF-36 score compared with the standard population (Rothbarth et al. 2001). Similarly, Damon and colleagues looked at clinical characteristics and quality of life in a cohort of 621 patients with fecal incontinence using the Wexner/CCF FIS and GIQoL index. They found a significant correlation between a Wexner/CCF FIS greater than or equal to 11 and the total GIQoL score. More recently, Brown et al. conducted the largest community-based fecal incontinence questionnaire to date, including 5,817 women 45 years and older (Brown et al. 2013). One of the questions asked was "Have you ever talked to a physician about accidental leakage of stool and/or gas?." Data was available for 938 women who responded "yes," and only 29 % of these responders with fecal incontinence reported that they sought care. Care seekers were more likely to have a primary care physician, have heard about fecal incontinence, and suffered longer with more severe leakage. Furthermore, the mean Wexner score of care seekers was 10.7 compared to non-care seekers who had a score of 7.5. Thus, a threshold of 10 or greater on the Wexner score also predicts women who seek care and predicts a significantly worse QoL.

In summary, the best and most used summary scores are the ones that balance between ease of use and a strong discriminatory ability. The Wexner/CCF and Vaizey/St. Mark's scores both achieve this balance, as is demonstrated by their widespread use, validity, reliability, and correlation with QoL measures. Furthermore, thresholds within the summary scores allow clinicians to assess the impact of interventions and researchers to plan protocols for future treatments. Judging by the "weight" of the number of citations in peer reviewed publications, the Wexner/CCF has been more widely accepted perhaps due to its simplicity and lack of potentially subjective weightings.

4 Assessment of the Impact of Fecal Incontinence on Quality of Life

Quality of life (QoL) instruments are designed to measure the subjective perception of a given patient's health state on their emotional and social life. Both generic and disease-specific QoL instruments have been used for fecal incontinence (Table 12.8). The most widely cited and well-validated QoL measure specifically designed to assess the impact of treatment for fecal incontinence is the Fecal Incontinence Quality of Life (FIQoL) scale. This scale was developed by the American Society of Colon and Rectal Surgeons in 2000 (Rockwood et al. 2000) and is composed

Table 12.8 Fecal Incontinence Quality of Life measures

Score	Author	Year	Range	Domains	Summary score	Validity tested	Reliability tested
FIQoL scale	Rockwood et al. (2000)	2000	0–29	4	Yes	+	+
Quality of life scale for FI in pediatrics	Bai et al. (2002)	2000	0–12	6	Yes	+	–
Manchester Health Questionnaire	Bugg et al. (2001)	2001	0–100	10	Yes	+	+
Pelvic Floor Impact Questionnaire and Disease Inventory (PFIQ-7, PFDI-20)	Barber et al. (2001)	2001	0–400	3	Yes	+	–
TyPE Specification	Wexner et al. (2002)	2002	–	14	No	+	–
Modified Manchester Health Questionnaire for phone interview	Kwon et al. (2006)	2003	–	8	No	+	–
Simple QoL FI questionnaire	Kyrsa et al. (2009)	2009	–	5	No	+	–

of four domains: lifestyle, coping/behavior, depression/self-perception, and embarrassment, and contains a total of 29 items (Table 12.9).

The FIQoL scale was determined to be reliable by test-retest through telephone interview assessments and was also determined to have strong internal consistency (Rockwood et al. 2000). The scale's face and content validity were demonstrated through a pilot patient sample and an expert panel, respectively (Rockwood et al. 2000). Furthermore, incontinent patients had significantly worse FIQoL scores than continent patients, FIQoL scores correlated with a generic QoL scale (SF-36) and Wexner/CCF scores, demonstrating construct validity (Rockwood et al. 2000). This disease-specific QoL scale is a well-accepted research end point and has been translated and validated in several languages. In 2004, Rullier and colleagues tested the validity and reliability of a French translation of the FIQoL scale in a multicenter study including 100 patients with fecal incontinence (Rullier et al. 2004). They observed a good correlation between the lifestyle, depression, and coping/behavior scales. They also found that the FIQoL had good internal reliability for each scale (Cronbach's alpha 0.78–0.92). Finally, they found that the FIQoL scale and French translation of the Wexner/CCF FIS had good correlation. Similarly, the Spanish translation of the FIQoL was assessed in a multicenter study of 118 patients with fecal incontinence and was found to have good to excellent internal reliability for all domains, and all domains significantly correlated with a generic questionnaire on health and scale of severity of fecal incontinence (Minguez et al. 2006).

5 Measuring Constipation

Constipation is another functional problem that can be difficult to describe. The symptoms and impact of these symptoms on a person's quality of life are often quite challenging to quantify. Gradual acceptance of lifestyle alterations due to the chronicity of the symptoms makes quantitative description of lifestyle compromise challenging. The complex constellation of symptoms with constipation includes frequency of bowel movements, methods of evacuation, symptoms associated with evacuation itself, abdominal pain, and others.

Many scoring scales for constipation set threshold values for significant constipation. This method is required due to the complex nature of constipation and the variable reporting of the functional impact of certain aspects of

Table 12.9 Items in the FIQoL scale – adapted from Rockwood et al. (Rockwood et al. 2000)

Scale 1: lifestyle
Que3B: I cannot do many of the things I want to do (agreement, 4 points)
Que2A: I am afraid to go out (frequency, 4 points)
Que2G: It is important to plan my schedule (daily activities) around my bowel pattern (frequency, 4 points)
Que2E: I cut down on how much I eat before I go out (frequency, 4 points)
Que2D: It is difficult for me to get out and do things like going to a movie or church (frequency, 4 points)
Que3L: I avoid traveling by plane or train (agreement, 4 points)
Que2H: I avoid traveling (frequency, 4 points)
Que2B: I avoid visiting friends (frequency, 4 points)
Que3M: I avoid going out to eat (agreement, 4 points)
Que2C: I avoid staying overnight away from home (frequency, 4 points)
Scoring = (Que3B + Que2A + Que2G + Que2E + Que2D + Que3L + Que2H + Que2B + Que3M + Que2C)/10
Scale 2: coping/behavior
Que3H: I have sex less often than I would like to (agreement, 4 points)
Que3J: The possibility of bowel accidents is always on my mind (agreement, 4 points)
Que2J: I feel like I have no control over my bowels (frequency, 4 points)
Que3N: Whenever I go someplace new, I specifically locate where the bathrooms are (agreement, 4 points)
Que2I: I worry about not being able to get to the toilet in time (frequency, 4 points)
Que3C: I worry about bowel accidents (agreement, 4 points)
Que2M: I try to prevent bowel accidents by staying very near a bathroom (agreement, 4 points)
Que2K: I can't hold my bowel movement long enough to get to the bathroom (frequency, 4 points)
Que2F: Whenever I am away from home, I try to stay near a restroom as much as possible (frequency, 4 points)
Scoring = (Que3H + Que3J + Que2J + Que3N + Que2I + Que3C + Que2M + Que2K + Que2F)/9
Scale 3: depression
Que1: In general, would you say your health is (excellent/poor, 5 points)
Que3K: I am afraid to have sex (agreement, 4 points)
Que3I: I feel different from other people (agreement, 4 points)
Que3G: I enjoy life less (agreement, 4 points)
Que3F: I feel like I am not a healthy person (agreement, 4 points)
Que3D: I feel depressed (agreement, 4 points)
Que4: During the past month, have you felt so sad, discouraged, hopeless, or had so many problems that you wondered if anything was worthwhile? (extremely so/not at all, 6 points)
Scoring = [(Que1 × 4/5) + Que3K + Que3I + Que3G + Que3F + Que3D + (Que4 × 4/6)]/7
Scale 4: embarrassment
Que2L: I leak stool without even knowing it (frequency, 4 points)
Que3E: I worry about others smelling stool on me (agreement, 4 points)
Que3A: I feel ashamed (agreement, 4 points)
Scoring = (Que2L + Que3E + Que3A)/3
Que = question

constipation among patients. For example, some patients may experience significant abdominal pain, while others may control symptoms with digitation or laxative use. The overall impact on quality of life and function may be the same, though the symptoms are different. Because of the difficulty establishing threshold cutoffs, these values may be somewhat arbitrary. However, the vast majority of the most commonly cited constipation scoring systems have been validated (see Table 12.10). Thus, these threshold cutoffs, though they may have been established arbitrarily based on clinical observation, have been confirmed and validated based on statistical

Table 12.10 Constipation scores available in the literature

Score	Author	Validity tested	Reliability tested
Constipation Assessment Scale	McMillan et al. (McMillan and Williams 1989)	+	+
Revised Constipation Assessment Scale	Broussard et al. (Broussard 1998)	+	+
Wexner/CCF Constipation Score (CCS)	Agachan et al. (1996)	+	+
Patient Assessment of Constipation	Frank et al. (1999)	+	+
Questionnaire for constipation and FI	Osterberg et al. (1996)	+	+
Knowles Eccersley Scott Symptom (KESS) score	Knowles et al. (2000)	+	–
Visual scale analog questionnaire (VSAQ)	Pamuq et al. (2003)	+	–
Garrigues questionnaire	Garrigues et al. (2004)	~	–

Table 12.11 Characteristics of scoring systems for constipation

Score	Author	Year	Items	Score range	Threshold score
Constipation Assessment Scale	McMillan et al. (McMillan and Williams 1989)	1989	8	0–16	No
Revised Constipation Assessment Scale	Broussard et al. (Broussard 1998)	1998	8	0–32	No
CCF Constipation Score (CCS)	Agachan et al. (1996)	1996	8	0–30	Yes
Patient Assessment of Constipation	Frank et al. (1999)	1999	12	0–48	No
Questionnaire for constipation and fecal incontinence	Osterberg et al. (1996)	1996	47	n/a	n/a
Knowles Eccersley Scott Symptom (KESS) score	Knowles et al. (2000)	2000	11	0–39	Yes
Visual scale analog questionnaire (VSAQ)	Pamuk et al. (2003)	2003	5	0–10	Yes
Garrigues questionnaire	Garrigues et al. (2004)	2004	21	n/a	n/a

probability. All of the major constipation scoring systems are self-reported by the patient and non-weighted. They are relatively simple to complete, with Table 12.11 showing the score ranges and presence of defined threshold scores for each scoring system.

5.1 Summary Scores for Constipation

The first summary score for scaling constipation was published by McMillan and Williams in 1989, and for a decade, no other major scoring systems were described (McMillan and Williams 1989). The Constipation Assessment Scale (CAS) sought to be a tool to measure the presence and severity of constipation in oncology patients, specifically those using opioids and chemotherapy (McMillan and Williams 1989). Their scale was found to be both valid and reliable in this group, as well as easy to complete in about 2 min. Table 12.12 shows the scoring tool and its components, with each of the components scored from 0 to 2, depending on the patient-reported severity of the symptoms. The authors suggested that the biggest benefits to the novel tool were readability of the instrument and the short amount of time required to complete it. It also appeared to have good reliability. A potential drawback to the tool is that it does not attempt to quantify frequency of bowel movements and leaves it entirely up to the patient to rate the severity of "less bowel movements." In the context of patients on active treatment for cancer, such a scale

Table 12.12 Constipation assessment scale (McMillan and Williams 1989)

	No problem (0)	Some problem (Baxter et al. 2003) (1)	Severe problem (Streiner and Norman 1994) (2)
Abdominal distension or bloating			
Change in amount of gas passed rectally			
Less frequent bowel movements			
Oozing liquid stool			
Rectal fullness or pressure			
Rectal pain with bowel movement			
Small volume of stool			
Unable to pass stool			

may very well be adequate to assess pre-, during, and posttreatment effects of chemotherapy or opioids on bowel habits. Interestingly, the CAS has been validated in other specific patient populations, including pregnant women and pediatric oncology patients (Woolery et al. 2006; Broussard 1998).

For nearly a decade following the publication of the CAS, there were no major publications on novel tools for assessing constipation. This hiatus was followed by a decade of numerous reports of scoring systems for constipation. By far, the most frequently cited scoring system for constipation is the Wexner/Cleveland Clinic Constipation Score (Agachan et al. 1996). Wexner and colleagues embarked on a large undertaking to identify factors that commonly contributed to patients' symptoms of constipation, develop a scoring system for constipation, and compare results with objective testing to assess for correlative factors. After surveying over 200 patients with constipation, eight factors were found to be commonly reported issues in constipation and were used to develop the scoring system shown in Table 12.13. Scores range from 0 to 30 and symptoms are patient reported. All patients with significant constipation were found to have scores of 15 or higher; thus, a score of 15 was set as the threshold score for significant constipation.

The Wexner/Cleveland Clinic Constipation Score has many important features. First, it attempts to quantify symptoms to make patient-reported outcomes more objective, which is a major advantage. It combines subjective symptoms as well as numeric responses of frequencies of specific events, such as frequency of movements. Second, important variables were chosen from among a larger pilot survey to find those that were significant and effective in distinguishing constipated from non-constipated patients. In addition, the Wexner/Cleveland Clinic Constipation Score was compared to physiology testing and found to correlate very well with objective signs of constipation. This is a unique feature of the study. The scores were compared to results of colonic transit time, anal manometry, defecography, and pudendal electromyography studies. All of the constipated patients with scores above 15 had some objective reason for their constipation, such as rectocele or paradoxical puborectalis function (Agachan et al. 1996). Thus, for patients with scores of 15 or greater, it is likely worth pursuing thorough investigations for a modifiable cause for constipation.

The Knowles Eccersley Scott Symptom (KESS) score was published as a modification of the Wexner/Cleveland Clinic Constipation Score, with additional outcomes added. Table 12.14 shows the KESS score, which again includes subjective and objective scoring of factors associated with constipation. In addition to the symptoms described by Wexner and colleagues, the KESS score added failure of evacuation less frequent than daily, duration of laxative use, bloating, and stool consistency (Knowles et al. 2000). The KESS score was found to correlate very well with the Wexner/Cleveland Clinic Constipation

Table 12.13 Wexner/Cleveland Clinic Florida constipation score (Agachan et al. 1996)

Frequency of bowel movements	**Score**
1×/1–2 days	0
2×/week	1
1×/week	2
Less than 1×/week	3
Less than 1×/month	4
Painful evacuation	
Never	0
Rarely	1
Sometimes	2
Usually	3
Always	4
Feeling incomplete	
Never	0
Rarely	1
Sometimes	2
Usually	3
Always	4
Abdominal pain	
Never	0
Rarely	1
Sometimes	2
Usually	3
Always	4
Time (min) in lavatory	**Score**
Less than 5	0
5–10	1
10–20	2
20–30	3
More than 30	4
Assistance	
Without	0
Stimulant laxatives	1
Digital or enema	2
Unsuccessful attempts per 24 h	
Never	0
1–3	1
3–6	2
6–9	3
More than 9	4
Duration of constipation (years)	
0	0
1–5	1
5–10	2
10–20	3
More than 20	4
Total score	**30**

Table 12.14 KESS score for constipation (Knowles et al. 2000)

Duration of constipation	**Score**
0–18 months	0
18 months to 5 years	1
5–10 years	2
10–20 years	3
Over 20 years	4
Laxative use	
None	0
PRN or short duration	1
Regular, long duration	2
Long duration, ineffective	3
Frequency of BM	
1–2x/1–2 days	0
2 or less x/week	1
Less than 1x/week	2
Less than 1x/2 weeks	3
Unsuccessful evacuation	
Never/rarely	0
Occasionally	1
Usually	2
Incomplete evacuation	
Never	0
Rarely	1
Occasionally	2
Usually	3
Always	4
Abdominal pain	
Never	0
Rarely	1
Occasionally	2
Usually	3
Always	4
Bloating	
Never	0
Perceived by patient only	1
Visible to others	2
Severe causing satiety	3
Severe causing vomiting	4
Enemas/digitation	
None	0
Occasionally	1
Regular	2
Manual evacuation occasionally	3
Manual evacuation always	4
Time taken (min)	
<5 min	0
5–10 min	1

(*continued*)

Table 12.14 (continued)

Duration of constipation	Score
10–30 min	2
>30 min	3
Painful evacuation	
Never	0
Rarely	1
Occasionally	2
Usually	3
Always	4
Stool consistency	
Soft/loose/normal	0
Occasionally hard	1
Always hard	2
Pellet like	3

Score, with a Pearson *r* correlation coefficient of 0.90. The authors attempted to predict the pathophysiology of a patient's constipation based on the KESS score. This was building on the work of Koch et al., who found that certain clusters of symptoms were unique, depending on the etiology of constipation (Koch et al. 1997). When looking at slow transit constipation, rectal evacuatory disorders, and mixed etiology constipation, the KESS scoring algorithm was able to predict the etiology of constipation in 55 % of patients (Knowles et al. 2000). While this represents the majority of patients in the study, clearly the tool is not ideal to predict etiology of constipation, and improvements should be made to the scoring system if it is to be applied to diagnosis prediction.

The Patient Assessment of Constipation (PAC) scoring system was developed around the same time. This scoring system grades patients' symptoms from 0 to 48, based on a 12-question survey (Frank et al. 1999 Sep). It is available online and has been validated to use to follow the course of a patient's treatment to monitor improvement (Frank et al. 1999). Symptoms are graded from 0 (absent) to 4 (very severe) for each of the 12 metrics (Table 12.15). This tool was developed with rigorous testing and retesting for validity. It was shown to be effective in predicting physician-assessed clinical severity and also showed improvement as patients progressed through a 6-week treatment regimen to improve constipation (Frank et al. 1999). It should be noted that the

Table 12.15 Patient assessment of constipation score (PROQOLID 2015)

Symptom	Score (0–4)
Discomfort in your abdomen	
Pain in your abdomen	
Bloating in your abdomen	
Stomach cramps	
Painful bowel movements	
Rectal burning during or after a bowel movement	
Rectal bleeding or tearing during or after a bowel movement	
Incomplete bowel movement like you didn't "finish"	
Bowel movements that were too hard	
Bowel movements that were too small	
Straining or squeezing to try to pass bowel movements	
Feeling like you had to pass a bowel movement but you couldn't	

mean age of the patients was 42 years and the vast majority (over 93 %) were female. As such, the results may not be applicable to all patients. The PAC scale described assesses symptoms only and does not address quality of life issues or frequency of bowel movements. Combined with other quality of life tools, this scale may be quite useful, as the questions are easy to understand and in very plain language.

A somewhat unique scoring tool was described by Pamuk and colleagues, who used a visual scale analog questionnaire (VSAQ) to assess for symptoms of constipation (Pamuk et al. 2003). Stool consistency was rated from 1 to 5 (from pellet to watery), whereas straining and incomplete evacuation were rated on a visual analog scale from 1 to 10. This tool was tested in healthy volunteers who completed the scoring tool and kept a stool diary for 7 days. Diaries were assessed by study personnel for clinical criteria of constipation. Of the patients who met 3 or more criteria for constipation based on their diaries, 92 % of them scored greater than 3 on the VSAQ (Pamuk et al. 2003). Therefore, a threshold score of 3 was chosen as the cutoff for constipation. The study found that the VSAQ was easy to complete and was sensitive to distinguish between constipated and non-constipated study participants. However, the VSAQ was better at detecting patients who were *not* constipated (based on their diary responses) than detecting those who *were* constipated. This highlights imperfections in such a simple scoring system and should not be used as the primary scoring tool for constipation.

5.2 Grading Questionnaires for Constipation

Garrigues and colleagues have described symptom-related scales and scoring systems for numerous diseases. They developed a questionnaire of 21 questions to assess for the prevalence of constipation in the Spanish population (Garrigues et al. 2004). As seen in Table 12.16, their questionnaire does not assess a score and was not validated to distinguish the severity of constipation. Using the Rome II definition of constipation, the authors found that self-reported constipation, anal blockage, straining, and hard stools were the patient-reported factors most accurate in predicting the presence of constipation (Garrigues et al. 2004). This tool may identify many patients with constipation, but was only shown to help predict the presence of constipation. That is, it is not validated to assess treatment effect, severity, or type of constipation. It is therefore not the ideal scale to use to guide treatment or communicate severity of constipation between healthcare providers, as many of the other scoring systems are able to do.

Another largely descriptive questionnaire that does not assign a score or distinguish thresholds is the questionnaire for constipation and fecal incontinence developed by Osterberg et al. (Osterberg et al. 1996). This tool is lengthier than the Garrigues questionnaire, mainly because it is meant to assess for both constipation and incontinence. The questionnaire was found to be valid and reliable. It is able to distinguish diagnoses and healthy controls. Table 12.17 shows all the items assessed (as patient-reported outcomes), but the factors found to be most reliable in the assessment of constipation were stool frequency, digitation to produce bowel movement, and straining (Osterberg et al. 1996). The benefit of this

Table 12.16 Garrigues et al.'s questionnaire for constipation (Garrigues et al. 2004)

1. Indicate your age 2. Indicate your gender 3. Indicate your educational level 4. Indicate your job 5. Indicate the amount of fiber in your diet: low/medium/high 6. How often do you perform physical exercise? Never/sometimes/ habitually 7. Indicate which drugs you are taking 8. Have you felt constipated? Yes/no	
9. Do you strain during a bowel movement? 10. Do you feel an incomplete emptying sensation after a bowel movement? 11. How often are your stools hard? 12. Do you feel a blockage in the anus that makes it difficult to pass the stool? 13. Do you need to press around the anus or vagina to complete a bowel movement? 14. Do you spend more than 10 min on the toilet to pass the stools?	Never, sometimes, often, always
15. How many bowel movements do you usually have each week?	
16. Do you take oral laxatives? 17. Do you need to use suppositories to have bowel movements? 18. Do you need to use enemas to have bowel movements?	Never, fewer than once a week, one or more times per week, every day
19. Have you visited a doctor because of constipation? Yes/no 20. Have you presented with abdominal pain more than six times this past year? Yes/no 21. Have you presented with loose or watery stools? Yes/no	

Table 12.17 Osterberg et al.'s questionnaire for assessment of constipation and fecal incontinence (Osterberg et al. 1996)

1. Stool frequency
2. Laxative use
3. Enema use
4. Stool consistency
5. Abdominal pain
6. Bloating
7. Excess flatus
8. Digitation
9. Time taken to pass stool
10. Need to strain
11. Percentage straining
12. Feeling of incomplete evacuation
13. Incontinence to gas
14. Incontinence to loose stool
15. Incontinence to solid stool
16. Use of pads
17. Pruritus ani
18. Sensibility for stool
19. Differentiate gas from solid stool
20. Urgency
21. Deferring time, loose stool
22. Deferring time, solid stool
23. Rectal pain
24. Painful defecation
25. Anal bleeding
26.Physical impact
27. Social impact
28. No. of stools per week
29. Urinary stress incontinence
30. Bladder emptying difficulties
31. Previous anorectal surgery
32. No. of childbirths
33. Forceps delivery
34. Vacuum extraction
35. Obstetric sphincter tear
36. Suture of perineal tear
37. History of pelvic prolapse
38. Surgery for pelvic prolapse

questionnaire is that it broadly assesses functional colorectal symptoms and disorders. Clearly, though, it is largely descriptive and for a focused and quicker assessment of constipation symptoms, some of the other scoring systems previously discussed would be more appropriate and provide more specific information about severity.

Both Garrigues et al. and Osterberg et al. provide useful population-wide screening tools to assess for constipation and highlight important symptoms that should be assessed in the diagnosis and management of constipation.

6 Measuring Other Colorectal Outcomes

6.1 Bowel Function After Proctectomy

Restorative, or sphincter preserving, rectal resections have become the standard of care wherever feasible. The avoidance of permanent colostomy is clearly preferred by most patients. As lower anastomotic levels become both accepted and technically feasible with new techniques, the question of bowel function after such procedures has come to light. Not all patients are good functional candidates for a restorative procedure as some patients report frequency, urgency, and impaired continence. Assessment of this cluster of symptoms, known as low anterior resection syndrome (LARS), has been of recent interest in the surgical literature. It is essential to be able to assess these symptoms in a standardized way and stratify severity based on symptom scoring.

In 2005, Temple et al. developed a bowel function instrument to assess bowel function after rectal cancer surgery (Temple et al. 2005). It was developed through focus groups and extensive piloting and was rigorously tested for validity and reliability. The result is an instrument meant to grade severity of symptoms, though no formal thresholds for mild versus severe impairment were set. Table 12.18 shows the instrument questions. The results of the instrument show that patients had a mean of 22 symptoms after rectal cancer surgery (Temple et al. 2005). The instrument was able to detect differences between type of surgery performed presence and timing of radiation therapy and correlated well with quality of life scores.

The LARS score was developed and tested on patients registered in the Danish National Colorectal Cancer Database (Emmertsen and Laurberg 2012). It is a summary score tool with set

Table 12.18 Bowel function instrument after rectal cancer surgery (Temple et al. 2005)

Over the last 4 weeks, how many bowel movements do you generally have in 24 h?	Number:				
	Always	Most of the time	Sometimes	Rarely	Never
Do certain solid foods increase the number of bowel movements in a day?					
Do certain liquids that you drink increase the number of bowel movements in a day?					
Do you feel like you have totally emptied your bowels after a bowel movement?					
Do you get to the toilet on time?					
Do you have another bowel movement within 15 min of your last bowel movement?					
Do you know the difference between having to pass gas (air) and needing to have a bowel movement?					
Have you used medicines to decrease the number of bowel movements (drugs like Imodium®, Lomotil®)?					
Have you had diarrhea (no form, watery stool)?					
Have you had loose stool (slight form, but mushy)?					
Have you been able to wait 15 min to get to the toilet when you feel like you are going to have a bowel movement?					
Have you been able to control the passage of gas (air)?					
Have you limited the types of solid foods you eat to control your bowel movements?					
Have you limited the types of liquids you drink to control you bowel movements?					
Have you had soilage (leakage of stool) of your undergarments during the day?					
Have you used a tissue, napkin, and/or pad in your undergarments during the day in case of stool leakage?					
Have you had soilage (leakage of stool) of your undergarments when you go to bed?					
Have you had to alter your activities because of your bowel function?					

thresholds for severity of LARS. The LARS score ranges from 0 to 42 and is divided into no LARS (0 to 20), minor LARS (Broussard 1998; Agachan et al. 1996; Knowles et al. 2000; Koch et al. 1997; Frank et al. 1999 Sep; Pamuk et al. 2003; Garrigues et al. 2004; Osterberg et al. 1996; Temple et al. 2005), (21–29) and major LARS (Emmertsen and Laurberg 2012; Juul et al. 2014; Wong et al. 2015; Ward et al. 1999; Sprangers et al. 1999; Sandborn et al. 1994; PROQOLID 2015; Parks 1975; Keighley and Fielding 1983; Hiltunen et al. 1986; Rudd 1979; Corman 1985; Wiliams et al. 1991) (30–42). It is an easy tool for patients to complete and captures most of the major impairments that can occur after anterior resection (Table 12.19). Not only does this tool stratify patients according to threshold symptom scores, but it was also found to be able to discriminate between height of anastomosis and presence of radiation therapy. The LARS score has been internationally validated in multiple European countries, showing similar abilities of the scoring system to discriminate between surgery and radiation types, as well as have a high test-retest probability (Juul et al. 2014). Scores have also been found to correlate well with quality of life scores (Juul et al. 2014). The LARS score will be a very useful tool going forward to test new treatment regimens with timing of radiation, expanded role of intersphincteric proctectomy for rectal

Table 12.19 Low anterior resection syndrome score (Emmertsen and Laurberg 2012)

	Score
Incontinence for flatus	
Never	0
<once a week	4
≥once a week	7
Incontinence for liquid stools	
Never	0
<once a week	3
≥once a week	3
Frequency of bowel movements	
>7 times a day	4
4–7 times a day	2
1–3 times a day	0
<once a day	5
Clustering of stools	
Never	0
<once a week	9
≥once a week	11
Urgency	
Never	0
<once a week	11
≥once a week	16
LARS score	0–42

cancer, and other therapies which may impair postoperative bowel function.

6.2 Quality of Life Scores for Colorectal Surgery

There have been many scores developed for both general and colorectal-specific assessment of quality of life. A recent systematic review of quality of life instruments for the assessment of colorectal cancer patients found that only a few of the scales were consistently rated the highest (Wong et al. 2015). The Functional Assessment of Cancer Therapy-Colorectal (FACT-C) instrument was the most commonly studied quality of life score. The European Organization for Research and Treatment of Cancer (EORTC) quality of life module developed specifically for the assessment of colorectal cancer (QLQ-CR38) was overall rated the highest and found to have the highest level of evidence for use. The bowel function instrument, developed by the group at Memorial Sloan Kettering Cancer Center and discussed earlier in this chapter, was also found to be a very useful instrument in this systematic review.

The FACT-C instrument was developed in the United States and first reported by Ward et al. (Ward et al. 1999). It combines elements of general quality of life of the related FACT-General instrument with colorectal-specific quality of life questions, such as assessment of diarrhea and bowel control (Table 12.20). The study found that the FACT-C instrument was valid and reliable in English and Spanish and has since been further tested in additional studies and languages. The addition of the colorectal cancer-specific questions relating to bowel function and ostomy care at the end was found to provide a more complete picture of quality of life, and this additional information is essential to identify possible modifiable elements that may improve quality of life in this patient population. The authors suggested the FACT-C would be most useful in further research on colorectal cancer treatments and specifically in patients who require an ostomy as part of their surgical care (Ward et al. 1999). Since publication in 1999, the FACT-C has indeed been applied to many aspects of colorectal cancer care.

The EORTC QLQ-CR38 was first published just 1 year after the FACT-C (Sprangers et al. 1999). The instrument was developed to assess patients at various phases of colorectal cancer treatments who were participating in clinical trials, in order to have a consistently reported instrument. It consists of 38 questions, of which 19 questions are completed only by certain patient groups, for example, patients with an ostomy. The Likert scale questionnaire assesses physical function and symptoms which potentially affect quality of life (Table 12.21). The QLQ-CR38 is longer and takes more time to complete, but assesses more treatment-related symptoms than the FACT-C. It was able to distinguish between patients with varying disease stage and Karnofsky performance status scores, as well as presence of an ostomy (Sprangers et al. 1999). Since publication, the QLQ-CR38 has been extensively used in EORTC trials of colorectal cancer treatments and has been validated in many languages.

Table 12.20 Functional assessment of cancer therapy-colorectal (FACT-C) quality of life instrument (Ward et al. 1999)

Below is a list of statements that other people with your illness have said are important. By circling one number per line, please indicate how true each statement has been for you during the past 7 days	
PHYSICAL WELL-BEING	
During the past 7 days	
1. I have a lack of energy	0 1 2 3 4
2. I have nausea	0 1 2 3 4
3. Because of my physical condition, I have trouble meeting the needs of my family	0 1 2 3 4
4. I have pain	0 1 2 3 4
5. I am bothered by side effects of treatment	0 1 2 3 4
6. I feel sick	0 1 2 3 4
7. I am forced to spend time in bed	0 1 2 3 4
8. Looking at the above 7 questions, how much would you say your PHYSICAL WELL-BEING affects your quality of life? (circle one number)	0 1 2 3 4 5 6 7 8 9 10
SOCIAL/FAMILY WELL-BEING	
During the past 7 days	
9. I feel distant from my friends	0 1 2 3 4
10. I get emotional support from my family	0 1 2 3 4
11. I get support from my friends and neighbors	0 1 2 3 4
12. My family has accepted my illness	0 1 2 3 4
13. Family communication about my illness is poor	0 1 2 3 4
14. I feel close to my partner (or the person who is my main support)	0 1 2 3 4
15. Have you been sexually active during the past year? No___Yes___ If yes, I am satisfied with my sex life	0 1 2 3 4
16. Looking at the above 7 questions, how much would you say your SOCIAL/FAMILY WELL-BEING affects your quality of life? (circle one number)	0 1 2 3 4 5 6 7 8 9 10
RELATIONSHIP WITH DOCTOR	
During the past 7 days	
17. I have confidence in my doctor(s)	0 1 2 3 4
18. My doctor is available to answer my questions	0 1 2 3 4
19. Looking at the above 2 questions, how much would you say your RELATIONSHIP WITH THE DOCTOR affects your quality of life? (circle one number)	0 1 2 3 4 5 6 7 8 9 10
EMOTIONAL WELL-BEING	
During the past 7 days	
20. I feel sad	0 1 2 3 4
21. I am proud of how I'm coping with my illness	0 1 2 3 4
22. I am losing hope in the fight against my illness	0 1 2 3 4
23. I feel nervous	0 1 2 3 4
24. I worry about dying	0 1 2 3 4
25. I worry that my condition will get worse	0 1 2 3 4
26. Looking at the above 6 questions, how much would you say your EMOTIONAL WELL-BEING affects your quality of life? (circle one number)	0 1 2 3 4 5 6 7 8 9 10
FUNCTIONAL WELL-BEING	
During the past 7 days	
27. I am able to work (include work in home)	0 1 2 3 4
28. My work (include work in home) is fulfilling	0 1 2 3 4
29. I am able to enjoy life	0 1 2 3 4
30. I have accepted my illness	0 1 2 3 4
31. I am sleeping well	0 1 2 3 4
32. I am enjoying the things I usually do for fun	0 1 2 3 4

(*continued*)

Table 12.20 (continued)

Below is a list of statements that other people with your illness have said are important. By circling one number per line, please indicate how true each statement has been for you during the past 7 days	
33. I am content with the quality of my life right now	0 1 2 3 4
34. Looking at the above 7 questions, how much would you say your FUNCTIONAL WELL-BEING affects your quality of life? (circle one number)	0 1 2 3 4 5 6 7 8 9 10
ADDITIONAL CONCERNS	
During the past 7 days	
35. I have swelling or cramps in my stomach area	0 1 2 3 4
36. I am losing weight	0 1 2 3 4
37. I have control of my bowels	0 1 2 3 4
38. I can digest my food well	0 1 2 3 4
39. I have diarrhea	0 1 2 3 4
40. I have a good appetite	0 1 2 3 4
41. I like the appearance of my body	0 1 2 3 4
Do you have an ostomy appliance? No__ Yes__ If yes, answer #42 and 43. If no, go to #44.	
42. I am embarrassed by my ostomy appliance	0 1 2 3 4
43. Caring for my ostomy appliance is difficult	0 1 2 3 4
44. Looking at the above 9 questions, how much would you say these ADDITIONAL CONCERNS affect your quality of life? (circle one number)	0 1 2 3 4 5 6 7 8 9 10

6.3 Pouchitis

Pouchitis can be a significant clinical problem in patient with an ileoanal J pouch for ulcerative colitis or familial adenomatous polyposis. It is a potentially easily treatable condition, but requires accurate diagnosis to distinguish it from Crohn's disease of the pouch. In 1994, Sandborn et al. published the Pouchitis Disease Activity Index (PDAI), a summative scoring system that combines patient-reported symptoms with endoscopic findings on pouchoscopy (Sandborn et al. 1994). The PDAI was found by the authors to be more sensitive when compared to older scoring systems and has probably become the standard scoring system for pouchitis for over two decades. It is frequently used in both surgical and gastrointestinal trials of the natural course and treatments for pouchitis. The combination of patient symptoms, visualization of the pouch by endoscopy, and histologic assessment of pouch biopsies for inflammation provides a comprehensive assessment (Table 12.22). A threshold of greater than 7 is generally used as the definition of pouchitis based on the PDAI. The PDAI score has been shown to correlate with severity of symptoms. In the original study of the PDAI, only patients who were symptomatic met the criteria for pouchitis, based on the PDAI (Sandborn et al. 1994).

7 Conclusion

Scoring tools in colorectal surgery can be very useful. They allow surgeons, physicians, and other healthcare professionals to communicate signs and symptoms in a standardized fashion. Using terminology such as “mild,” “moderate,” and “severe” clearly introduces physician and patient bias. On the contrary, using validated scoring systems provides universal, specific, and objective information. This may make triage, treatment discussions, physician communication, and clinical trials reporting more efficient and accurate. Most of the instruments presented in this chapter have been repeatedly well validated and are certainly widely cited in international literature. When discussing functional problems and their treatments among specialties, across languages, and in surgical literature, these types of validated instruments are certainly invaluable.

Table 12.21 EORTC colorectal cancer-specific quality of life questionnaire (QLQ-ER38) (Sprangers et al. 1999)

Content area of items	Number of items
Function	
Body image (BI)	3
Feeling less attractive	
Feeling less feminine/masculine	
Dissatisfied with body	
Sexual functioning (SX)	2
Interest in sex	
Sexual activity	
Sexual enjoyment (SE)	1
Future perspective (FU)	1
Symptoms	
Micturition problems (MI)	3
Frequency of urination/day	
Frequency of urination/night	
Pain while urinating	
Symptoms in the area of the gastrointestinal tract (GI)	5
Bloated feeling in stomach	
Abdominal pain	
Pain in buttocks	
Bothered by gas (flatulence)	
Belching	
Chemotherapy side effects (CT)	3
Dry mouth	
Thin or lifeless hair	
Different taste	
Problems with defecation (only for patients with intact sphincters) (DF)	7
Frequency of bowel movements/day	
Frequency of bowel movements/night	
Urge without producing stools	
Unintentional release of stools	
Blood with stools	
Difficulty in moving bowels	
Painful bowel movements	
Stoma-related problems (only for patients with a stoma) (STO)	7
Afraid about stoma noise	
Afraid about smell of stools	
Worry about possible leakage	
Caring for stoma	
Irritated skin	
Embarrassment	
Feeling less complete	
Male sexual problems (only for men) (MSX)	2
Problems with erection	
Problems with ejaculation	
Female sexual problems (only for women who have been sexually active) (FSX)	2
Dry vagina	
Pain during intercourse	
Weight loss (WL)	1

Table 12.22 Pouchitis Disease Activity Index (PDAI) (Sandborn et al. 1994)

Criterion	Score
Clinical	
Stool frequency	
Usual postoperative stool frequency	0
1–2 stools/day > postoperative usual	1
3 or more stools/day > postoperative usual	2
Rectal bleeding	
None or rare	0
Present daily	1
Fecal urgency or abdominal cramps	
None	0
Occasional	1
Usual	2
Fever (temperature >37.8 °C)	
Absent	0
Present	1
Endoscopic inflammation	
Edema	1
Granularity	1
Friability	1
Loss of vascular pattern	1
Mucous exudate	1
Ulceration	1
Acute histologic inflammation	
Polymorphic nuclear leukocyte infiltration	
Mild	1
Moderate + crypt abscess	2
Severe + crypt abscess	3
Ulceration per low-power field (mean)	
<25 %	1
25–50 %	2
>50 %	3

In choosing which scoring tool to utilize for a given functional problem, one can see certain commonalities in the most widely cited and utilized tools. First, the symptoms should be patient reported and account for clinically relevant symptoms or signs. The tool should be physician validated, ideally using multiple methods of validation. The results of the symptom scoring tool must be valid and reliable in capturing the severity of patient symptoms. An ideal tool has a validated threshold to mark changes in severity of the given disease and should be able to detect small but clinically significant differences. Lastly, symptom scoring tools should be easy to use (by both patients and physicians), and the results must be easy to interpret.

As new scoring instruments become available, psychometric testing should be conducted to validate the instrument, especially to compare new tools to well-validated and established scoring systems. When assessing the utility of a new scoring tool, there are two main considerations. First, a new tool may be designed to address a new problem or cluster of symptoms or a new and specific population. This, for example, was done when low anterior resection syndrome became an issue as more restorative proctectomies became commonplace for lower rectal cancers. Second, an existing symptom scoring tool may be modified to improve how one captures a symptom or functional problem. Such modified tools must be compared to their original counterparts to ensure that the validity and reliability of the instrument is not only maintained but is also improved with the new instrument. The establishment of an ideal instrument for each functional problem will continue to be pursued so as to allow physicians and healthcare providers to easily communicate and direct treatments.

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Diagnosis in Coloproctology: From the General Practitioner to the Tertiary Referral Center

13

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Contents

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Abstract

Anorectal disorders are daily encountered by general practitioners, gastroenterologists, and general surgeons. Most symptoms and complaints that patients refer are common and may be caused by a wide spectrum of benign and malignant conditions. Many anorectal conditions are benign and can be easily managed. Suspicion for colorectal cancer should always be remembered and ruled out when indicated. A correct diagnosis is basic to prevent benign conditions becoming invalid or affecting quality of life. It also prevents malignant conditions presenting as advanced or noncurable diseases. To achieve this, the fundamental steps in the diagnostic process are to collect a careful medical history from the patients and a comprehensive physical examination. The aim of this chapter is to describe how to approach patients with anorectal disorders, focusing firstly on the most common coloproctologic symptoms, secondly on how to perform the anorectal physical examination, and lastly on analyzing the diagnostic process of the most frequent coloproctologic diseases from the point of view of both the general practitioner and the colorectal surgeon.

1 Introduction

Anorectal disorders are daily encountered by general practitioners, gastroenterologists, and general surgeons. It has been estimated that up to 75 % of individuals have symptomatic hemorrhoids at least once in their lifetime. Most symptoms and complaints that patients refer are common and may be caused by a wide spectrum of benign and malignant conditions. Some of these may not even be directly related to coloproctologic disorders. Although many anorectal conditions are benign and can be easily managed, suspicion for colorectal cancer should always be remembered and the disease must be ruled out when indicated. A correct diagnosis is basic to prevent benign conditions becoming invalid or affecting quality of life. It also prevents malignant conditions presenting as advanced or noncurable diseases. To achieve this, the fundamental steps in the diagnostic process are to collect a careful medical history from the patients and a comprehensive physical examination keeping in mind that, especially for coloproctologic pathology, patients may delay their consultation or occult important and relevant personal information, because of embarrassment or fear of cancer (Table 13.1).

The aim of this chapter is to describe how to approach patients with anorectal disorders, focusing firstly on the most common coloproctologic symptoms, secondly on how to perform the anorectal physical examination, and lastly on analyzing the diagnostic process of the most frequent coloproctologic diseases from the point of view of both the general practitioner and the colorectal surgeon.

2 Symptoms

A detailed and well-collected medical history can provide a diagnosis by itself for many coloproctologic disorders. Initiating the interview following a "symptom-based" approach is advisable in order to relax and put at ease the patient before performing the anorectal examination. Embarrassment does not help the patient when explaining symptoms, therefore directing questions is helpful (Table 13.2).

Anal pain. When visiting a patient with **anal pain,** it is important to investigate the localization (inside or around the anus), chronology (acute pain or chronic pain), the pain characteristics (intermittent or constant), if it is associated with bowel movements, and especially if those worsen or improve the pain. Association with other clinical conditions such as constipation, diarrhea, bleeding, mucus or purulent anus discharge, perianal mass, or tenderness and systemic symptoms like fever must be investigated too. The differential diagnosis for anal pain includes perianal abscesses, anal fissure, thrombosed external hemorrhoid, chronic proctalgia (levator ani syndrome), and proctalgia fugax, among others.

Rectal bleeding. Any kind of rectal bleeding must be considered seriously until cancer is ruled out. Investigation should focus on the starting

Table 13.1 Sign and symptoms in the most frequents coloproctologic pathology

	Bleeding	Anal/perianal pain	Itching	Perianal mass	Prolaps	Constipation	Diarrhea	Soiling	Fecal incontinence	Rectal tenesmus	Fever	Abdominal pain	Abdominal mass
External hemorrhoids	–	++ **(if complicated)**	+	++ **(if complicated)**	–	–	–	–	–	–	–	–	–
Internal hemorrhoids	++	–	–	–	++	–	–	+	–	–	–	–	–
Anal fissure	+	++	–	–	–	+	–	–	–	–	–	–	–
Perianal abscess	–	++	–	++	–	–	–	–	–	+	++	–	–
Perianal condylomas	+	+	++	++	–	–	–	–	–	–	–	–	–
Anal fistula	+	+	+	+	–	–	–	–	–	–	–	–	–
Diverticulosis	–	–	–	–	–	+	+	–	–	–	–	+	–
Diverticulitis	–	–	–	–	–	–	+	–	–	–	++	++	++
Rectal prolaps	+	+	+	+	++	++	+	++	+	+	–	–	–
Anal cancer	+	++	++	++	–	+	–	+	+	++	–	–	–
Rectal cancer	++	+	–	–	–	++	–	–	+	++	–	+	–
Colon cancer	+	–	–	–	–	++	+	–	–	–	–	+	+

"–" uncommon; "+" common; "++" very common

Table 13.2 Complementary explorations in the most frequents coloproctologic pathology

	Anoscopy	Rigid rectoscopy	Flexible sigmoidoscopy/ colonoscopy	Endoanal US	Endorectal US	Abd US	Pelvic MR	Abd MR	CT	RX	Defecography/ cinedefecography	MRI defecography
Internal hemorrhoids	++	–	+	–	–	–	–	–	–	–	–	–
Anal fissure	++	–	+	–	+	–	–	–	–	–	–	–
Perianal abscess	++	–	+	?	–	–	?	–	?	–	–	–
Anal fistula	++	–	+	?	–	–	?	–	?	–	–	–
Perianal condylomas	++	+	–	–	–	–	–	–	–	–	–	–
Anal cancer	++	–	++	++	++	–	++	–	++	–	–	–
Rectal cancer	–	++	+	–	++	?	++	–	++	–	–	–
Retrorectal tumors	–	+	?	–	?	–	++	–	++	–	–	–
Prolapso rectal	–	–	+	–	–	–	–	–	–	–	++	?
Rectocele	–	–	–	–	–	–	–	–	–	–	++	?
Crohn's colitis	–	–	++	–	–	–	–	?	?	?	–	–
Ulcerative colitis	–	–	++	–	–	–	–	–	–	?	–	–
Irritable bowel syndrome	–	–	?	–	–	–	–	–	–	–	–	–
Diverticulosis	–	–	++	–	–	–	–	–	–	–	–	–
Diverticulitis	–	–	–	–	–	?	–	–	++	–	–	–
Colon cancer	–	–	++	–	–	?	–	–	++	–	–	–

"–" no necessary; "+" differential diagnosis; "++" diagnosis; "?" optional; *US* Ultrasound; *MRI* magnetic resonance; *CT* computerized tomography point, if it is continues or intermittent, occasional or frequent. The patient should also be interrogated about its relationship with bowel movements. The color of the blood, if the blood forms clots, drips in the toilet after defecation, paints the stools, or stains the toilet paper are important questions that the physician should ask. Associated symptoms such as constipation of new onset, diarrhea, mucous discharge from the anus, anal pain during defecation, rectal tenesmus, asthenia, abdominal pain or discomfort, and recent history of nonvoluntary weight lost must be investigated. The age, significant family history of bowel disease or cancer, and persisting bleeding in spite of previous treatment are to be considered during the diagnostic process. Differential diagnosis for **rectal bleeding** must consider benign conditions such as hemorrhoids, anal fissure, rectal prolapse, rectal solitary ulcer and colorectal polyps, as well as malignancies.

Perianal itching. **Perianal itching** or burn sensation in the perianal area is a very commonly reported symptom. Since perianal itching is associated with a wide range of mechanical, infectious, dermatologic, hygienic, and systemic conditions, it is of great importance to approach patients collecting an accurate anamnesis, general and specific, although some cases turn out to be idiopathic (*pruritus ani*). First of all, it is essential to know if dealing with a chronic problem or one of recent onset, if it is constant during the day, at night, or intermittent, and investigate its intensity (continuous need to scratch and/or does not permit sleeping). Given that causes of itching could be related to bowel movement, the presence of diarrhea must be investigated, as well as the sensation of seepage or moisture around the anus, mucous soiling, or fecal incontinence. It is important to ask patients about their intimate hygienic habits because those can be related to overcleaning of the perianal area with soap, frequent scrubbing, as well as direct application of certain types of soap, perfume, and other irritant cosmetic agents on or around the anus. Because certain substances like cola, coffee, citric foods or drinks, chocolate, and calcium have been found to be potentially related to pruritus ani, any abuse of these foods should be investigated. Medication for other pathologies like certain chemotherapy, colchicine, and quinidine as well as topical use of corticoids in perianal cutaneous area have also been related to anal itching, thus its use should be included in the interrogatory.

Fecal incontinence. Normal continence depends on the balance between consistency of the stool (chronic diarrhea or liquid stool), anal sphincter function and/or its integrity, and the normal central and peripheral nervous system regulation. The approach to patients referring **fecal continence** problems must be global, so systemic nervous illnesses, diabetic neuropathy, and previous complex pelvis injuries must be ruled out. Before making a choice among the different diagnostic tools that we can use to better define our patient's incontinence level, the anamnesis is going to be, once again, crucial. We need to know when the problem started, if the patient is capable of controlling gas, as well as liquid and solid fecal stool. Frequency of the episodes of **incontinence** (occasionally or daily) and the need for diaper use should be investigated. The Wexner (Jorg and Wexner 1993) and Vaizey score (Vaizey et al. 1999) and the creation of a defecation diary are both tools of great use as they allow to have a better knowledge on how incontinence affects the patient quality of life and, therefore, helps with the design of the best diagnostic-treatment schema. Any possible triggers such as recent coloproctologic and gynecologic surgery, pelvic radiation, and pelvic and anal injuries must be investigated. A thorough obstetric medical history must always be collected, as a lesion to the sphinteric complex or the pudendal nerves may have occurred during delivery and might contribute to the incontinence. Association with other symptoms such as urinary incontinence, rectal and genital prolaps, pelvic and anal pain, new-onset constipation, and rectal **tenesmus** must be taken into consideration in the differential diagnosis between idiopathic or secondary fecal incontinence.

Perianal mass. Anal or **perianal masses** may be due to a wide spectrum of benign and malignant conditions. Before proceeding to the anorectal exploration, we must know when and how the patient first noticed the mass, if it is painful or not, and if it has been growing over time (days or months). Associated symptoms like anal or perianal pus discharge, itching, bleeding, fecal incontinence of recent onset, changes in bowel movement habit, rectal tenesmus, pelvis or back pain, and fever need to be investigated. Localization of the mass and its relations with the sphincter complex and pelvic floor are important for surgical treatment.

Prolapse. When approaching a patient that refers *"something is coming out from my anus,"* it is important to know when the patient first noticed the problem. Association with pain, bleeding, mucus discharge, and itching must be ruled out. Any relationship between the symptoms and changes in bowel movement frequency must be investigated, especially constipation of recent onset, episodes of **fecal impaction** that may require digitation and/or enemas, and fecal and/or urinary incontinence. Given that many times during examination at the office, the referred **prolapse** may not be evident, the physician must enquire about the protrusion features, if it is reducible, if it comes out and back inside spontaneously or needs to be reduced manually. It is also important to enquire about transvaginal protrusion. Previous pelvic surgery such as hysterectomy must be investigated too. Differential diagnosis must be made between internal prolapse versus external hemorrhoids, rectal prolapsing polyps, among others.

Constipation. **Constipation**, defined as infrequent or difficult, even painful bowel movements, is a symptom related to a number of causes. In a patient who complains of constipation, organic problems such as colorectal cancer, or colonic stenosis must be excluded before assuming the cause of the problem is functional. The presence of systemic associated signs and symptoms such as anorexia, anemia, asthenia, abdominal pain or distension, abdominal palpable mass, and rectal bleeding must be ruled out. The use of laxatives or enemas must be recorded. Previous pelvic or abdominal surgery or any other systemic diseases that can alter the intestinal transit must be investigated. A complete dietary history must be obtained, as low intake of fiber and fluids as well as a sedentary lifestyle could be the cause of the patient condition. When focusing on functional constipation disorders, it is important to consider stool consistency (loose, lumpy, or hard), rule out straining during defecation, number of bowel movements per week, sensation of incomplete evacuation, or the need for the use of manual maneuvers to facilitate defecation like digital evacuation (Roma III Diagnostic Criteria for Functional Constipation).

3 Anorectal Examination

It is advisable to start examining the abdomen to rule out any abdominal abnormality that might be related to the problem.

A proper anorectal examination has three basic steps: inspection, **digital rectal examination,** and **anoscopy**. Patients' dignity minimizing embarrassment should be always kept in mind, and the patient should feel as comfortable as possible. Draping should be used to expose only the perineum.

The patient will then be asked to lie in left lateral decubitus position, with the knees bent toward the abdomen and the pelvis at the edge of the table. This is the usual and best tolerated position for anal examination in the office. Alternatively, a jackknife position for improved anorectal visualizations could be chosen if needed. The presence of an assistant at the time of examination is always recommended. It is important to walk the patient through the different steps that will be followed, for further comfort. It must be remembered that directional terms such as anterior/posterior and right side/left side are preferred instead of clock descriptions to document the findings, in order to avoid confusion.

Inspection. The anorectal area is exposed distracting the buttocks and examination of the perianal area started, looking at the condition of the perianal skin, presence of hemorrhoids (external, thrombosed, prolapsed, or skin tags), external condylomas, sentinel tags related to anal fissures, external fistula-in-ano opening, anal protrusions, and any other perianal mass should be reported.

Anal fissures can be seen by distracting the perianal area with the fingers and asking the patients to performe a valsalva effort. The buttocks, base of the scrotum, vulva, and sacrococcyygeal region should always be examined.

Digital Rectal Examination. Palpation of an external lesion provides important information on its nature (fluctuant, fixed, infiltrant, and painful). Therefore, if the patient is not experiencing pain, a digital examination should be performed. A lubricated gloved index finger should be gently inserted in the anal canal and rectum examining the complete circumference. Attention must be focused on the presence of any palpable mass or tenderness. If detected, attention should be paid to its size, features (fixed or mobile, soft or hard, infiltrating, fluctuant, painful, ulcerated, exofitic, etc.), position, relationship to the anorectal ring, and distance to the anal verge. Digital examination must include the assessment of the sphinteric tone (resting and squeeze) as well as the symmetry and defects of the anal sphincter. A transanal prostate palpation could be performed in men. In case of pain in the set of an acute inflammatory condition, digital examination could be inconclusive and a proper exploration under anesthesia performed.

Anoscopy. The office evaluation of the anorectum should be completed with the anoscopy. The exploration can generally be performed with minimal patient discomfort if detailed explanation of the procedure is previously given to the patient. Different anoscopes are commercially available. It is important to achieve a good exposition with a good light focus. A well-lubricated anoscope is gently inserted in the anal canal upper to the rectum and slowly withdrawn under direct vision of the entire circumference looking for polyps, masses, internal condylomas, internal hemorrhoids, fistula-in-ano internal orifice, fissure, signs of proctitis, and bleeding. The dentate line must be visualized and should be used as a landmark for lesion description. During anoscopy, it is useful to ask the patient to do valsalva; hemorrhoids or rectal prolapse then become evident. Rigid rectoscopy, flexible sigmoidoscopy, and complete colonoscopy are indicated in selected patients.

4 Anorectal Benign Diseases

4.1 Hemorrhoids

Symptomatic hemorrhoidal disease may affect external hemorrhoids that are defined as the dilated inferior hemorrhoid plexus distal to the dentate line. The external hemorrhoids may swell and patients with acute external hemorrhoid thrombosis present with a painful lump at the anal verge.

Internal symptomatic **hemorrhoids** are associated with bleeding bright red and painless that occurs at the end of defecation with or without the presence of prolapse (Kluiber and Wolff 1994). Symptomatic internal hemorrhoids are classified in four degrees. First degree: bulge into the anal canal and bleeding with defecation; second degree: prolapse during defecation and spontaneous reduction; third degree: prolapse during defecation that requires digital reduction; fourth degree: prolapse that cannot be manually reduced (Cataldo et al. 2005)

4.1.1 Examination

Inspection may reveal variable degrees of protrusion of internal hemorrhoids or normal appearance. The severity of the prolapse can be evaluated by straining while sitting in the toilet. At the same time, the degree of descending perineum can be evaluated.

Digital examination will exclude anal canal neoplasms and will enable assessment of the tone of the anal sphincter.

4.1.2 Investigation

A regional examination should include anoscopy. With the anoscope in place, the patient is asked to strain as if having a bowel movement so that the amount of prolapse can be assessed.

Flexible sigmoidoscopy or colonoscopy must be performed to exclude carcinoma, adenoma, and inflammatory bowel disease. Complete colonic evaluation is recommended for individuals over 40 years of age with a risk for colorectal cancer and for all patients 50 years and older (Imperiale et al. 2002)

4.2 Anorectal Abscess

Most common symptoms are related to inflammation: redness, warmth, pain, and tumor. Depending on the phase of development, other local signs and symptoms present such as difficulty in sitting down and painful defecation, constipation due to **sphincter spasm**, purulent discharge when spontaneous drainage has occurred, and fever (Abcarian 1976). Swelling and induration in the perianal region may not occur with an intersphincteric abscess (Vasilevsky and Gordon 1984) making that diagnosis highly dependent on suspicion.

4.2.1 Physical Examination

Inspect the perianal area for signs of inflammation or **purulent discharge**. Palpation of the induration, localized tenderness, and fluctuance reveal the location of the abscess (Parks and Thomson 1972).

Rectal examination is exquisitely painful or impossible and not essential at the acute phase of disease

4.2.2 Complementary Examinations

Anoscopy may show pus arising from an anal crypt or the base of a chronic fissure. Flexible sigmoidoscopy or colonoscopy allows examination of the rectal and colonic mucosa and helps to rule out Crohn's disease. A rectal biopsy should be performed if inflammatory bowel disease is suspected. These examinations, when indicated, are usually performed once the abscess has been drained.

Imaging including **endoanal ultrasound** (EUS), computed tomography (CT), and **magnetic resonance imaging** (MRI) can reveal deep or complex anorectal abscesses and can be useful in patients with multiple or recurrent abscess formation (Garcia-Granero et al. 2014).

4.3 Anal Fistula

Pain, discharge, bleeding, or a history of an abscess that was drained either surgically or spontaneously are common as fistulas represents the chronic phase of the disease.

4.3.1 Physical Examination

An external opening may be seen discharging pus. According to **Goodsall's rule**, an opening seen posterior to a line drawn transversely across the perineum will originate from an internal opening in the posterior midline of the anal canal, and an anterior external opening will originate through a radial fistula from the nearest crypt (Cirocco and Reilly 1992). The internal opening in most cases is not apparent.

Digital rectal examination may reveal a cordlike structure in cases of superficial fistulas.

4.3.2 Complementary Examinations

Anoscopy should be performed to try to localize a primary opening in the dentate line. Flexible sigmoidoscopy or colonoscopy allows examination of the rectal and colonic mucosa to determine whether there is underlying proctitis and helps to rule out Crohn's disease or inflammatory processes that sometimes may drain through the pelvis and reach the perineum.

During anal EUS, hydrogen peroxide injection through the external opening is helpful in identifying the fistula tract, and MRI may be of help with complex fistulas (Hussain et al. 1996).

4.4 Anal Fissure

Most common symptom is intense pain in the anus during and after defecation. Usually, it is described as a tearing sensation during the actual passage of stool and may persist from a few minutes to hours. The pain is usually accompanied by a small amount of bright red blood per rectum (Hananel and Gordon 1997) usually staining paper.

4.4.1 Physical Examination

Inspection is the most important step. Gentle separation of the buttocks and anal margin can be sufficient to expose the external extent of the open fissure, in posterior midline in 99 % of males and 90 % of females. Spasm may keep the anal orifice closed, and the finding of spasm of the sphincter is suggestive of anal fissure (Beck and Wexner 1992).

Some patients may be too tender for this maneuver, and an examination under anesthesia may be necessary.

A triad of a chronic fissure has been described including **sentinel pile**, anal ulcer, and hypertrophied anal papilla (Petros et al. 1993)

4.4.2 Complementary Examinations

Anoscopy will reveal the fibers of the internal anal sphincter and a hypertrophied anal papilla at the ápex if the fissure is chronic.

Rigid rectoscopy or **flexible sigmoidoscopy** at a subsequent visit should be performed to exclude distal inflammatory bowel disease or concurrent malignancy.

4.5 Anal Condylomata

Anal condylomas, also known as anal warts, are related to the human papillomavirus (HPV) infection and represent one of the most common sexually transmitted diseases in the world.

4.5.1 Symptoms

Patients with anal warts generally refer to the physician for anal bleeding with defecation, anal itching, perianal wetness, pain while cleaning with toilet paper, or simply because they note a perianal lump.

4.5.2 Physical Exploration

Macroscopic aspect of condylomas is often clearly diagnostic. They can appear as cauliflower-like masses of different sizes or pinhead-sized lesions. Multiple lesions are common. In the advanced disease, the lesions may affect the entire anal circumference. In patients with perianal condylomas, the anal canal must be explored with a digital rectal examination and anoscopy to rule out intra-anal lesions.

4.5.3 Complementary Explorations

Because associated genital wart lesions are very common, a careful perineal exploration and gynecologic evaluation in women with a Pap smear is mandatory. Penis and distal urethra must be explored in men. Rigid sigmoidoscopy could be useful to rule out rectal concomitant sexually transmitted or infectious diseases. Tests for other sexually transmitted diseases like HIV, syphilis, and gonorrhea should be evaluated. Differential diagnosis of anal warts must be made with syphilis anal lesion (Condyloma latum), anal hypertrophic papillae, and molluscum contagiosum. Excision biopsy must be considered for recurrent or atypical lesions and in HIV-positive patients. Since that HPV infection and anal condylomas are been associated with anal intraepithelial neoplasia (AIN), experts recommend HIV-positive patients to participate in anal squamous cancer screening programs with anal cytology and high-resolution anoscopy. Sexual partners of persons with genital warts should be counseled for physician evaluation (Beck and Whitlow 2009).

5 Anorectal Tumors

5.1 Rectal Cancer

Rectum cancer may be asymptomatic and discovered as part of a routine proctosigmoidoscopy or colonoscopy.

The most common symptom is **bleeding**. Altered bowel habits may be present with constipation or diarrhea or decreased stool caliber because of stenosis of the lumen (Beart et al. 1995).

If located low in the rectum, a carcinoma may cause pain because of invasion of the anal sphincter and a feeling of incomplete evacuation known as tenesmus.

Abdominal distention may occur from circumferential narrowing of the rectosigmoid or **anal canal** evolving to bowel obstruction.

5.1.1 Physical Examination

Digital rectal examination should assess tumor size, position (anterior, posterior, lateral), fixity of the lesion to the underlying rectal wall and pelvic structures, and distance of the tumor from the anorectal ring and the anal verge (Nicholls et al. 1982). Physical examination may also demonstrate findings of pelvic mass, hepatomegaly, or enlarged Virchow's or inguinal nodes.

5.1.2 Complementary Examinations

Rigid rectoscopy or flexible sigmoidoscopy records the lesion's size, lower margin distance from the anal verge, and the percentage of circumference of the lumen occupied by the lesion (Nivatvongs and Fryd 1980) as well as its gross appearance, whether the lesion is polypoid or ulcerating.

A colonoscopy should be performed to rule out synchronous colon neoplasms. Multiple biopsies by rectoscopy or colonoscopy should always be taken to confirm the diagnosis and establish the grade of the carcinoma.

Endorectal US (Marohn 1997) and Pelvic MRI (Beets-Tan and Beets 2003) are currently used to preoperatively assess local-regional stages being essential to management strategy. Toracoabdominal CT assesses for metastasis (Kerner et al. 1993).

5.2 Anal Cancer

5.2.1 Symptoms

Anal cancer has a wide range of clinical signs and symptoms. In its early stages, the disease clinical presentation includes very common coloproctologic symptoms, such as anal pain, tenesmus, bleeding, anal discharge, and persisting anal itching, that can delay its diagnosis. Up to 20 % of patients with early anal cancer may be asymptomatic when diagnosed (Scholefield et al. 2011). In advanced stages, the disease can cause soiling or fecal incontinence, especially if the disease compromises the sphincteric apparatus. In case of palpable inguinal nodes, anal cancer must be ruled out, even in the absence of anal symptoms (Glynne-Jones et al. 2014).

5.2.2 Physical Exploration

The disease typically appears as an ulcerated mass or fissure, generally exophytic, with indurate margins and irregular thickening. Perianal skin fistulization and concomitant inflammatory signs could be present. Digital examination of the anus, rectum, and vagina in female patients provides important data on the localization of the tumor, its local extension, the relationships with the anal sphincteric apparatus, and the presence of anal stenosis. Distinction between anal canal and anal margin malignancies must be well established, as diseases affecting the anal margin have a much aggressive behavior. When intense pain makes it difficult or even impossible to perform a proper exploration, it should be made under anesthesia. Inguinal regions must always be explored looking for pathological inguinal nodes. Gynecologic exploration in women should be performed in order to rule out concomitant squamous genital neoplasia (Salmo and Haboubi 2011).

5.2.3 Diagnostic Tests and Extension Study

Patients under clinical suspicion of anal and perianal malignancies must be promptly referred to a third-level hospital for evaluation in a specialized coloproctologic unit. All new patients, with confirmed diagnosis of anal cancer after rectoanoscocopy and positive biopsy, will be taken care of by a multidisciplinary team that includes the colorectal surgeon, dedicated clinical oncologists, radiologists, and pathologists. Biopsy will provide the histological patterns of the tumor (squamous cell carcinoma, anal adenocarcimoma, verrucous carcinoma, small-cell carcinoma, neuroendrocrine melanoma, lymphoma, leiomyosarcoma, among others). Colonoscopy should be performed to rule out synchronous colorectal neoplasm based on standard age and risk profile assessment. Magnetic resonance imaging (MRI) is fundamental to assess locoregional disease, as it gives tumor size, depth of invasion, infiltration of the anal sphincter and levator ani muscle, as well as the invasion of other pelvic structures such as vagina, urethra, prostate, and bladder. Perirectal, iliac, and inguinal nodal status is also assessed. Endoanal ultrasound is a useful tool for staging small lesions. Positron emission tomography/computed tomography with fluorodeoxyglucose (FDG-PET/CT) is recommended in identifying systemic lymph node involvement. In the setting of clinically palpable nodes in the groin area or the existence of inguinal nodes in CT or MRI greater than 10 mm, biopsy could be considered for confirmation (Branagan 2011). The existence of distal metastasis must be assessed by thoraco-abomino-pelvic computed tomography (Steele et al. 2012). Anal cancer staging should be performed in accordance with the American Joint Committee on Cancer (AJCC, 7th edition) and the International Union Against Cancer (Edge et al. 2010). Tumors of the anal margin (below the anal verge) are classified as skin malignancies (AJCC, 6th edition) (Sobin et al. 2009). Considering that anal cancers are more common in HIV patients and that the highly active antiretroviral therapy (HAART) affects the treatment and the outcome is recommended, perform an HIV test and CD4 counts in all patients diagnosed with anal cancer (Renehan and O'Dwyer 2011).

5.2.4 Screening and Prevention

Squamous anal cancer and high-grade anal intraepithelial neoplasia (AIN III) have been associated with chronic human papillomavirus (HPV) infection (subtypes 16 and 18). In patients at increased risk of persistent HPV infection (frequent anal intercourse and high number of sexual partners) and altered host response as HIV infection and other cause of immunosuppressant condition has been observed a higher incidence of anal cancer respect the rest of population. To prevent progression from dysplasia to invasive cancer, screening programs with anal cytology and high-resolution anoscopy and treatment of the lesion with high-grade dysplasia have been proposed in HIV men who have sex with men (MSM) and HIV women with a history of anal intercourse or other HPV-related anogenital malignancies (Scholefield et al. 2011).

6 Retrorectal Tumors

Retrorectal tumors are frequently asymptomatic. If pain is present, it is poorly localized as low back, perianal, or rectal and is frequently postural, associated with sitting or standing and often related to local trauma such as a fall on the sacrum or coccyx. If the sacral plexus is involved, patients may refer pain in the legs or buttocks. Infection may occur with fever and perianal suppuration.

Large masses may interfere with the passage of stool or with the pelvic parasympathetic supply; this may cause disturbances in bladder function.

6.1 Physical Examination

Inspecting the perianal area, a postanal dimple may suggest the presence of a developmental cyst.

Careful digital rectal examination, under anesthesia if appropriate, and a solid retrorectal mass may be obvious (Jao et al. 1985).

6.2 Complementary Examinations

Procedures to classify the lesion as cystic or solid and to delineate its extent must be done. Pelvic CT (Stewart et al. 1986) or MRI (Wolpert et al. 2002) show the amount of sacrum or central nervous system and pelvic structure involvement. Endorectal US or endoscopy determine involvement of the rectal wall.

Preoperative biopsy of a lesion considered to be operable is not recommended, because of the risk of seeding of malignant cells if the lesion is solid, infection if the lesion is a cyst, and meningitis if meningocele is present.

7 Pelvic Floor Diseases

7.1 Rectal Prolapse

A mass extrudes initially only with defecation; later, this extrusion occurs with the mildest straining, or even when the patient stands up.

In the early stage, symptoms may include difficulty in bowel regulation, discomfort, the sensation of incomplete evacuation, and tenesmus.

Tenesmus, bleeding, and mucus discharge are associated symptoms for advanced-stage prolapse. Incontinence may range from mucus leakage to complete fecal incontinence.

7.1.1 Physical Examination

Examination of perianal area and visualization of everted bowel with concentric folds. During the early stage, if prolapse is not evident, the patient should be examined while straining. In its florid form, a large red mass is quite unmistakable.

7.1.2 Complementary Examinations

A colonoscopy is indicated to exclude another associated disease such as a neoplasm, inflammatory bowel disease, or diverticular disease (Rashid and Basson 1996).

If prolapse cannot be demonstrated, **defecography and cinedefecography** may show an internal intussusception (Dvorkin et al. 2005). MRI-defecography can help to diagnose underlying anatomic and pathophysiologic disorders (Healy et al. 1997).

7.2 Rectocele

Most common symptoms are difficulty to empty the rectum during defecation. These patients will often feel a protrusion of tissue through the introitus on straining and usually will need to push upward on the perineal body, or to apply backward pressure on the posterior wall of the vagina, to aid in rectal emptying.

7.2.1 Physical Examination

On digital rectal examination, one can easily feel the anterior wall defect.

7.2.2 Complementary Examinations

Cinedefecography is usually the most helpful diagnostic study. Defecography will show the size of a rectocele as well as the remaining contrast in the **rectocele** after emptying of the rectum. Anal manometry or endorectal ultrasound may help to detect any associated sphincter dysfunction.

As a general rule, functional pelvic disorders associate and present as complex combined entities. It must be kept in mind during clinical investigations and when concluding in order to set surgical indications.

8 Colonic Nonneoplastic Diseases

8.1 Crohn's Colitis

Most common symptoms are abdominal pain, diarrhea, and weight loss.

Abdominal pain is a characteristic of ileocolitis and diarrhea and bleeding per rectum typical symptoms of Crohn's colitis (Hanauer and Meyers 1997).

8.1.1 Physical Examination

Other than abdominal findings, perineum, anal canal, and extraintestinal manifestations, attention must be paid to overall and nutrition status as these patients may fear eating and lose nutrients after chronic diarrhea.

8.1.2 Complementary Examinations

Colonoscopy and biopsy is the most sensitive tool making the initial diagnosis of Crohn's colitis and in determining its extent and severity. However, sometimes it is impossible by endoscopy and even for the pathologist to distinguish Crohn's colitis from ulcerative colitis (Schratter-Sehn et al. 1993).

A flat and upright abdominal x-ray can exclude colonic distention or obstruction.

A computerized tomography scan has been described to detect abscesses and rule out extraintestinal complications. MRI is helpful assessing the activity (Koh et al. 2001).

8.2 Ulcerative Colitis

Most common symptoms are rectal bleeding in the main manifestation. Tenesmus, constipation, abdominal pain, and extraintestinal manifestations may occur in some patients (Sands 2004).

8.2.1 Physical Examination

There may be evidence of malnutrition with anorexia and weight loss. The patient will often appear cushingoid and with complications of corticosteroid treatment (Corman 1993). The perineum is usually disease free.

8.2.2 Complementary Examinations

The rectum is involved in all cases and diseased mucosa continuous from distal to proximal. Therefore, proctoscopy or flexible sigmoidoscopy could be sufficient in order to assess a single episode. Moreover, colonoscopy is contraindicated during the acute attack due to the risk of perforation. After patients have recovered, it should be performed to determine the extent of the disease although a finding of normal rectum almost always excludes ulcerative colitis. A biopsy may not distinguish between ulcerative colitis and **Crohn's disease**. (Corman 1993). In the long term, colonoscopy biopsies help in monitoring dysplasia.

A flat and upright abdominal x-ray is useful to assess bowel distention, particularly in a patient with toxic colitis.

A double-contrast barium enema is useful for evaluation of patients with UC (Altaras 1994). However, it is contraindicated in an acute stage.

9 Irritable Bowel Syndrome

Frequently, patients complain of chronic gastrointestinal symptoms including abdominal pain, altered bowel habits alternating constipation, and diarrhoea and abdominal bloating.

Other symptoms are pain relieved by defecation, incomplete evacuation of the rectum, mucous discharge per rectum, looser stools, and increased frequency with pain onset (Taley et al. 1991).

Symptoms are common to many intestinal entities leading to differential diagnosis more than to a single clinical suspicion.

9.1 Physical Examination

Nonspecific physical findings are characteristic.

9.2 Complementary Examination

Irritable bowel syndrome diagnosis is by exclusion. Invasive procedures other than sigmoidoscopy or colonoscopy if a patient is under 40 should be reserved for those patients with atypical symptoms, abnormal physical findings, or recent change in bowel habits suspicious for colon cancer (Schuster 1993).

9.3 Diverticulosis/Diverticulitis

Most often, **diverticulosis** is asymptomatic. Some patients may experience the presence of left iliac fossa or lower abdominal discomfort as well as vague unspecific intestinal complaints. Left iliac fossa pain and tenderness, fever, and tachycardia are the most common symptoms and signs of inflammation. Some patients experience chronic symptoms secondary to stenosis.

9.3.1 Physical Examination

In patients with diverticulosis, no abnormal abdominal examination will be found. In patients with diverticulitis, lower abdominal tenderness with no appreciable mass in case of diverticular inflammation; tender mass and voluntary guarding in the lower abdomen if peridiverticular abscess or phlegmon is present and persistent fever with severe diffuse abdominal pain and septic general status when generalized peritonitis develops.

9.3.2 Complementary Examinations

In the uncomplicated scenario, the diagnosis is most often established through a barium enema examination, but colonoscopy is mandatory to exclude a colonic carcinoma (Hunt 1979).

In acute diverticulitis, ultrasound and particularly abdominal CT are important for the diagnosis but also to differentiate disease confined to the colonic wall from pericolic extension or abscess (Ambrosetti et al. 2000) (Ripolles et al. 2003).

10 Colonic Neoplastic Pathology

10.1 Colon Cancer

The most common symptom is bleeding. In right-sided lesions, this may be occult and cause of anemia. Depending on the location of the carcinoma in the bowel, bleeding may be either black, dark purple, or bright red (Beart et al. 1983).

Either change in bowel habits, constipation, or diarrhea can be observed. Changes in stool caliber are referred in some cases. Abdominal pain may be vague and poorly localized or colic in relation to defecation.

10.1.1 Physical Examination

Most often, abdominal examination does not reveal specific findings. Nutrition status is not usually altered. Advanced cases may present pale, with hepatomegalia or a palpable abdominal mass.

10.1.2 Complementary Explorations

Patients with positive screening of **hemoccult stools** demand prompt evaluation.

Colonoscopy and biopsy is preferable (Bernard et al. 1987). Chest radiography is routinely obtained for preoperative staging as well as toracoabdominopelvic CT or liver ultrasound (Kerner et al. 1993) to rule out metastatic disease or local extension of the tumor. Carcinoembryonic antigen (CEA) values are helpful in monitoring disease in the long term.

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Instruments for the In-Office Diagnosis 14

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Contents

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Abstract

The diagnosis and management of colorectal disease that is initiated during outpatient consultation frequently requires specialized equipment and instrumentation. Adequate positioning of patients for examination and the performance of in-office procedures often require maneuverable tables. Additionally, unique tools facilitate the performance of procedures in the clinic. In-office **endoscopy**, including anoscopy, rigid proctosigmoidoscopy, and flexible sigmoidoscopy, is a valuable diagnostic and therapeutic tool that has become common place. Advances is technology have made **anal manometry** and **anorectal ultrasonography** readily available in the outpatient setting. The maintenance and cleaning of these instruments as well as the disposable of non-reusable equipment and medical waste is regulated by local, state, and federal entities. Knowledge of all aspects of the instruments for in-office diagnosis in colorectal disease is key to the creation and maintenance of a successful practice.

1 Introduction

Once a problem-specific history has been taken, a focused physical examination should be performed. Physical examination for manifestations of colorectal diseases often requires

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specialized equipment to obtain the requisite information for diagnosis and treatment. Examination of the perineal region in the office includes visual inspection and digital palpation complemented by endoscopic, physiologic, and radiologic evaluations. Colorectal and general surgeons are the best equipped to complete this task as physical examination of the distal colon, rectum, and anus often requires invasive techniques.

2 Table Requirements

The examination table is where the practice of medicine takes place – the interaction between the physician and the patient that is initiated while obtaining the history is completed during the physical examination. The **examination table** should be maneuverable enough to accommodate several positions (Fig. 14.1a–c). Additionally, the table must allow for adequate exposure and efficient use of equipment as well as provide for patient and examiner comfort. These tables must have sufficient patient weight capacity and be made of materials that are durable and easily cleaned between patients. Tables that allow the surgeon to place patients in a wide variety of position including **prone jackknife**, **lithotomy**, **left lateral,** and **knee-to-chest positions** are key to the practice of colorectal surgery. The selection of patient position is at the discretion of the examiner, however, there are some advantages and disadvantages of each.

Fig. 14.1 Examination table configurations for examination positions. (**a**) Prone jackknife position. (**b**) Lithoomy position. (**c**) Left lateral (Sims' position)

3 Patient Positions

3.1 Prone Jack-Knife Position

The prone jackknife position provides excellent exposure of the perineum, anus, and gluteal cleft. This position requires the use of a maneuverable table to facilitate exposure. The patient is asked to kneel on the table platform, bending forward and placing their chest on the table with elbows forward, palms on the table, and the back in a slight sway back position. After being appropriately draped, the patient is warned to remain in this position. The table is then raised and tipped forward for exposure and inspection.

Appropriate patient selection for this position is key. Patients with significant cardiopulmonary disease may not tolerate this position for prolonged periods as compression of the chest and abdomen may reduce ventilatory capacity and decrease preload due to compression of the IVC. Furthermore, physical factors that would prevent the patient from lying prone, such as obesity, pregnancy, and tense ascites, may require the use of a different position. Additionally, patients with orthopedic conditions that limit their range of motion including significant osteoarthritis, kyphosis, or have a history of lower extremity joint replacement should be placed in other positions.

3.2 Lithotomy Position

The **lithotomy position** is most frequently used for gynecologic examinations and procedures; however, it may be used for the examination of the perineum and anus. The patient is asked to lie in the supine position on the examination table, place their heels in stirrups, and then move their buttocks to the edge of the table. The advantages of this position include the ability to perform anorectal, pelvic, abdominal, and bimanual examinations. For additional exposure, the table may be placed in slight trendelenburg position. While this position provides some technical advantages for the surgeon, the patient (especially if the patient is male) may feel uncomfortable in this position and appropriate coaching may be necessary. The posterior position of the anus limits the surgeon technically in performing some procedures, including endoscopy.

3.3 Left Lateral Position

In the **left lateral decubitus position,** the patient lays left side down with the buttocks brought to the edge of the table or even a slight bit over the edge of the table. The back is slightly flexed and both arms are extended with the hips and knees flexed. A variation of this is the **Sim position** where in the patient is similarly positioned with the exception that the left leg is kept straight. This is the most comfortable and well-tolerated position for the patient. Patients who cannot tolerate prone jackknife or knee to chest positions due to significant cardiopulmonary disease are best suited for this position. However, access to the perineum and anus is less optimal. Adequate exposure requires retraction of the buttocks to examine and perform interventions on the perineum and anus. This can be facilitated by the aid of an assistant.

3.4 Knee-Chest Position

The **knee-chest position** is accomplished by placing the patient in the prone position, then having them bring their knees to their chest with their arms extended forward. Like prone jack-knife, this position provides excellent exposure to the anus and perineum. This position maybe favorable for pregnant patients as their gravid abdomen may prevent them from comfortably laying in the prone position. However, the cardiopulmonary limitations associated with the prone jackknife position also apply to the knee to chest position.

4 Offices-Based Endoscopy

4.1 Anoscopy

Anoscopy offers the best means to detect pathology of the anal canal. Anoscopy allows the examiner to visually inspect the terminal 10 cm of the

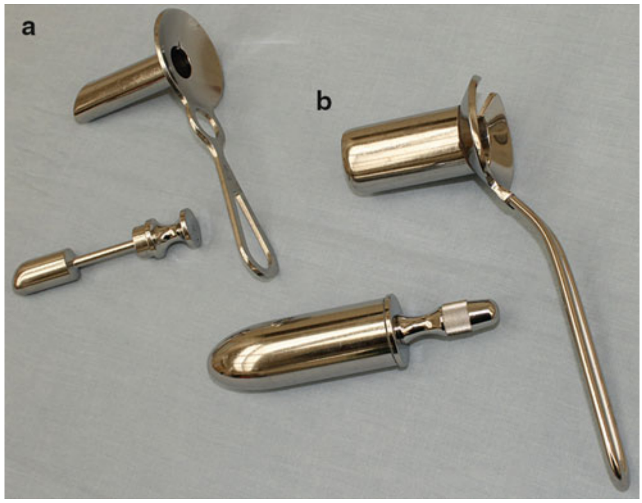

Fig. 14.2 Anoscopes. (**a**) Kelly Anoscope (*left*), (**b**) Chelsea-Eaton Anoscope (*right*)

gastrointestinal tract (Fig. 14.2). Additionally, it is essential in the performance of procedures to treat conditions of the anal canal. Numerous anoscopes and specula are available. Anoscopes are either reusable or disposable with some having a **light source** that fits into the instrument. The use of a fiberoptic light source is optional as a headlamp or a simple gooseneck lamp works well. Once the patient is adequately positioned, a digital rectal exam is performed. The anoscope is lubricated and the instrument is introduced with the obturator in place. When rotating the anoscope around the anal canal circumference, it is helpful to reinsert the obturator to turn the instrument. By doing so, the tendency to drag or pinch the anal canal or perianal skin is minimized. Finally, when pathologic features are noted or treated, the site should be recorded as follows: right anterior, left lateral, and right posterior.

4.2 Rigid Proctosigmoidoscopy

The **rigid sigmoidoscope** is one of the most valuable diagnostic instruments available in the office setting. The rigid sigmoidoscope is the optimal instrument for evaluation of the rectum. Examination with the sigmoidoscope may reveal a mucosal excrescence, a polypoid lesion, cancer, inflammation, stricture, vascular malformation, or anatomic distortion from an extraluminal mass. Indications for its use include localization of sources of bleeding, including polyps and rectal cancer as well as the evaluation of proctitis.

Reusable or disposable rigid sigmoidoscopes are available with or without fiberoptics. Reusable instruments require care and cleansing, with the need for sterilization equipment. Disposable ones are discarded and are treated as **medical waste**. The decision to utilize disposable versus reusable **proctoscopes** is dependent upon physician preference and the cost-benefit of having a number of instruments readily available for which the expense of maintenance and cleansing must be justified. These instruments are available in several diameters, ranging from 1.1 to 2.7 cm. In addition to the speculum tube, the instrumentation includes a light source, a proximal magnifying lens, and an attachment for the insufflation of air (Fig. 14.3).

Prior to the examination, a small-volume enema may be used unless otherwise contraindicated. Suction should be available to remove any residual liquid stool or fluid. Regardless vigorous catharsis, the day before the examination and dietary restrictions are unnecessary. Once the patient is positioned, a digital rectal examination precedes instrumentation. In addition to providing valuable information, this procedure permits the sphincter to relax sufficiently to accept an instrument. A well-lubricated rigid proctosigmoidoscope is then inserted and passed

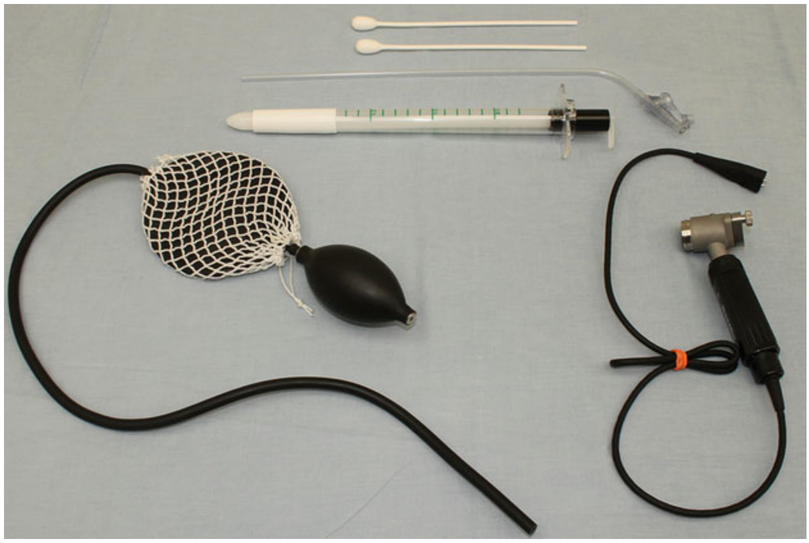

Fig. 14.3 Rigid proctosigmoidoscope with light handle, insufflator, suction tip, cotton tip applicators

to the maximal height as quickly as possible while causing minimal discomfort to the patient. Successful insertion of the **proctosigmoidoscope** requires familiarity with the anatomy of the rectum and sigmoid colon. When the proctosigmoidoscope is inserted, the low rectal and mid-rectal areas are midline structures. As the upper rectum is reached, the bowel bends slightly to the left.

Minimal air insufflation is used to visualize the mucosa while the instrument is carefully withdrawn viewing the entire circumference of the bowel wall and flattening mucosal folds to be certain that no lesions are missed. The **valves of Houston** are rectal folds on the lateral aspect of the rectal wall. The upper and lower folds are located on ipsilateral walls while the middle fold is located on the contralateral wall. The valves can serve as useful sites for performing rectal biopsy when the mucosa is grossly normal because of technical ease as well as the limited risk for perforation.

4.3 Flexible Sigmoidoscopy

Flexible sigmoidoscopy inspects more bowel surface area than is possible with the rigid proctosigmoidoscope. The flexible sigmoidoscope evaluates the terminal 60 cm of the colon and rectum as well as the anus during retroflexion of the scope in the rectal canal (Fig. 14.4a). The flexible fiberoptic sigmoidoscope is available through several companies and though the specifications of the instruments vary somewhat among the manufacturers the channel size ranges between 2.6 and 3.8 mm, the instrument diameter varies from 12.2 to 14.0 mm, and lengths range from 60 to 71 cm. The working channel allows the passage of biopsy forceps, cytology brushes, snares, and electrocautery as well as a number of other specialized instruments (Fig. 14.4b). Additionally, the working channel permits suction and irrigation. The tip of the instrument is deflected by rotation of the larger dial in each direction, while the smaller dial deflects the tip from side to side. If both dials are turned maximally, it produces a tight bend producing **retroflexion** of the tip of the instrument (Fig. 14.4c).

Prior to flexible sigmoidoscopy, bowel preparation with small volume enemas assists with the clearance of the majority the stool burden within the distal colon and rectum. Dietary restrictions and oral laxatives are generally unnecessary. In some cases, however, **oral bowel prep preparations** may be beneficial. Once the patient is placed in the left lateral (or Sims' position), a digital rectal exam is performed and the instrument is inserted. The endoscopist maneuvers the dials with one hand and guides the instrument with

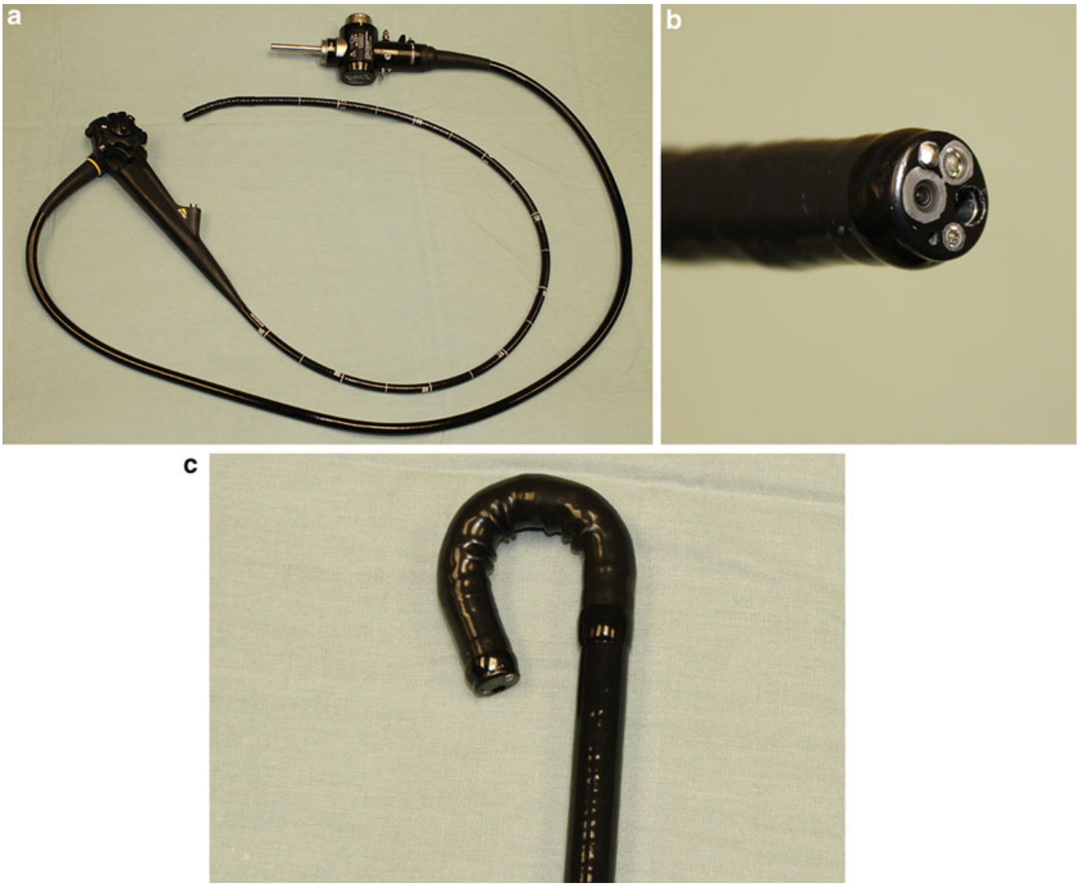

Fig. 14.4 (**a**) Flexible sigmoidoscope. (**b**) Tip of the endoscope with working channel, air port, camera lens, and fiberoptic light. (**c**) Sigmoidoscope with retroflexed tip

the other. While insufflating air rather than redirecting the tip, the examiner passes the instrument to a depth of 10 or 12 cm. This will permit the visualization of the rectal ampulla. The instrument is then passed with the lumen seen either under direct visualization or with the mucosa seen sliding past. If further passage is impeded, the instrument is withdrawn slightly, the lumen is searched out by dial manipulation and rotation, and the instrument is advanced again. Negotiation of the sigmoid colon is the most difficult part of the procedure. Sedation may be required to accomplish this, but this may not be available for office examinations. After the instrument has been passed to its full length or as far as is possible, it is carefully and slowly withdrawn. Suction, irrigation, and air insufflation are alternately employed as indicated to obtain clear visualization of the entire mucosa. Biopsy, with or without **electrocoagulation**, is obtained if appropriate. The scope is withdrawn to the distal rectal canal and retroflexed. The tip of the sigmoidoscope is then placed in the neutral position and the instrument is removed.

Flexible sigmoidoscopy is a great tool for in office evaluation, diagnosis, and management of colorectal disease; however, there are some disadvantages to this examination, including cost in the form of capital expense, maintenance, and repairs as well as the risk of complications including the transmission of communicable disease, perforation, and hemorrhage. The cost of equipment may exceed $15,000, including light source and accessories (Fig. 14.5). In addition to the outlay

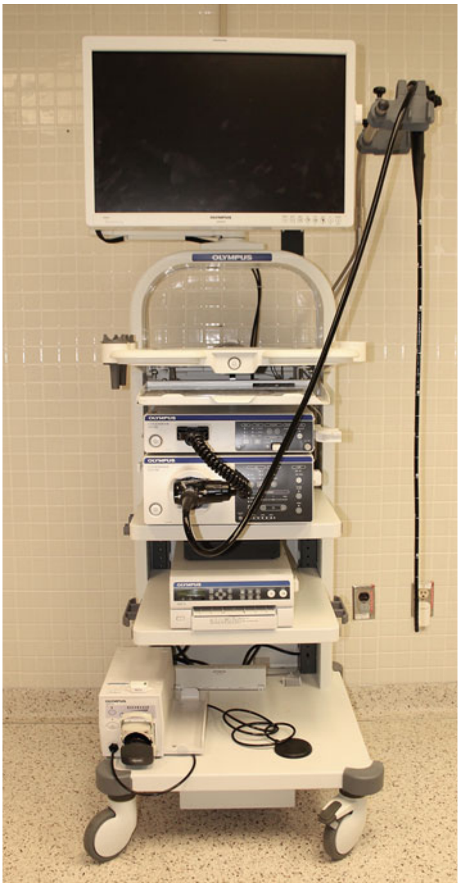

Fig. 14.5 Flexible endoscope with light source, video processor, printer, and endoscopic flushing pump

for the capital expense and repairs, there are the costs of personnel (patient preparation as well as instrument cleansing and maintenance). This procedure comes at a considerable higher cost to the patient, ranging from 25 % more than for rigid proctosigmoidoscopy to as much as 200 % more.

5 Specialized Instrumentation

The diagnosis and treatment of anorectal disease could not be completed without the aid of essential specialized equipment. These tools of the trade facilitate the crucial work of the colorectal surgeon in the ambulatory setting. While not exhaustive the following instruments are essential to the office practice of colorectal surgery.

Fig. 14.6 Anal retractor. (**a**) Hill-Ferguson retractor (*left*). (**b**) Pratt bivalve retractor (*right*)

Anal retractors and **specula** are designed to allow inspection of the anal canal and lower rectum. Standard and self-retaining specula are valuable for different uses and allow surgical procedures to be carried out by granting adequate exposure of the area. Anal retractors should always be well lubricated prior to insertion and used with care to avoid damage to the anal sphincter.

The ***Hill-Ferguson retractor***, also known as a half-moon retractor, allows inspection of half the circumference of the anal canal without applying tension to the area. It is available in different sizes allowing for serial dilation of the anal canal (Fig. 14.6a). An assistant can hold the retractor in place while procedures are performed using this instrument.

The ***Pratt bivalve retractor*** is a self-retaining anal retractor that helps expand and expose the anal canal. The speculum has rounded blades that open by squeezing the handles together and is held open by a screw adjacent to the hinge (Fig. 14.6b). This device enables the surgeon to proceed with interventions that require both hands

Fig. 14.7 Anal Retractors. (**a**) Sims retractor (large and medium). (**b**) Sawyer rectal retractor

to operate within the anal canal with minimal aid from an assistant. However, care must be demonstrated when expanding the speculum to prevent severe pain. This can be done by slowly expanding the blades sequentially until adequate exposure can be achieved.

The ***Sawyer rectal retractor*** is a hand-held retractor with a right angle convex blade that extends to a hollow grip handle (Fig. 14.7). The curved blade at the end of the handle holds back the edges of the anus for better access to the lower rectum for inspection and interventions.

In patients with fistulous disease, the passage of a **probe** can be attempted once adequate exposure is obtained. The internal or external opening can be probed to confirm the tract's location and course. Care must be taken to never force a probe down a tract to prevent the creation of false passages.

The ***Buie fistula probe*** is ideal for examination of anal or anorectal fistulas. The probe has a flat, widened handle at one end for better grip and control with a thin rigid tapered probe (Fig. 14.8).

Lacrimal duct probes are sided instruments typically with a flat metal surface at its central point to enhance handling. This device comes in numerous sizes and is malleable. Thinner probes are less durable but easily fit in the orifices of small fistula openings (Fig. 14.8).

Fig. 14.8 (From *left* to *right*) Buie Fistula probe. Lacrimal duct probe. Retrograde fistula probe. Large and small curette

The treatment of symptomatic grades I, II, and III internal hemorrhoids **by *rubber band ligation*** is a common office procedure. Rubber band ligation is commonly performed with either a suction or forceps ligator. A suction ligator draws the hemorrhoid in to the ligator with subsequent placement of the preloaded band around the hemorrhoid tissue. A forceps ligator operates in a similar fashion to a suction ligator with the exception that an atraumatic clamp is used to pull the hemorrhoidal tissue into the ligator (Figs. 14.8 and 14.9).

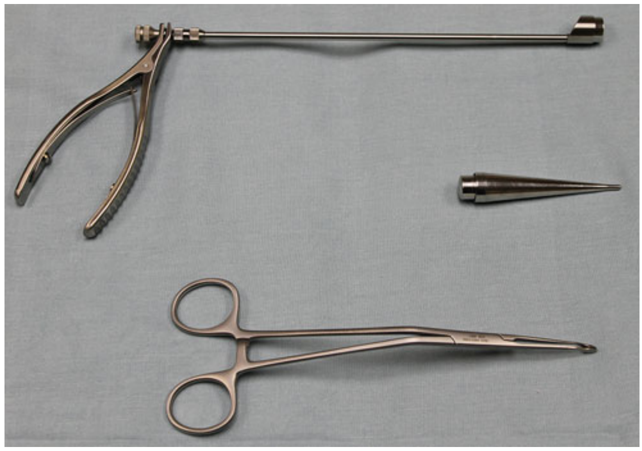

Fig. 14.9 Forceps and rubber band ligator

The removal of condyloma accuminata and other skin lesions can be facilitated by cryotherapy and electrodessication. Once local anesthesia is achieved cryotherapy and electrodessication can be used to effectively treat small lesions. **Cryotherapy** is performed by applying a cotton swab that has been dipped in liquid nitrogen to the lesions or via a device that sprays liquid nitrogen in a controlled fashion on the lesions. Cryotherapy can be an effective method of treatment, but adequate ventilation and maintenance of compressed liquid nitrogen systems are required. During electrodessication and curettage of skin lesions, the affected areas are treated with electrocautery followed by debridement with a curette. Maintenance of electrosurgical units (Fig. 14.10) in the office with disposable or reusable curettes is cost efficient and requires infrequent maintenance. However, the smoke plume generated by electrodissecation is potentially hazardous. Several studies have demonstrated that these **smoke plumes** may contain toxic gas and vapors (e.g., benzene, hydrogen cyanide, formaldehyde) as well as particulate matter including viable bacteria and viruses. This particulate matter has been implicated in transmitting disease, and it is recommended that smoke evacuation practices be implemented and as well as the use of high filtration surgical masks.

Fig. 14.10 Electrosurgical unit with electrocautery pad and grounding pad

Another role for **electrocoagulation** in the outpatient setting can be the treatment of bleeding telangiectasias due to radiation proctitis. There are a variety of endoscopic coagulation devices that are effective for control of bleeding. The technique of most endoscopists is coagulation of the focal bleeding telangiectasias rather than the entire friable mucosa of the rectum. Several treatment sessions are often required. Scarring and re-epithelization with more normal tissue tend to occur over time.

Suction is a necessity in the ambulatory setting while performing in office procedures. Suction can be utilized from a central source or via a portable unit. Single use suction tubing, canisters, and suctions tips (Yankauer) can be stored and disposed of in the office. The usefulness of adequate suction cannot be underestimated, especially during in office endoscopy.

6 Anal Manometry and Anorectal Ultrasonography

Manometry and ultrasonography of the anus and rectum complement physical examination and endoscopy by confirming diagnosis. **Manometry** is used to quantify the function of the internal and external sphincters. While ultrasonography provides excellent anatomic detail of the anal sphincter complex and the rectal wall, allowing for accurate delineation of sphincter defects, anal fistula tracts, and depth of penetration of rectal cancers.

Preparation of manometer and ultrasonography is limited to administration of a small volume rectal enema prior to the procedure to remove all stool and mucous from the rectum. The patient is then placed in the left lateral position (or Sims' position) as previously described. A digital rectal exam is then performed.

In the case of anal manometry small, flexible catheter, about the size of a thermometer, with a balloon at the end is inserted into the rectum. The catheter is connected to a **transducer** that measures the pressure. The patient is asked to squeeze, relax, and push at various times. A perfused-tube catheter is pulled through the **anal sphincter** in 1 cm increments. The anal sphincter muscle pressures are measured during each of these maneuvers at various locations. To squeeze, the patient tightens the sphincter muscles as if trying to prevent defecation. To push or bear down, the patient strains down as if trying to have a bowel movement.

Voluntary contraction of the external sphincter produces an increase in anal pressure, superimposed on the basal tone. This increase in pressure is maximal in the distal part of the anal canal, where the bulk of the external sphincter is situated. To determine the functional activity of the different parts of the external sphincter, the recording device has to be withdrawn stepwise. After each step, the patient is asked to squeeze at full strength. In this way, it is possible to measure the maximal squeeze anal pressure at every level of the anal canal. It has been shown that maximal squeeze anal pressure is higher in male than in female subjects and that it is reduced as subjects get older.

The internal sphincter reflex in response to rectal distention can be mimicked by inflation of a rectal balloon. Transient inflation of a balloon with relatively small volume of air results in an initial rise in pressure, caused by a transient contraction of the external sphincter. Almost immediately after this initial increase in pressure, a transient reduction in anal canal pressure can be observed as a result of relaxation of the internal sphincter.

Different techniques have been used, including fluid-filled open-tipped catheters, closed multiple balloon systems, and, more recently, microtransducers with readings registered on a recording device. Each method has advantages and disadvantages, and each method has its advocates, but their goals are similar.

In **endoanal ultrasonography** a 2D ultrasound scanner with a 7 or 10 MHz rotating endoprobe is used providing a 360° axial view of the anal canal. After the probe is inserted into the anal canal up to approximately 6 cm it is gently withdrawn down the anal canal, during which cross-sectional images of the puborectalis muscle, the longitudinal muscle, external anal sphincter, internal anal sphincter, and the anal epithelium are obtained.

Endoanal ultrasound is the diagnostic test of choice for the evaluation of the anal sphincter anatomy and the identification of sphincter defects associated with fecal incontinence. It has particular value in the diagnosis of complex perianal fistulas. Furthermore, endoanal ultrasound is used in the staging and follow-up of anal neoplasms.

The equipment used is the same as the equipment used for **endorectal ultrasound**, with a minor modification. A translucent plastic cap is placed over the transducer and is filled with water, which provides the acoustic medium. Prior to endorectal ultrasound examination rigid proctoscopy follows, using an instrument measuring 20 mm in diameter to accommodate the endorectal ultrasound probe. Following proctoscopy, a balloon is placed over the crystal of the endorectal ultrasound and the probe inserted into the rectum either under direct vision or through the lumen of the proctoscope. Once inserted, the balloon is inflated with water. Imaging of the rectum is initiated as the probe is withdrawn. During the exam, the probe is kept centered within the rectal lumen to maintain image clarity.

While anal manometry is a valuable diagnostic tool the main criticisms of its use are that the equipment is costly and the procedure has poor sensitivity and specificity in anorectal disorders. At times, abnormal measurements do not correlate with disease entities or explain the symptoms. Normal range of various parameters measured is highly variable and poorly reproducible. Clinical outcome after intervention does not correlate with alteration in the measurements obtained. However, anorectal manometry provides many useful data regarding anorectal function. It does provide information that assists in the management of conditions such as constipation, anismus, Hirschsprung's disease, and fecal incontinence. Anorectal manometry indicates the prognosis of treatment, particularly in the management of sphincter injuries, and may be used in biofeedback treatment of anismus and solitary rectal ulcer syndrome.

Anorectal ultrasound is an effective diagnostic tool for detailing anatomic details especially in evaluating rectal cancer. The procedure is easy to perform, well-tolerated, and readily usable in the clinic environment. After the initial investment in the specialized probes, ultrasound machine, and accessories, maintenance costs are low. Although it is operator-dependent, with a steep learning curve, the dedicated practitioner can master anorectal ultrasound readily. In addition, to tumor staging, anorectal ultrasound is useful in evaluating adenomas for foci of malignancy, assessing tumor response to neoadjuvant therapy, and in post-treatment surveillance. The ongoing development of ultrasound guided biopsies, microprobes, and **3D ultrasonography** offers the potential for further improvement in staging of lymph nodes and poorly accessible tumors, as well as prediction of response to therapy.

7 Cleaning and Maintenance

Creation and maintenance of a safe environment for patient care in the ambulatory care setting through the application of infection control principles and practices for cleaning, disinfection, and sterilization of medical equipment as well as safe disposal of medical waste is essential. Microorganisms are present in great numbers in moist, organic environments, and some can persist under dry conditions on medical equipment. Contaminated surfaces have been associated with the transmission of bacterial and viral infections, including Clostridium difficile, Hepatitis B, Hepatitis C, human immunodeficiency virus, norovirus, Salmonella species, Pseudomonas aeruginosa, and methicillin-resistant Staphylococcus aureus.

The transfer of a microorganism from an environmental surface to a patient can occur via direct contact with medical equipment and devices that are inadequately cleaned. In order to minimize the impact of this transfer, cleaning and disinfecting environmental surfaces is fundamental in reducing their potential contribution to the incidence of healthcare acquired infections.

Sterilization describes a process that destroys or eliminates all forms of microbial life and is carried out in health-care facilities by physical or chemical methods. Steam under pressure, dry heat, ethylene oxide gas, hydrogen peroxide gas plasma, and liquid chemicals are the principal sterilizing agents used in health-care facilities.

Disinfection describes a process that eliminates many or all pathogenic microorganisms, except bacterial spores, on inanimate objects. In health-care settings, objects usually are disinfected by liquid chemicals or wet pasteurization. Factors that affect the efficacy of both disinfection and sterilization include prior cleaning of the object; organic and inorganic load present; type and level of microbial contamination; concentration of and exposure time to the germicide; physical nature of the object; presence of biofilms; temperature and pH of the disinfection process; and in some cases, relative humidity of the sterilization process.

Cleaning is the removal of visible soil from objects and surfaces and normally is accomplished manually or mechanically using water with detergents or enzymatic products. Thorough cleaning is essential before high-level disinfection and sterilization because inorganic and organic materials that remain on the surfaces of instruments interfere with the effectiveness of these processes.

Decontamination removes pathogenic microorganisms from objects so they are safe to handle, use, or discard.

7.1 Disinfecting Endoscopes

Endoscopes represent a valuable diagnostic and therapeutic tool in modern medicine and the incidence of infection associated with their use reportedly is very low (about 1 in 1.8 million procedures), however, more healthcare–associated outbreaks have been linked to contaminated endoscopes than to any other medical device. To prevent the spread of **health care–associated infections**, all reusable endoscopes must be properly cleaned and subjected to high-level disinfection after each use. High-level disinfection can be expected to destroy all microorganisms, although when high numbers of bacterial spores are present, a few spores might survive. Because of the types of body cavities they enter, flexible endoscopes acquire high levels of microbial contamination during each use.

Flexible endoscopes are particularly difficult to disinfect and easy to damage because of their intricate design and delicate materials. Meticulous cleaning must precede any sterilization or high-level disinfection of these instruments. In general, endoscope disinfection or sterilization with a liquid chemical sterilant involves five steps after testing to ensure structural integrity:

1. **Clean**: mechanically clean internal and external surfaces, including brushing internal channels and flushing each internal channel with water and a detergent or enzymatic cleaners (leak testing is recommended for endoscopes before immersion).
2. **Disinfect**: immerse endoscope in high-level disinfectant (or chemical sterilant) and perfuse (eliminates air pockets and ensures contact of the germicide with the internal channels) disinfectant into all accessible channels, such as the suction/biopsy channel and air/water channel and expose for a time recommended for specific products.
3. **Rinse**: rinse the endoscope and all channels with sterile water or filtered water. If the disinfectant chemicals are not thoroughly rinsed away these chemicals can result in chemical colitis. Several case studies have demonstrated that chemical colitis can be induced as a result of contamination of endoscopes by disinfecting solutions containing glutaraldehyde or hydrogen peroxide.
4. **Dry**: rinse the insertion tube and inner channels with alcohol, and dry with forced air after disinfection and before storage.
5. **Storage**: store the endoscope in a way that prevents recontamination and promotes drying (hung vertically). Drying the endoscope is essential to greatly reduce the chance of recontamination of the endoscope by microorganisms that can be present in the rinse water.

8 Medical Waste

Governmental and local guidelines and regulations specify the categories of **medical waste** that are subject to regulation and outline the requirements associated with treatment and disposal. In addition to complying with regulation, the most practical approach to medical waste management is to identify wastes that represent a sufficient potential risk of causing infection during handling and disposal and for which some precautions are necessary. Regulated waste includes the following:

- Liquid or semiliquid blood or other potentially infectious materials
- Contaminated items that would release blood or infections materials in a liquid or semiliquid state, if compressed
- Items that are caked with dried blood or infections materials and are capable of releasing these materials during handling
- Contaminated sharp instruments (e.g., needles, scalpel blades)
- Pathologic and microbiologic wastes containing blood or infectious waste

Medical wastes require careful disposal and containment before collection and consolidation for treatment. Regulatory agencies have dictated initial measures for discarding regulated medical-waste items. Medical wastes should be kept in labeled, leak proof, puncture-resistant containers under conditions that minimize or prevent foul odors. The storage area should be well-ventilated and be inaccessible to pests. Any facility that generates regulated medical wastes should have a regulated medical waste management plan to ensure health and environmental safety regulations are adhered to per governmental regulations.

9 Cross-References

► Anorectal Manometry
► Colonic and Rectal Endoscopy
► Colonic and Rectal Endosonography
► Integration of Diagnostics in Proctology: Assessment, Choice of Treatment, and Evaluation of Results
► Physiology of the Rectum and Anus
► Ultrasound of the Colon and Rectum: Procedures and Indications

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Authorship The authors listed contributed to this manuscript through the conception, design, acquisition of data, analysis and interpretation of data as well as drafting the article and revising it critically for important intellectual content. Clyde N. Ellis, MD, gave final approval of the version to be published.

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Colonic and Rectal Endoscopy

15

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Contents

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Abstract

Colonoscopy represents the main procedure for diagnosis and treatment of many colorectal disorders. Thanks also to the development of other endoscopic devices, using modern "high-definition" endoscopes in association with a correct technique, we can today offer patients a "high-quality" diagnostic and operative colonoscopy. In this chapter, we describe the basic components of modern endoscopes, actual indications and contraindications of colonoscopy, management of antithrombotic drugs, the different types of bowel preparation, principles of sedation, quality indicators and technique of colonoscopy, colonoscopy findings, and, finally, principles of operative colonoscopy. We also include indications for procedures alternative to colonoscopy, such as capsule colonoscopy and CT colonography.

1 Introduction

The first colonoscopy was probably performed in 1969 by W. Wolf and H. Shinya at Beth Israel Hospital, in New York, and was first described 2 years later. After some decades of evolution, this examination has today reached a high-quality level. Colonoscopy, by direct internal visualization of the entire colon and the distal part of the ileum, is the *gold standard* for diagnosis and treatment of many colorectal disorders. Today, the technical improvements and the

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advancements in bowel preparation and sedation make this examination generally well tolerated, safe, and accurate.

2 Basic Components of Colonoscope

Modern colonoscopes contain the experience gained over the years and the adaptation of the instrument to the human anatomical features with the needs of endoscopists. Modern colonoscopes concentrate all technology in a tube that is 133–166 cm in length and 9.5–12.8 mm in diameter (Fig. 15.1). Pediatric colonoscope can be useful also in adults in case of fixed, angulating sigmoid colon (especially in case of previous pelvic surgery) or in presence of stricture.

From the structural point of view, the colonoscope is composed – from the outside inwards – of an outer cover of polymer which coats a stainless steel wire mesh, then of two metal spiral bands. This envelops the functional components of the endoscope which are represented by: four rods or angulation wires, the spiral metal wire of the variable stiffness system, two fiber-optic bundles for light transmission, the operative channel, a channel for air insufflation/lens cleaning, the auxiliary water-jet channel, and the electrical connection to the CCD (charge-coupled device).

The images collected by the CCD are processed by the central CPU and sent to the HD monitor. In recent years, the main manufacturers of endoscopes have introduced the so-called enhanced endoscopy or electronic chromoendoscopy that gives a greater contrast of microvascular structures through the use of a special optical filter that exploit the characteristics of the absorption spectrum of hemoglobin (NBI by Olympus) or by a computer-based processing (i-Scan by Pentax and FICE by Fujifilm), enabling in both cases a better definition of the identified lesions (Ginsberg et al. 2012).

Fig. 15.1 Standard colonoscope 12.8 mm. Pediatric colonoscope 11.8 mm

3 Indications and Contraindications for Colonoscopy

Following ASGE 2012 guidelines (ASGE Standards of practice committee 2012), colonoscopy is generally indicated in the following circumstances:

Indications for colonoscopy	Indications not for colonoscopy	Contraindications for colonoscopy
Evaluation of an abnormality on barium enema or other imaging study that is likely to be clinically significant, such as a filling defect and stricture	Chronic, stable, irritable bowel syndrome or chronic abdominal pain; there are unusual exceptions in which colonoscopy may be done once to rule out disease, especially if symptoms are unresponsive to therapy	Fulminant colitis
Evaluation of unexplained GI bleeding: Hematochezia Melena after an upper GI source has been excluded Presence of fecal occult blood	Acute diarrhea	Documented acute diverticulitis
Unexplained iron-deficiency anemia	Metastatic adenocarcinoma of unknown primary site in the absence of colonic signs or symptoms when it will not influence management	

(continued)

Indications for colonoscopy	Indications not for colonoscopy	Contraindications for colonoscopy
Screening and surveillance for colonic neoplasia: Screening of asymptomatic, average-risk patients for colonic neoplasia Examination to evaluate the entire colon for synchronous cancer or neoplastic polyps in a patient with treatable cancer or neoplastic polyp Colonoscopy to remove synchronous neoplastic lesions at or around the time of curative resection of cancer followed by colonoscopy at 1 year and, if normal, then 3 years and, if normal, then 5 years thereafter to detect metachronous cancer Surveillance of patients with neoplastic polyps Surveillance of patients with a significant family history of colorectal neoplasia	Routine follow-up of inflammatory bowel disease (except for cancer surveillance in chronic ulcerative colitis and Crohn's colitis)	
For dysplasia and cancer surveillance in select patients with long-standing ulcerative or Crohn's colitis	GI bleeding or melena with a demonstrated upper GI source	
Clinically significant diarrhea of unexplained origin		
Intraoperative identification of a lesion not apparent at surgery (e.g., polypectomy site, location of a bleeding site)		
Treatment of bleeding from lesions such as vascular malformation, ulceration, neoplasia, and polypectomy site		
As an adjunct to minimally invasive surgery for the treatment of diseases of the colon and rectum		
Management or evaluation of operative complications (e.g., dilation of anastomotic strictures)		
Foreign body removal		
Excision or ablation of lesions		
Decompression of acute megacolon or sigmoid volvulus		
Balloon dilation of stenotic lesions (e.g., anastomotic strictures)		
Palliative treatment of stenosing or bleeding neoplasms (e.g., laser, electrocoagulation, stenting)		
Marking a neoplasm for localization		

(*continued*)

4 Informed Consent and Risk Assessment

Informed consent must be obtained from the patient before performing the procedure. It must be a clear discussion about risks, benefits, and alternative to the procedure. Common risks of colonoscopy such as perforation, bleeding, infections, sedation-related adverse events, missed lesions, and intravenous site adverse events must be discussed. If an operative procedure is planned, the patient must be informed about the specific risks related to the scheduled procedure.

5 Management of Patients Under Anticoagulant and/or Antiplatelet Agents Therapy

The management of patients under therapy with anticoagulant and/or antiplatelet agents before and after an endoscopic operative procedure must be carefully evaluated. In order to decide whether to continue or discontinue these therapies it's critical to stratify the risk continue or discontinue these therapies is fundamental the stratification of the risk of bleeding of the endoscopic procedure scheduled and the risk of a thromboembolic event related to interruption of these drugs in accord with the patient's consultant (e.g., cardiologist/neurologists) or treating physician.

The anticoagulants include: warfarin, heparin, low molecular weight heparins (LMWH), and, more recently, the new direct-acting oral anticoagulants (DOAC).

The antiplatelet agents (APA) are aspirin, nonsteroidal anti-inflammatory drugs (NSAIDs), thienopyridines (e.g., clopidogrel, prasugrel, and ticlopidine), and inhibitors of the receptor GPIIb/IIIa.

Recommendations of ASGE/ESGE (Boustière et al. 2011; ASGE Standards of Practice Committee 2009) are summarized in Table 15.1.

6 Bowel Preparation for Colonoscopy

Bowel preparation is crucial for the outcome of colonoscopy and for a high-quality examination, improving adenoma detection rate and reducing costs, lowering the rate of rescheduled examinations due to inadequate preparation (Harewood et al. 2003; Froehlich et al. 2005; Rex et al. 2002b).

Some endoscopists prescribe a low-fiber diet during the 3 or 5 days preceding colonoscopy, but

Table 15.1 Management of patients on anticoagulant and/or antiplatelet drugs undergoing endoscopies procedures

		Risk of thromboembolic event	
		Low-risk condition	High-risk condition
Procedure risk for bleeding	**Low-risk procedures** Diagnostic colonoscopy ± biopsies Colonic polypectomy < 1 cm Digestive stenting	Maintain APA therapy Maintain aspirin Discontinue thienopyridines Discontinue warfarin	Maintain dual APA therapy Maintain warfarin or considering bridging therapy with LMWH
	High-risk procedures Colonic polypectomy ≥1 cm EMR ESD	Stop aspirin 5–7 days In patients taking a thienopyridine alone, it is recommended to substitute with aspirin Discontinue warfarin and considering bridging therapy with LMWH	Maintain aspirin Discuss temporary cessation of: Clopidogrel, 5–7 days Prasugrel, 7–10 days Or consider postponing procedure to time when thromboembolic risk is low Discontinue warfarin and considering bridging therapy with LMWH

APA antiplatelet agents
EMR endoscopic mucosal resection
ESD endoscopic submucosal dissection
LMWH low molecular weight heparins

the benefits of this diet have not been well assessed. Based on literature data, a low-fiber diet the day before the examination seems to be enough. In order to reduce the presence of bubbles and foam, which are frequently encountered during colonoscopy (32 %–57 % of patients), the addition of simethicone to the bowel preparation is useful, which reduces the surface tension of air bubbles and improves endoscopic view (Hassan et al. 2013; Tongprasert et al. 2009; Shaver et al. 1988).

Actually the three widely accepted bowel preparations for colonoscopy are:

– Polyethylene glycol (PEG)-based solutions: this is the first osmotically balanced solution; it is an inert, nonfermentable, nonabsorbable polymer, which does not induce fluid or electrolyte absorption or secretion. For this reason, it is safe and could be used in patients with comorbidities as liver, heart, or kidney failure. It is associated with a good cleansing efficacy, but approximately 19 % of patients are unable to complete the preparation because of its large volume (4 L) and unpalatable taste. A "low-dose" 2 L PEG (MoviPrep, Norgine in Europe and Australia, Salix Pharmaceuticals in the USA) was recently developed. This reduced volume solution has the same safety, almost the same efficacy, and is better tolerated than "high-dose" PEG (Jansen et al. 2011; Ell et al. 2008).
– Sodium phosphate-based solutions: this is a smaller volume preparation, which acts by exerting an hyperosmotic effect and by stimulating stretch receptors to increase peristalsis. This preparation has been shown to be equally effective and better tolerated than PEG. Sodium phosphate preparations must be used carefully because they have the potential to cause electrolyte disturbances including serious hyperphosphatemia and hypocalcemia. For this reason, this preparation is not recommended in elderly, in patients with heart, renal, and liver failure and other electrolyte imbalances. Also, this preparation should be avoided in patients with possible inflammatory bowel disease because it could cause colonic inflammation and aphthous ulcerations in 25 % of cases, which is less common in patients using PEG (2–3 %). ESGE suggest that oral sodium phosphate can only be advised in selected cases of specific needs that cannot be met by an alternative product (Hassan et al. 2013).

Sulfate-based solutions: magnesium is a well-known traditional laxative which increases water in the gastrointestinal tract and stimulates peristalsis. A combination of magnesium sulfate and sodium picosulfate (Picoprep or Citrafleet) was compared with PEG and sodium phosphate-based solutions in a meta-analysis showing that PEG provided a satisfactory colon cleansing in a similar proportion of patients, with less adverse events such as nausea, vomiting, or abdominal pain, but sodium phosphate produced better cleansing than magnesium sulfate and sodium picosulfate (Tan and Tjandra 2006). An adequate oral intake of water during preparation is essential. Given the potential for dehydration and dangerous hypermagnesemia, this combination is relatively contraindicated in the presence of congestive cardiac failure and impaired renal function.

Other nonabsorbable sugars like mannitol, sorbitol, and lactulose are ineffective and must be avoided because they may be metabolized by colonic bacteria carrying the risk of colonic explosion during procedures requiring electrosurgery.

6.1 Timing of Bowel Preparation

Many RCT have shown that timing of bowel preparation is very important in order to obtain a good cleansing efficacy. "Split" dose of cathartic half the day before and half the day of the examination, or a same-day regimen (for afternoon colonoscopy), improves significantly the effect of colonic cleansing. "Split" regimen is better tolerated against the single dose. Finally, for better results, the preparation should end 6–4 h before starting the examination (Marmo et al. 2010; Parra-Blanco et al. 2006; Varughese et al. 2010; Matro et al. 2010; Longcroft-Wheaton and

Bhandari 2012; Eun et al 2011; Siddiqui et al. 2009).

ASGE/ACG recommend that examination should be considered adequate if it allows detection of polyps >5 mm in size (Rex et al. 2002b). Quality of bowel preparation must be documented in each colonoscopy report. Validated preparation scores, such as Boston (Calderwood and Jacobson 2010) or Ottawa bowel preparation (Rostom and Jolicoeur 2004) score, should be used.

7 Sedation in Colonoscopy

Sedation and analgesia are usually recommended in order to improve examination quality, reduce procedure time, and minimize discomfort during colonoscopy. The reasons to use sedation and analgesia during colonoscopy are the possible pain caused by the procedure and patient's anxiety. The level of sedation can be generally considered on a spectrum from no sedation to general anesthesia. The choice of the level of sedation depends on patient comorbidities, complexity of the procedure, discomfort expected or experienced, and availability or unavailability of the anesthesiologist. All patients that undergo colonoscopy must be under continuous monitoring before, during, and after sedation with pulse oximetry and for high-risk patients with hemodynamic measurements and continuous electrocardiogram.

The level of sedation most frequently used is conscious sedation that could be reached using benzodiazepines and centrally acting narcotic opioids, such as meperidine or fentanyl. This sedation aims at maintaining the patient's collaboration for all the time requested to complete the procedure with drug-induced depression of consciousness, during which the patient responds purposefully to verbal commands, either alone or accompanied by light tactile stimulation. Usually the patient breathes spontaneously, without assisted ventilation. Oversedation, hypoventilation, aspiration, and airway obstruction are the most commonly reported sedation-related complications (Freeman 1994; Benjamin 1996). Elderly patients and subjects with compromised renal and hepatic functions may be at a greater risk of adverse events. One advantage of the use of conscious sedation with benzodiazepine and opioids is the possibility, in case of oversedation or in the presence of adverse event, to use narcotics and benzodiazepines antagonists such as naloxone and flumazenil. Conscious sedation does not require the presence of an anesthesiologist, but the endoscopist must be well trained in the management of complications of sedative drugs. After colonoscopy patients must be monitored in a recovery room for 30–60 min and can return to normal activities on the next day.

Some endoscopists perform unsedated colonoscopy in order to reduce the risk of sedation-related complications and costs, but the pain associated with colonoscopy can affect the quality of the examination and the patient's compliance. It has been demonstrated that for unsedated colonoscopy, the experience of the endoscopist is fundamental. Recently, to reduce pain of scope insertion caused by stretching of intestinal wall, carbon dioxide (CO_2) insufflation, water immersion (WI), and water exchange (WE) techniques have been proposed.

- CO_2 insufflation is readily absorbed causing less wall tension.
- Water-assisted colonoscopy is performed using water infusion and aspiration during insertion of the instrument (WE) or during withdrawal (WI).

Many studies on colonoscopy insertion demonstrated that WI and WE techniques reduce insertion pain when compared to air or CO_2 insufflation. The least painful technique was WE with increased completion of unsedated colonoscopy (Hsieh et al. 2014; Amato et al. 2013; Bretthauer 2010; Garborg et al. 2015).

For painful or prolonged procedures or for patients intolerant to colonoscopy under conscious sedation, deep sedation based on the use of propofol, a hypnotic drug, is commonly used. Deep sedation is defined as a drug-induced depression of consciousness, during which the patient cannot be easily aroused but responds purposefully to repeated or painful stimulations. Patient's spontaneous ventilation is usually maintained, but sometimes ventilation assistance may be required. Deep sedation with propofol during colonoscopy is associated with improved patient's and physician's satisfaction, because of its rapid onset and action (induces sedation within 30–60 s), its amnesic properties, and its short context-sensitive half-life of 2–8 min. For these reasons, it is considered an attractive drug for colonoscopy. Propofol can be administrated as monotherapy or in a combination with opioids and benzodiazepine. When used in combination, total dose of propofol can be reduced. Propofol may cause respiratory depression, apnea, or hypotension. Moreover, in 14 % of patients tremors, twitches, hypertonus, and hiccups can occur. Pulmonary edema, hypertension, cardiac arrhythmias, bronchospasm, or laryngospasm happens more rarely. For this reason, monitoring and managing the airway and breathing is more critical and requires the presence of anesthesiologists with additional costs.

8 Quality Indicators for Colonoscopy

Quality indicators for colonoscopy (Rex et al. 2015) are summarized in Table 15.2.

9 Technique of Colonoscopy

For a high-quality colonoscopy, it is essential to refer to the correct principles of technique. This is essential not only to improve the technical quality of the examination but also to eliminate or minimize visceral pain during the exam. Basic rules of colonoscopy involve proper anterograde push of colonoscope, precise movements of the tip, constant identification of the bowel lumen, appropriate twist on the instrument's longitudinal axis, and controlled suction.

The discomfort caused by colonoscopy is mostly dependent by the stretching of the mesenteric supports of the colon and, to a lesser extent, by the distension due to the air blown in the colon. Elder patients are more tolerant of the curves and twists caused by the colonoscope because the elasticity of the mesentery rises with age, while women, due to the lower abdominal area in which the colon and the angle are located that the sigmoid presents between the uterus and left inguinal area, are generally subject to more frequent traction and straightening of the mesentery, resulting in increased tenderness.

Despite the considerable individual variability, definitely the presence of loops causes pain. That is why this examination, according to a correct technique based on frequent retraction movements, aims to keep the endoscope as

Table 15.2 Quality indicators for colonoscopy

Pre-procedure	Intra-procedure	Post-procedure
Appropriate indications	Correct administration of sedation	Incidence of perforation for all examination (<1:500)
Informed consent and risk assessment	Cecal intubation identifying cecal landmarks (appendiceal orifice and ileocecal valve with photo-documentation) and a detailed mucosal inspection	Incidence of perforation for screening examination (<1:1000)
Correct management of antithrombotic drugs	Quality of bowel preparation documentation	Incidence of postpolypectomy bleeding (<1 %). The risk of bleeding increases with polyp size; for polyps >2 cm of diameter, bleeding rates may exceed 10 %
Sedation plan	Frequency of adenoma detection rate (ADR), that is, the primary measure of the quality of mucosal inspection and the single most important quality measure in colonoscopy	Appropriate recommendation for timing of repeat colonoscopy after histologic findings is reviewed
Timeline of procedure	Withdrawal time >6 min is demonstrated in several studies that increase ADR	

straight as possible, reducing the probability of marked tenderness.

Colonoscopy may be conducted by only one endoscopist that holds the instrument with his right hand, while his left hand handles the controllers on the chassis, or the doctor who commands with both hands grips the endoscope and the assistant (usually the nurse) that pushes and rotates the instrument. Most experts believe that the first option is preferable because it allows you to have a greater sensitivity about the thrust force applied, and it allows you to correct any loops as well as greater readiness to overcome difficult stretches as stenosis, particularly curves angled or rigid segments with greater caution. Only in certain moments, which require mandatory concurrent use of both knobs, the instrument is held by the assistant.

Most endoscopists use the left lateral decubitus position as a start position for the colonoscopy, although some prefer the supine position and very few the right side. A careful inspection of the perianal region and a digital exploration of the rectum should precede the introduction of the colonoscope. This procedure allows a lubrication of the anal canal, the evaluation of the sphincter tone, and the exclusion of pathological conditions such as the presence of fistulous orifices, leakage of secretions or blood, presence of palpable stenosis, etc. The colonoscope is then inserted through the anus gently holding it at about 10 cm from the tip.

Once distended the distal rectum by inflation and aspirated residues, a retroflection maneuver should be performed carefully to inspect the rear portion of the mucosa immediately above the dentate line as small lesions at this level cannot be detected in front view only. The left lateral decubitus position is more favorable to give a great exposure of the distal rectum. The rear view is performed by angling to the maximum the tip of the colonoscope while it is introduced and displacing it gently sliding along one of the side walls so turning the optics toward the internal anal orifice. This maneuver can easily lead to traumatic injuries of the mucosa and should be done with caution. If the operator encounters difficulties during the retroversion of the instrument, it is good practice to desist in order to avoid complications and observe carefully the distal rectum by front view only.

Once the rectum is explored, the progression through the sigmoid colon, the descending colon, and the splenic flexure is probably the most delicate moment of the procedure and is usually being associated with the formation of loops and onset of pain. The endoscope is advanced using a minimum thrust associated with slight movements of clockwise and counterclockwise torque impressed to the instrument by endoscopist's right hand. This avoids having to use too often the right/left knob and makes the insertion more fluid. The direction of the intestinal lumen can be identified thanks to some details such as folds convergence, presence of a less illuminated or "shadow" area on the visual field, the observation of transverse haustral folds and small translucent interhaustral lines, or the identification of a single longitudinal fold introflexed in the lumen formed by one of the three taeniae coli. For each curve, it is necessary to exactly locate the direction of the lumen and preferentially rotate the instrument positioning the tip in the upper part of the visual field (12 h). The tip must be angulated and slightly anticipating the thrust movement. If the lens is found in direct contact with the mucosa, it is essential to retract the instrument until the lumen is seen again, identifying the right lumen direction.

Some sigmoid corners are angled to the point that their overcoming must be performed "blindly." In these cases, the endoscopist must be extremely confident on the lumen direction and should push the endoscope using gradually the maximum angulation of the tip. During this maneuver, small translations from the axis of the curve must be corrected by locating mentally the lumen direction and consequently turning the instrument. After having passed a good part of the curve, a full rotation of the right/left knob can be associated to increase the angle of the tip and to facilitate the transition.

To aspirate residual fluids, it is convenient to place them at 6 o'clock position and aspirate while blowing air, thus preventing complete collapse of the bowel wall and unwilling suctions of the mucosa.

Sigmoid can be thought as a very elastic tube that follows a curve in posterior–anterior direction and when filled with air becomes longer (about 40–70 cm) and angled, while when it is not inflated, it is much shorter (30–35 cm) and straight. To pass the sigmoid is generally convenient to progress with the instrument blowing the minimum amount of air required and retracting often the scope after each curve, making the bowel straight as possible. In this way the accentuation of curves and angles as well as the longitudinal stretching of the organ itself can be prevented. A compression made by the assistant with the hand flat on the suprapubic region/left iliac fossa may help to reduce the anterior loop forming near the abdominal wall.

However, despite the use of a correct technique, often the passage of the sigmoid and the reaching of the descending colon still cause the formation of various types of loops. It is important to underline that, despite the colonoscope seems to proceed along the bowels in a straight way and without obstacles or angles, it is possible that a loop is developing. Without the aid of fluoroscopy or magnetic systems (ScopeGuide, Olympus), it is not easy to detect the presence and type of loops that develop during the examination. The presence of a loop is suspected in the case in which the pushing movements or retraction of the endoscope is not simultaneously transmitted to the tip of the endoscope or even causes the opposite movement. In such cases, many endoscopists prefer to still proceed by pushing the colonoscope, counteracting the extension of the loop by manual compression. This maneuver can cause the patient considerable tenderness, reduces the usable length of the colonoscope by about 40–50 cm, and causes a considerable instability of the distal tip. The use of deep sedation probably could lead to favor the spread of this approach.

The original technique of colonoscopy, instead, suggests that once the presence of a loop is detected, the scope should be advanced up to the proximal descending colon or the splenic flexure. This point is well recognized as a landmark for the view of the distal transverse lumen, which has a triangular shape and is clear of fluids when the patient lies on his left side. By angling the scope tip and maintaining the bowel lumen at the center of the visual field, the scope should be firstly rotated in a clockwise direction, reaching even to a complete rotation of 360°. If it were possible to retract the endoscope of 30–40 cm during this rotation while maintaining the distal tip in the same position, most likely we will have reduced an N loop or an alpha loop. In case the clockwise rotation increases the resistance of the instrument without reducing the loop, we will probably be in the presence of an inverse alpha loop (“reverse alpha loop”), which will require a maneuver of counterclockwise rotation.

The colonoscope is usually straight when the tip, located at the splenic flexure, is at about 50 cm from the anal verge. Then, to enter the transverse colon is often sufficient to reduce the tip angulation, and gently push the instrument while turning it slightly clockwise. If the scope does not advance, probably a sigmoid loop is reforming. If a variable stiffness scope is used, it will be useful at this point to stiffen it up; otherwise, a manual compression on the right iliac fossa/hypogastrium toward the left iliac fossa can be performed by the assistant. If this maneuver has no effect, a decubitus change can be done, by placing the patient supine or on his right side.

After the splenic flexure is reached and overtaken, the distal transverse colon generally runs smoothly. The presence of a more or less accentuated mid transverse ptosis is usual: this may appear as one of the two flexures. Retracting a little the scope after air aspiration and with a slight counterclockwise instrument twist may help lifting the transverse and more easily achieving the hepatic flexure. In case of difficulty, an upward compression on the mesogastrium to lift the transverse can be made by the assistant, or the decubitus can be changed to supine position.

The hepatic flexure often forms an acute angle, even of 180°. When the right direction to overcome it is found, it may be useful to aspirate air, so reducing the angulation. Other useful maneuvers are to place the patient in the supine or left lateral position, to ask him to inhale deeply, or to make compression on the right hypochondrium, downward and medially, with the intent to “lower” the flexure.

After the ascending colon is reached, the cecum can easily be intubated with the patient in the supine position, aspirating air and simultaneously retracting the scope. A frequent problem regarding the ascending colon is to see the cecum in the distance but being unable to reach it. At times, this maneuver can prove extremely difficult for the formation of loops at the level of sigmoid and/or transverse. In this case a manual compression on the rear right side ("Waye point") or upwards in the left iliac fossa or even in the right iliac fossa can sometimes solve the problem.

A total colonoscopy requires a clear recognition of the cecum through the identification of the convergence of the three taeniae coli, of the appendiceal orifice, and of the ileocecal valve.

To explore the distal ileum, the ileocecal valve has to be passed. To do so, the scope tip must be retracted 2–4 cm from the cecal wall. Imagining the appendiceal orifice shape as a bow, an imaginary arrow in the appendix shows the ileocecal valve entering direction. With this trick ("bow and arrow sign"), the ileocecal valve can be passed even blindly, retracting smoothly the scope tip from the bottom cecal wall.

The retraction phase of the colonoscope must be performed slowly (at least 6 min) to allow a proper examination of the mucosa, alternating short inward movements to recover any involuntary slip off of the instrument. Each colonic segment is explored thoroughly, removing residues adhering to the wall with water flushing (preferably with peristaltic pumps or through the accessory channel) and aspiration or changing the patient's decubitus to move fluids. It must be remembered that, for the gravity, the hepatic flexure opens in left lateral decubitus position, while the vision of the transverse colon, the splenic flexure, and the descending colon is simplified by the patient in the supine or right lateral.

Once completed, the examination of a colonic segment is good practice to draw air to prevent excessive distension remaining.

A correct technique of colonoscopy grants many advantages, such as less pain and higher rate of cecal intubation. Moreover, it is very important when operative endoscopic techniques are needed.

10 Alternatives to Colonoscopy (Capsule Colonoscopy and CT Colonography)

10.1 Video Capsule Endoscopy (VCE) of the Colon

Capsule endoscopy represents a major development of minimally invasive examination of GI tract. First experimental use in animals was made in 2000 (Iddan et al. 2000). Next, the first reported use in humans was described in 2001 (Appleyard et al. 2000; Appleyard et al. 2001). The colon capsule is 11 mm in diameter and 32 mm long (Fig. 15.2). It was mainly used for observation of small bowel in particular condition such as obscure gastrointestinal bleeding, unexplained iron-deficiency anemia, nonstricturing small bowel Crohn's disease, refractory or complicated celiac disease, hereditary polyposis syndromes, and small bowel tumors (Ladas et al. 2010).

Colon VCE has limited indications and is generally used as a complement to incomplete colonoscopy and where conventional colonoscopy is either refused by patients or poses substantial risk to them. According to the meta-analysis (Rokkas et al. 2010), the sensitivity and specificity of colon VCE for the detection of significant colon adenomas (Fig. 15.3) and carcinomas are 69 % and 86 %, respectively, suggesting that although it is a promising tool, colon VCE needs improvement before it can be an alternative to colonoscopy for colon cancer screening.

Fig. 15.2 Colon capsule

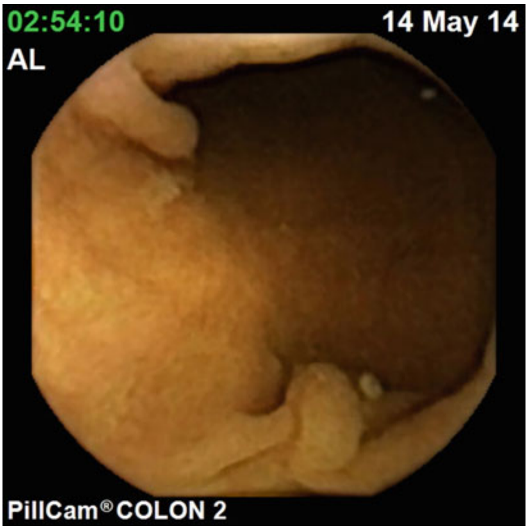


Fig. 15.3 Colonic small pedunculated polyp

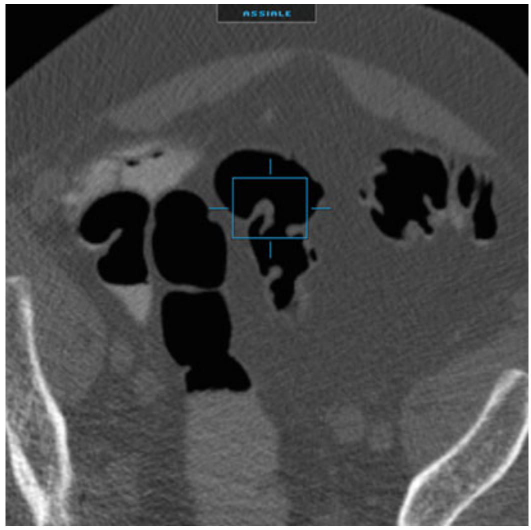


Fig. 15.4 CTC polyp detected

The PillCam colon capsule (Given Imaging) has some technical differences from the small bowel capsule: it is approximately 6 mm longer; it has dual cameras that enable the device to acquire video images from both ends, optics with more than twice the coverage area of the small bowel capsule; automatic light control; and a frame rate of four frames per second. After initial capsule activation and 5 min of image transmission, the capsule enters a delay mode of approximately 2 h, after which it spontaneously "wakes up" and restarts the transmission of images for approximately 10 h. The recommended bowel preparation consists of conventional colonoscopy preparation plus ingestion of domperidone before capsule ingestion and boosts of sodium phosphate purge and bisacodyl suppositories during the examination (Eliakim et al. 2006; Schoofs et al. 2006).

Previous studies showed that the colon capsule is expelled within 10 h post ingestion from 74 % to more than 90 % of patients (Spada et al. 2014). No examination-related adverse events have been reported.

10.2 CT Colonography (CTC)

CTC is an imaging technique, first introduced 20 years ago and evolved becoming now the best radiologic diagnostic test for detecting colorectal cancer (CRC) and polyps. As colonoscopy, CTC needs preparation with 1–3 days of low-fiber diet and bowel purgation in order to perform an optimal examination. Also to improve polyp detection and reduce the number of false-positive, the "tagging" of residual stools by oral administration of water-soluble iodinated contrast or diluted barium sulfate suspension is recommended.

Figures 15.4 and 15.5 show colonography appearance of polyps.

Recently, ESGE/ESGAR guidelines of clinical indications for CTC were published (Spada et al. 2014). Main recommendations are:

– CTC is recommended as the radiological examination of choice for the diagnosis of colorectal neoplasia. ESGE/ESGAR do not recommend barium enema in this setting.
– In case of incomplete colonoscopy, CTC is recommended the same day or next day. Delay of CTC should be considered following endoscopic resection. In case of obstructing colorectal cancer, preoperative contrast-enhanced CTC may also allow location or staging of malignant lesions.
– CTC is recommended when endoscopy is contraindicated or not possible as an

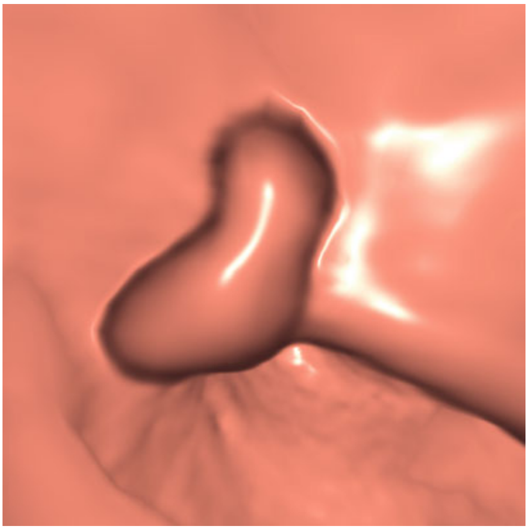


Fig. 15.5 CTC reconstruction

acceptable and equally sensitive alternative for patients with symptoms suggestive of colorectal cancer.

– A patient with at least one polyp ≥ 6 mm in diameter detected by CTC must be a referral for endoscopic polypectomy. CTC surveillance may be clinically considered if patients do not undergo polypectomy.
– CTC is not recommended as primary test for population screening or in individuals with a positive first-degree family history of colorectal cancer (CRC). However, it may be proposed as a CRC screening test on an individual basis provided the screenee is adequately informed about test characteristics, benefits, and risk.

11 Pathological Colorectal Findings

Endoscopic evaluation of the colon and rectum starts always with careful evaluation of anal and perianal region with observation and digital rectal examination in order to identify, if present, abnormalities such as the presence of rectal prolapsed, hemorrhoids, anal fissure, anal fistula, or anal cancer. This evaluation is completed after the scope insertion with the retroflexion maneuver that visualizes the circumference of the anus, the dentate line, and the distal part of the rectum. The anus is also better evaluated with rigid proctoscope. Pathologic findings of the anus, including hemorrhoids, anal fissure, anal fistula, and anal carcinoma, are described elsewhere in this book.

Fig. 15.6 Colonic diverticula

In this chapter subsection, we describe the most frequent pathologic findings during colonoscopy; it is not the purpose to cover more than the most obvious points of endoscopic pathology which are fully featured in the various available atlases of endoscopy (Fig. 15.6).

11.1 Colonic Diverticula

"Diverticula" occur when colonic mucosa and submucosa herniate through defects of muscle layer of the colon (Fig. 15.6). These could be found during colonoscopy as orifices that can be small or larger than the true lumen. The most frequent localization of diverticula is the sigmoid colon, but they could be found in all colonic segments.

Acute inflammation of diverticula, called diverticulitis, represents a contraindication to colonoscopy because infection could compromise the colonic wall with high risk of bleeding or perforation. For this reason if colonoscopy is indicated, in order to exclude other diagnoses (polyps and/or malignancy), it should be performed 6–8 weeks after the acute episode of diverticulitis has resolved (Jacobs 2007). Sometimes, during elective colonoscopy performed for other reasons, it is possible to see pus coming out from a diverticular orifice associated with inflammation of colonic mucosa in this segment that could suggest a recent episode of diverticulitis. Another risk in patients with diverticulosis is bleeding. This event represents one of the major causes of colonic bleeding (Longstreth 1997). In case of severe bleeding, the diagnostic sensitivity of colonoscopy to detect a diverticular bleeding is 72 % (Caos et al. 1986). Usually diverticular bleeding stops spontaneously without therapeutic measures in over 76 % of cases. In case of severe continuous bleeding, colonoscopy may be performed in order to identify the site of active bleeding or the presence of adherent clot or visible vessel and treat with adrenaline or endoclip. Also, in case of therapeutic failure, the endoclip could be used as marker for arterial embolization (Vallappan et al. 2015). Actually embolization of diverticular bleeding is considered a first-line therapy in case of massive lower bleeding with a successful rate in 85 % of cases (Khanna et al. 2005).

Sometimes it is possible to find, especially in the sigmoid colon, a mucosal inflammation called segmental colitis associated with diverticulosis (SCAD) with an incidence reported from 0.3 % to 2 % (Tursi et al. 2010). Endoscopic findings of SCAD are macroscopic and microscopic inflammation of the sigmoid and sometimes of descending colonic interdiverticular mucosa without involvement of the diverticular orifice. The rectum and the proximal colon are usually normal. Endoscopic and histology findings suggest that this condition includes pathogenetic characters of inflammatory bowel disease. So it is not well defined if it is an autonomous entity or a complication of diverticular disease (Tursi 2011). Based on endoscopic appearance of mucosal damage, some authors have proposed an endoscopic classification (Tursi 2011):

– "Crescentic fold disease" pattern with the presence of some inflamed red patches of 0.5–1.5 cm in diameter, without hemorrhage or ulceration; there is no involvement of diverticular orifices.
– "Mild to moderate ulcerative colitis-like" pattern, with edema, hyperemia, erosions, and reducing or loss of vascular pattern. There is no involvement of diverticular orifices.
– "Crohn's disease colitis-like" pattern with aphthous ulcers within a normal colonic mucosa. There is no involvement of diverticular orifices.
– "Severe ulcerative colitis-like" pattern with marked submucosal and mucosal edema, hyperemia, and ulceration sometimes with reduction of the lumen. The diverticular orifices are not always visible, and it is possible to find a reduction of the lumen due to the inflammation.

11.1.1 Submucosal Lesions

Submucosal lesions, which may be very difficult to diagnose, include secondary carcinoma, endometriosis, and a few large vessel hemangiomas, lipomas, or carcinoid.

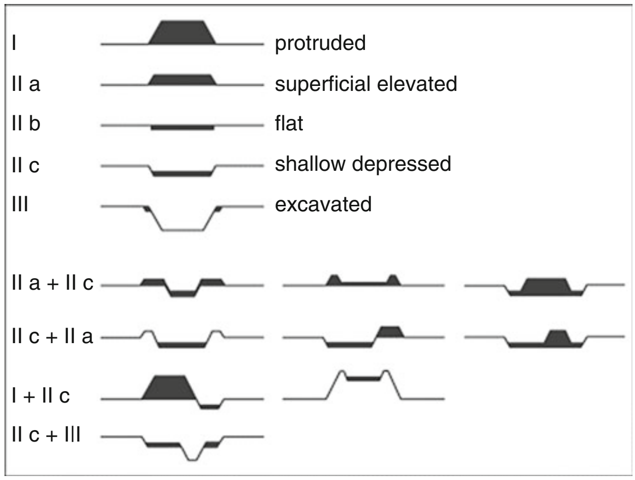
Superficial Neoplastic Lesions

The Japanese Gastric Cancer Association (JGCA) classification of neoplastic lesions, modified from Borrmann classification proposed in 1926 for gastric cancer, includes:

– Type 0: superficial polypoid, flat/depressed, or excavated tumors
– Type 1: polypoid carcinomas, usually attached on a wide base
– Type 2: ulcerated carcinomas with sharply demarcated and raised margins
– Type 3: ulcerated, infiltrating carcinomas without definite limits
– Type 4: nonulcerated, diffusely infiltrating carcinomas
– Type 5: unclassifiable advanced carcinomas

Of them, type 0 represents the "superficial" neoplasia, defined as the lesions in which endoscopic appearance suggests that the depth of penetration in the gastrointestinal wall is not more than submucosa (Fig. 15.7). Type 0 and its subtypes first used for classification for gastric lesion were applied on the esophagus and next for colorectal lesions. In 2002 in a Paris workshop, this classification was evaluated by an international group of endoscopists, surgeons, and pathologists

Fig. 15.7 Paris classification



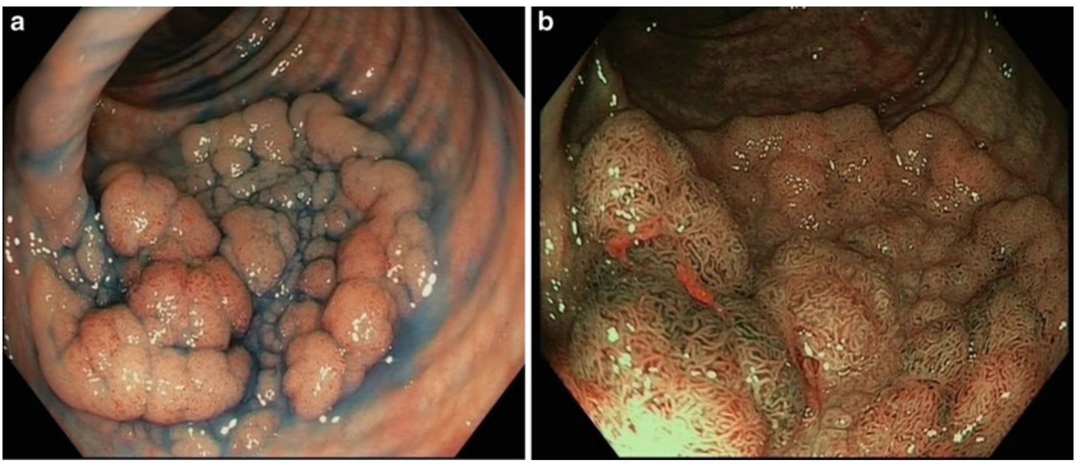


Fig. 15.8 (**a**) Lateral spreading tumor granular mixed type. (**b**) Chromoendoscopy evaluation, below NBI evaluation

in order to define its clinical relevance, assessing the risk of submucosal invasion and the risk of lymph node metastasis. The conclusion of workshop was summarized in 2003 in Osaka (Participants in the Paris Workshop 2003).

Superficial neoplastic lesions (type 0) according to Paris classification (Participants in the Paris Workshop 2003) and later update (Endoscopic Classification Review Group 2005) are divided into:

1. Polypoid
   – Protruded, sessile (0-Is)
   – Protruded, semipedunculated (0-Isp)
   – Protruded, pedunculated (0-Ip)
2. Nonpolypoid lesions
   – Superficial, elevated (0-IIa)
   – Flat (0-IIb)
   – Superficial shallow, depressed (0-IIc)
   – Excavated (0-III)

Nonpolypoid lesions have superficial and depressed morphology and were classified as elevated with depressed area (IIa-IIc) when there is more elevated area or depressed with elevated area (IIc-IIa) when there is more depressed area than the elevated (Fig. 15.8).

When nonpolypoid lesions are large (≥10 mm), they are called lateral spreading tumor

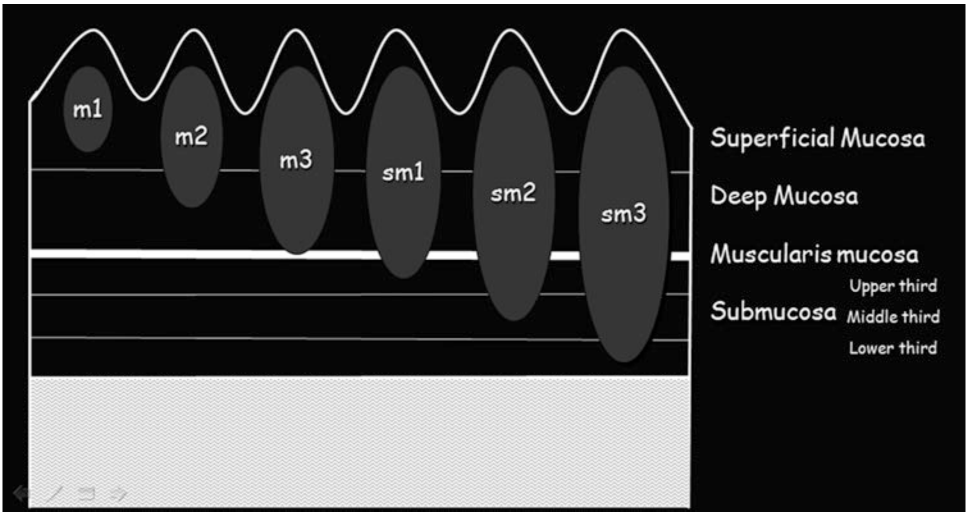


Fig. 15.9 Mucosal and submucosal invasion

(LST) (Fig. 15.8). Based on the morphology we distinguish:

1. Granular type
   – Homogeneous type
   – Mixed type
2. Nongranular type

Endoscopic morphology of type 0 neoplastic lesions according to the Paris classification supplemented with magnifying endoscopy, image-enhanced endoscopy, and lifting sign has an important predictive value for depth of invasion that correlates with the risk of nodal metastasis. This influences greatly the decision making as to whether to approach these lesions by endoscopic resection or surgery. Micrometer pathological evaluation on specimen obtained after endoscopic resection, correctly prepared, stretched, and pinned on cardboard with the mucosal surface up and placed in formalin, classified according to Vienna classification (Shlemper et al. 2000; Dixon 2002) of epithelial neoplasia, will give the definitive diagnosis on R0 resection and depth invasion of the lesion that for colorectal lesions must be <1000 μm.

Risk of lymph node metastasis for colorectal superficial lesions evaluated in a large series from Akita and Yokoyama hospitals (Endoscopic Classification Review Group 2005) reports (Fig. 15.9):

– Sm1 invasion, frequency of lymph node metastasis <1 %
– Sm2 invasion, frequency of lymph node metastasis <6 %
– Sm3 invasion, frequency of lymph node metastasis <14 %

The classification's guideline is critical in order to differentiate patients who need endoscopic resection from those who need a surgical one.

Carcinomas

Malignant polyps appear irregular, may bleed easily from surface ulceration or be paler (Fig. 15.10), and typically also firmer to palpation with the biopsy forceps. The diagnosis of large carcinoma is often obvious when it appears as friable mass with irregular surface, sometimes with depressed and ulcerated area. Multiple biopsies should always be obtained in order to minimize the risk to find adenoma and to distinguish condition that could mimic malignancy such as granulation tissue masses at an anastomosis, larger granulation tissue polyps in chronic ulcerative colitis, and, most rarely, the acute stage of an ischemic process. The size of colonic neoplastic lesion is not an indicator of malignancy. In fact sometimes large colonic lesion could be benign with area of dysplasia, and small polyps may be cancerous.

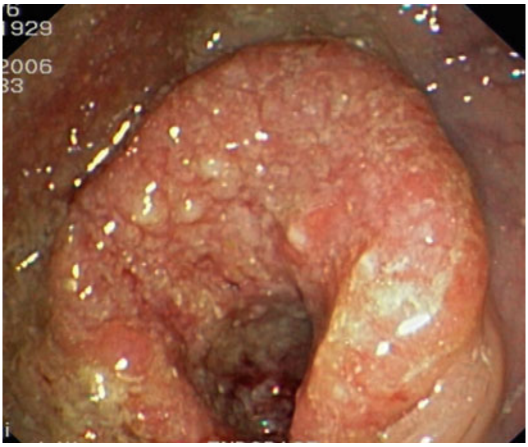


Fig. 15.10 Carcinoma of the colon

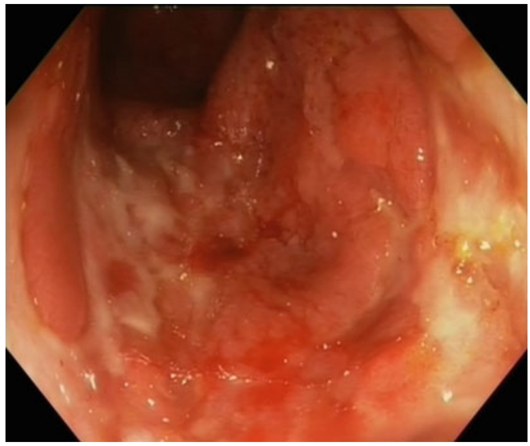


Fig. 15.11 Inflammation of terminal ileum in patient with Crohn's disease

However, diagnosis is less obvious in case small early cancers do occur, typically 6–20 mm in diameter with a slightly depressed center. The "non-lifting sign" for lesions without presence of scar (previous partial resection or biopsies) is considered as sign of deeper submucosal infiltration, but when it is used alone as predictive value of early cancer with deeper infiltration (>1 mm), it had a sensitivity of 61.5 % and specificity of 98.4 % (Kobayashi et al. 2007). Endoscopic diagnosis using magnifying colonoscopy and submucosal injection could be useful for diagnostic evaluation of invasion depth with high sensitivity and sensibility when used together. However, sensitivity and specificity of both these diagnostic modalities might depend on the expertise of the investigator.

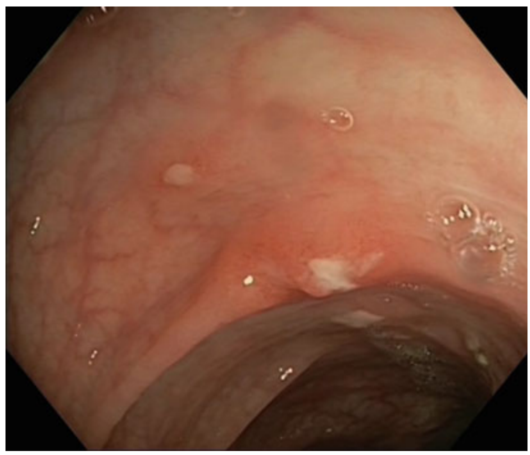


Fig. 15.12 "Aphthoid" ulcers

Inflammatory Bowel Disease (IBD)

IBD is defined as a group of disorders characterized by recurrent destructive inflammation of gastrointestinal tract due to unknown etiology. The most common forms are:

– **Crohn's disease (CD)** that could engage any segment of the gastrointestinal tract from mouth to the rectum and involve all wall layers. The most common symptoms are cramping abdominal pain, diarrhea, fever, and weight loss. Distal ileum is frequently involved (Fig. 15.11). Inflammation of the bowel is often discontinuous with erythema, erosions, sometimes with irregular and serpiginous ulceration, mucosal edema with appearance of "cobblestone," and with reducing or absence of vascular submucosal pattern. It is common with the presence of multiple, small, flat, or volcano-like "aphthoid" ulcers (Fig. 15.12) set in a normal vascular pattern characteristic but not pathognomonic.

– **Ulcerative colitis (UC)** could involve the entire colon or a colon segment or more commonly the rectum. It sometimes starts from the rectum but can spread in a proximal (oral) direction to all segments of the colon. Inflammation is circular affecting only the mucosal

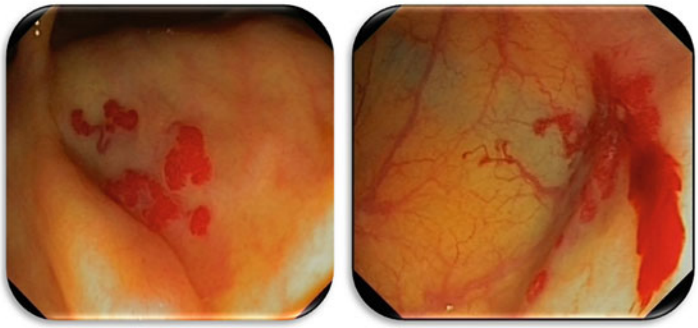


Fig. 15.13 Angiodysplasias

membrane. When inflammation affects the area of ileocecal valve that becomes open and incontinent, it is possible to note an inflammation of terminal ileum, due to the reflux of colonic contents (backwash ileitis).

Pathological findings of UC are erythema, edema, granularity of mucosa, ulceration, and reduction or absence of vascular submucosal pattern.

– Collagenous colitis, lymphocytic colitis, and atypical microscopic colitis are very uncommon. In these cases, mucosal surface could appear normal and diagnosis is made based on symptoms and histological findings of endoscopic biopsies (at least four should be taken at intervals around the colon) that must be made in any patient with chronic diarrhea.

The differentiation between UC and CD is based on endoscopic and histological findings; it is sometimes not easy. In fact it is possible that definitive diagnosis is made based on the course of the disease.

The role of colonoscopy with biopsy in case of IBD is to make diagnosis (when possible), to evaluate extension of bowel involvement and severity of mucosal activity, and to evaluate response to therapy and surveillance of cancer developing especially for long-standing colitis.

Angiodysplasias

Angiodysplasias are uncommon and mainly occur in the small intestine, in the cecum, or in the ascending colon in elderly patients. They appear as solitary or multiple (often two or three) and are always bright red, but they can be small (1–5 mm diameter) vascular plaques or spidery telangiectasias (Fig. 15.13).

Ischemic Colitis

Ischemic colitis (IC) is one of the most common causes of low gastrointestinal bleeding. It is common in elderly patients, but it can occur at any age. The true incidence of IC is not known. Any colonic segment could be affected, but it is more frequent in the left colon and the superior part of the rectum due to their blood supply. The main causes of IC are atherosclerosis, heart failure, cardiac arrhythmias, abdominal aortic surgery, shock, and hyperviscosity states. Also cocaine and some medications such as nonsteroidal anti-inflammatory drugs (NSAIDs), products of digitalis, alosetron, and migraine agents have been related to IC. If the patient has IC, colonoscopy must be performed carefully, with minimum insufflation of air (better with CO_2 if available). Typically the endoscopic appearance is from mild, patchy submucosal hemorrhages with ulcerations to necrosis of colonic wall with segmental involvement of the left colon called "watershed area" (Wang et al. 2014); biopsies could be used to differentiate IC from infectious disease, IBD. In some cases, IC is self-limiting and does not need any medical or surgical therapy, but in case of necrosis of colonic wall with peritoneal inflammation or in case of perforation colonic resection, it is needed.

Infectious Colitis

Infectious colitis may affect all colonic segments and sometimes the distal ileum. Endoscopic appearance could be mimicking CD or UC with mucosal edema and erythema, and ulcerations are difficult to distinguish. In case of infections by *Clostridium difficile*, the presence of yellow-white plaques of exudate pseudomembranes is possible, but not pathognomonic. Diagnosis of different infectious colitis could not be made based only on endoscopic findings, but it must be reached also according to symptoms, pathological evaluation on biopsy, and stool microbiological evaluation.

12 Principles of Operative Colonoscopy

Colonoscopic polypectomy can be used for removal of polypoid lesions as sessile, semipedunculated, and pedunculated polyps. Small sessile semipedunculated polyps <6 mm in diameter can be removed by biopsy forceps or cold snare. Polypectomy for polyp lesions greater than 6 mm is resected using snare with cautery coagulating and "auto-cut" current produced by the circuitry of some "intelligent" electrosurgical units, which will automatically adjust power output to match the resistance of the tissue being heated, the intention being to produce a predictable rate of transection. Polyps can be aspirated into a suction trap (polyp trap or a gauze in a suction line), which is a convenient way of managing polyps up to 5–8 mm in diameter. Larger polyp could be taken away using Roth Net basket or a snare.

12.1 Endoscopic Mucosal Resection (EMR)

EMR is widely indicated for endoscopic resection of sessile or nonpolypoid lesions that require resection at the submucosa to ensure cure. EMR is performed with previous submucosal injection (saline, saline plus indigo carmine, glycerol, etc.) that provides to lift the lesion submucosa. This procedure aims at first to evaluate the presence of lifting sign and create a "safety cushion" of engorged submucosal stroma that protects the bowel wall from heat damage. Then with a snare, the lesion could be resected. When possible the lesion must be resected "en bloc" for an appropriate pathological evaluation, but for lesions larger than 20 mm as LST, it is sometimes difficult or impossible to be resected en bloc, and then in these cases, the resection could be made by endoscopic piecemeal mucosal resection (EPMR) with the risk of incomplete resection which can lead to local recurrence (Fig. 15.14).

12.2 Endoscopic Submucosal Dissection (ESD)

Endoscopic submucosal dissection (ESD) was developed for en bloc resection of larger, flat or with slight depression lesions, usually up to 20 mm in diameter; ESD must be performed because these lesions present a higher risk of invasion of submucosal layer. The en bloc specimen with mucosa and submucosa is needed for pathological evaluation.

ESD is a complex technique that requires a high level of expertise of the operator and more time to be performed than EMR.

Actually there is no consensus about what is the optimal endoscopic resection for colorectal superficial lesions. A recent meta-analysis (Wang et al. 2014) compared the clinical outcomes of ESD vs EMR suggesting that ESD has considerable advantages over EMR for colorectal tumors regarding en bloc resection rate and local recurrence rate, without increasing complication rate, but on the other hand, ESD needs a long operation time.

Anyway EMR and ESD are considered as advanced endoscopic resection techniques that also require experience in management of complication as microperforation and immediate or delayed bleeding.

Tattooing should mark the site of any suspicious or partially removed polyp, whether for follow-up or possible surgery.

12.2.1 Postpolypectomy Syndrome

Sometimes after difficult polypectomy, especially piecemeal removal of a large lesion in the

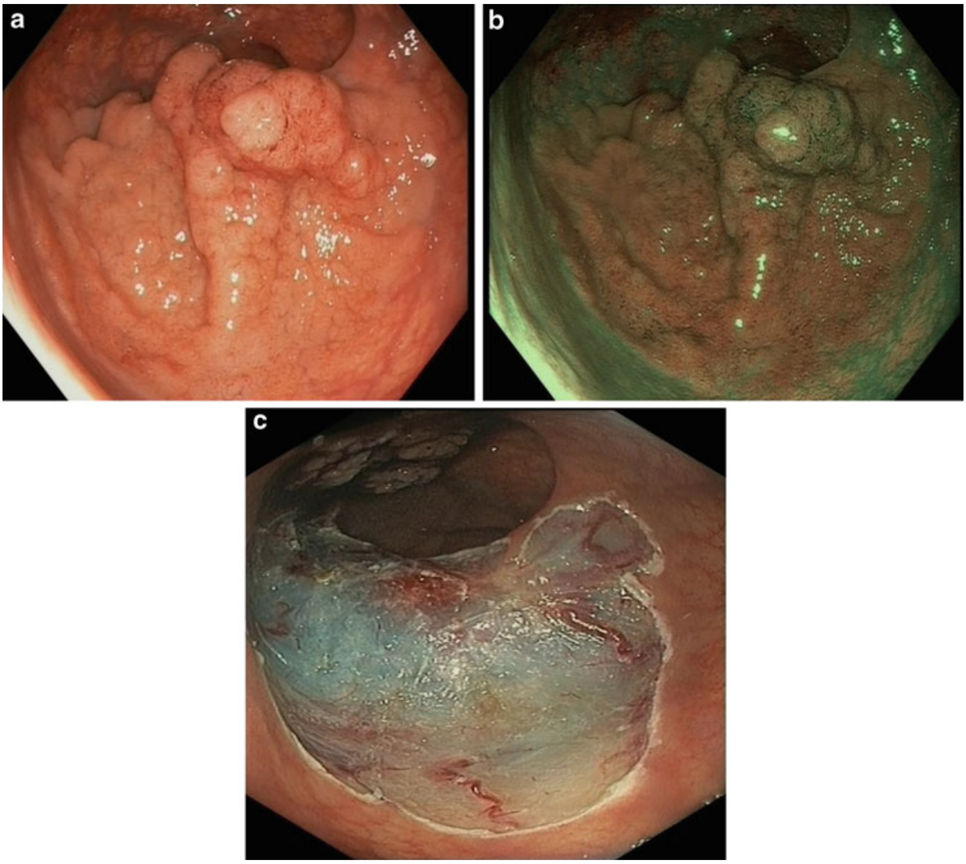


Fig. 15.14 (**a**) LST granular type. (**b**) Evaluation using NBI. (**c**) The lesion was resected with endoscopic piecemeal mucosal resection (*EPMR*)

proximal colon, it is possible that the patient presents fever, pain, and peritonism. It is called the "postpolypectomy syndrome," due to a "closed perforation" with full-thickness heat damage to the bowel wall. Usually abdominal pain and fever persist for 12–24 h following polypectomy, but without free gas on X-ray or signs of generalized peritonitis. The inflammatory reaction of the peritoneum is usually a self-limiting event. This condition is managed with bed rest and systemic antibiotics. If symptoms and signs do not decrease rapidly, the patient must be referred to surgical consultation.

12.2.2 Other Therapeutic Procedures

Balloon dilation of short benign anastomotic stricture is easy with "through-the-scope" (TTS) balloons. Considering the size of the stricture, usually the treatment starts with small balloon (perhaps dilate to 12–15 mm initially and repeat to a larger diameter on another occasion). Balloons of at least 18 mm diameter give the best long-term results. Balloons must be fluid distended, using either water or dilute contrast material, because air is too compressible. A pressure gun and manometer are used, and the maximum dilatation reached must be maintained for 2 min to dilate effectively. The overall perforation rate for stricture dilation in different series ranges between 4 % and 10 %. Very scarred, ulcerated, or angulated strictures are more likely to split under dilation.

Volvulus: colonoscope can be used to deflate a sigmoid volvulus. Deflation alone, when possible, is usually sufficient for the torsion to reverse spontaneously, so endoscopic manipulation is usually unnecessary. If the segment appears in advanced stage of ischemia, surgery is indicated because of the high risk of perforation.

Angiodysplasias in symptomatic patients can be treated endoscopically preferably using argon plasma coagulation because of its ease, efficacy, and relative safety. Electrocoagulation (mono- or bipolar), heater probe, or laser can be carefully applied; even minor whitening and edema will progress to produce remarkable local ulceration within 24 h.

12.2.3 Insertion of Self-Expanding Metal Stents

Insertion of colorectal self-expanding metal stents is used for management of malignant large colorectal obstruction. Today different colorectal stents are available, and their application depends on the site and morphology of stricture and clinical indications such as palliation or bridge to surgery (Harris et al. 2001; Stimac 2008). Stents are uncoated and their nitinol "memory metal" construction is deliberately made to be immovable. Stents are placed with a combined endoscopic–fluoroscopic-controlled procedure in order to check location and expansion of the distal end of the stent. Colonic stents are not indicated in patients in which perforation is suspected or in case of active bleeding. Also rectal stenting is contraindicated for stricture involving the distal part of the rectum (<5 cm from the anal verge) because it causes tenesmus, pain, and fecal incontinence.

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Colonic and Rectal Endosonography 16

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Contents

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1 Introduction

Accurate staging of rectal carcinoma is crucial for planning surgery and indicating adjuvant therapy. Although computed tomography and magnetic resonance imaging are very sensitive in detecting metastatic disease, the local staging of rectal cancer with these techniques has been disappointing. Endorectal ultrasound (ERUS) is today with rectal MRI the most accurate methods for staging rectal and anal cancer.

Continued research and development has made the instrumentation for ERUS more accurate and user-friendly. New techniques that have contributed significantly to the evolution of ERUS include three-dimensional ERUS, high-frequency miniprobes, and transrectal ultrasound-guided biopsy techniques. Further improvements can be expected from contrast enhancement with microbubbles and color Doppler imaging.

In this new millennium, new developments in ERUS, such as tri-dimensional ERUS and radial electronic probing, widen the role of ERUS in the staging of rectal carcinoma, as well as for inflammatory conditions.

The place of colonic EUS is less well define, main indication is the use of linear echoendoscope to reach difficult lesions for a biopsy or the use of mini-probes to access submucosal lesion or extrinsic compressions.

Over the past two decades, anorectal ultrasound has been progressively used and is now considered to be an integral part of the

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investigation of various anorectal diseases. The technique is reliable for staging rectal tumors and is widely used in the assessment and follow-up of rectal carcinoma.

2 Rectal Cancer

Colorectal cancer is among the most common cancers affecting adult men and women.

Nearly 45,000 new rectal cancers are diagnosed each year in the USA. Although part of a functional continuum, rectal cancers can be distinguished from colon cancers on the basis of some very real anatomical, prognostic, and practical differences. These differences demand staging and treatments unique to rectal lesions. Stage-based therapy for rectal cancer has achieved broad acceptance and is considered to be the standard of care.

2.1 Rectal Anatomy

The rectum originates beneath the peritoneal reflection, extending 15–20 cm from the anal verge. The rectum is contained within the narrow pelvis, is confined by the pubic bones anteriorly and the lumbosacral spine and coccyx posteriorly, and is surrounded by structures vital to urinary and sexual function. Using transrectal EUS, the urinary bladder, seminal vesicles, prostate, and urethra are well seen in the male. The urinary bladder, uterus, and vagina are less well appreciated in women.

The anatomy of the anorectum is specifically designed for the storage and controlled evacuation of the fecal bolus. Defecation and continence require the coordinated interaction of several muscular structures in and surrounding the anorectum. The circular muscle of the anus forms a prominent internal anal sphincter (IAS), which provides tonic closure of the anus. Specialized skeletal muscles descending from the levator ani apparatus provide a muscular sling and terminate to form the external anal sphincter (EAS). When viewed with a radial scanning echoendoscope, at the level of the anal verge, the IAS and EAS can be viewed as two distinct rings (Schwartz et al. 2002). The lymphatic drainage of the rectum follows the route of its venous drainage along the inferior, middle, and superior hemorrhoidal veins to the inferior mesenteric veins, along the iliac veins and on to the portal vein.

The prognosis of rectal cancer correlates with the pathological stage at the time of diagnosis. Consequently, management is predicated on tumor stage at diagnosis and response to induction therapy. A wide variety of surgical techniques have been developed for rectal neoplasms in consideration of the anatomical constraints, preservation of function, and intent to achieve a cure (Ramamoorthy and Fleshman 2002). These are associated with disparate rates of postoperative morbidity.

Cancer containing superficial villous adenomas can be cured with endoscopic mucosal resection. Lesions confined to the wall may be resected by transanal excision or low anterior resection. Lesions involving, or in close proximity to, the anus may warrant APR, preserving anal sphincter function. Patients with locoregionally advanced lesions (extension on to the perirectal fat and/or perirectal or pelvic adenopathy) should be considered for neoadjuvant chemoradiotherapy. Neoadjuvant therapy has been shown to reduce local recurrence and permit an increased likelihood of a sphincter-sparing operation, with less toxicity compared with postoperative regimes (Santiago et al. 2002). Thus, unlike more proximal colon cancer, the optimal method of management of rectal carcinoma is critically dependent on accurate preoperative staging of the disease, as shown in Table 16.1 (Ahmad et al. 2002).

2.2 Equipment and Technique

ERUS can be performed with either blind, rigid probes (Fig. 16.1) or flexible echoendoscopes. ERUS is an ambulatory procedure, and intravenous sedation is optional. Patients prepare the rectum with two Fleets enemas in advance. With the patient in the left-lateral decubitus position, a digital rectal examination should be performed. Digital rectal examination should allow an assessment of sphincter tone and palpation of the lesion. If palpable, the lesion should be described in terms of location, distance from the anal verge, and fixation or mobility. Forward-viewing sigmoidoscopy should be performed to image the lesion in both the forward and the retroflexed scope positions. This allows familiarity with the anatomical configuration of the patient's rectum and the location and distribution of the tumor.

The echoendoscope is inserted and advanced beyond the lesion, under direct vision, to the rectosigmoid junction. ERUS imaging should begin at 5–7.5-9 and 12 MHz during withdrawal of the scope. The lumen is deflated, and the water-filled balloon is adjusted for acoustic coupling. Tip deflection should be passive, allowing the transducer to find the right axis to the lumen. During this phase of the examination, the operator is looking for surrounding adenopathy. Any lymph nodes seen should be assessed for size, shape, and echo qualities. The scope is then withdrawn to the level of the anal verge.

Table 16.1 Tumor stage and lesion location on endorectal ultrasound determine the treatment options for rectal cancer

Tumor stage/ location	Treatment option
Polyploid T1m cancer	Snare polypectomy
Sessile T1m cancer	EMR or ESD TAEX
T1sm	ESD or TAEX
T2, no	LAR
T2, no/low	RT-capecitabine followed by LAR
T2, T3, N1/high	LAR
T2,T3, N1/low	RT-capecitabine followed by LAR or APR
T4, any N	RT-FOLFOX followed by LAR or APR

High R2 cm from the dentate line; low % 2 cm from the dentate line. *EMR* endoscopic mucosal resection, *ESD* endoscopic submucosal dissection, *TAEX* transanal excision, *LAR* low anterior resection, *APR* abdominoperineal resection, *FOLFOX* chemotherapy 5FU-Folinic Acid-Oxaliplatin, *RT* neoadjuvant radiotherapy, *RT-capecitabine* neoadjuvant radiochemotherapy using capecitabine

Next, the tumor itself should be targeted to determine its depth of penetration into or through the rectal wall. The choice of frequency is dependent on the lesion size, but 5 and 7.5 MHz are most commonly employed for T-staging. The degree of tip deflection and water-balloon fill should be adjusted to avoid false findings owing to tumor compression, tangential imaging, and air artifact. Filling the lumen with water through the accessory channel is often necessary to achieve optimal imaging.

The echoendoscope is advanced and withdrawn over the lesion to achieve satisfactory imaging over the length of the lesion.

Finally, the scope is withdrawn to the anal verge to assess the anal sphincters for tumor invasion. This is an active process and should incorporate voluntary squeezing and relaxation of the muscles by the patient during imaging.

2.3 ERUS Staging of Rectal Cancer

The American Joint Committee of Cancer has identified the TNM classification as the preferred

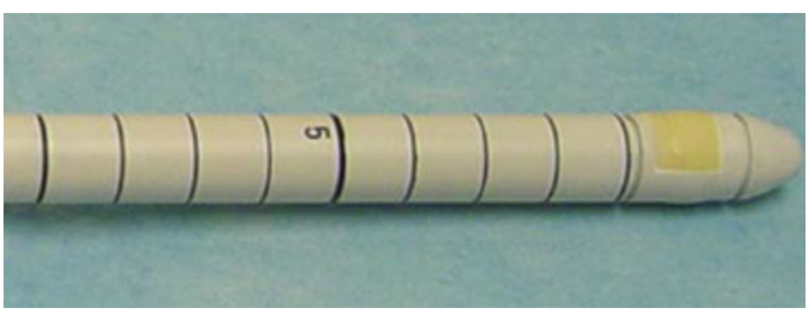


Fig. 16.1 Radial rigid ERUS Probe

staging system (American Joint Committee on Cancer 1992). This system is based on the determination of the depth of tumor invasion (T-classification), the presence of regional lymph node metastases (N classification), and the presence of distant metastases (M-classification). The individual classifications are combined to provide an overall stage.

2.3.1 ERUS Tumor Stage

Endosonographically, the rectal wall is seen as five alternating hyper- and hypoechoic layers (Fig. 16.2). The histological correlation of the echo-layers is as follows:

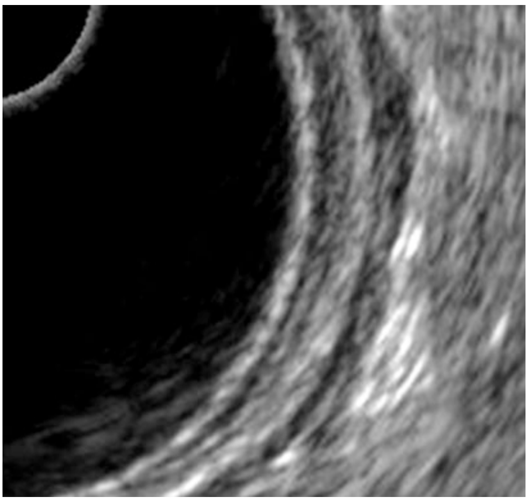


Fig. 16.2 Normal rectal wall

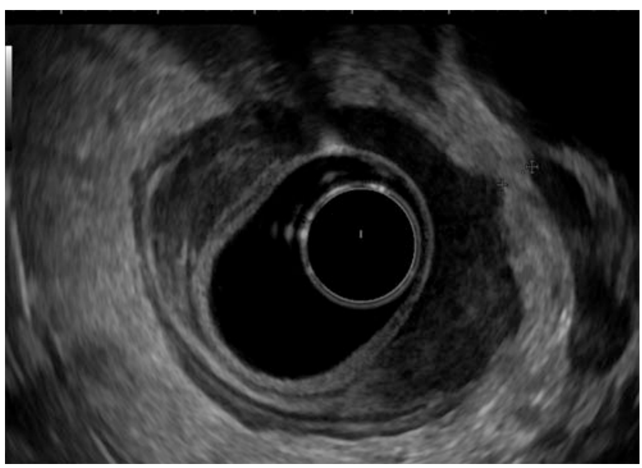
- **First layer (hyperechoic)** – interface between water or a water-filled balloon and the superficial mucosa
- **Second layer (hypoechoic)** – represents the deep mucosa and muscularis mucosa
- **Third layer (hyperechoic)** – denotes the submucosa and its interfaces
- **Fourth layer (hypoechoic)** – represents the muscularis propria
- **Fifth layer (hyperechoic)** – indicates the interface between the serosa and perirectal fat

Rectal cancer appears as homogeneous hypoechoic soft tissue, and invasion appears as disruption of the normal wall echo-layer pattern. A tumor that, on ERUS, appears to be limited to the mucosa or submucosa (first three echo-layers) is classified as a T1 lesion, whereas a tumor that invades the muscularis propria (the hypoechoic fourth ERUS layer) is a T2 lesion. A T3 lesion penetrates through the rectal wall, extending beyond the five echo-layers and into the surrounding perirectal fat (Fig. 16.3). A T4 lesion displays direct invasion into an adjacent organ such as the prostate gland, sacrum, vagina, or bladder.

2.4 ERUS Lymph Node Staging

Endosonographically, lymph nodes appear as round or oval structures (Fig. 16.4) that are hypoechoic compared with the surrounding

Fig. 16.3 US T3 rectal cancer



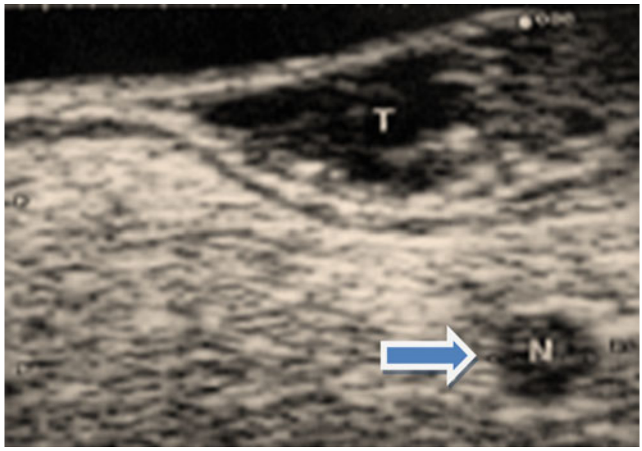


Fig. 16.4 Peri-rectal lymph node (malignant) (*arrow*)

perirectal fat. Endosonographic criteria applied to perilesional adenopathy in other regions of the digestive tract for the determination of malignancy versus benignity may not be so well applied in rectal cancer. Data obtained primarily in patients with esophageal carcinoma have identified four sonographic criteria predictive of malignancy: large size (1 cm), hypoechoic echodensity, sharply demarcated borders, and round (rather than ovoid or flat) shape (Catalano et al. 1994).

These criteria may not apply as well to rectal carcinoma in that up to 50 % of metastatic lymph nodes associated with rectal cancers are smaller than 5 mm (Spinelli et al. 1999). Although EUS-guided fine-needle aspiration (FNA) of an individual lymph node might confirm accuracy, it is only rarely called upon for this purpose in initial staging.

The accuracy of tumor and nodal staging depends on the experience and expertise of the endosonographer (Boyce et al. 1992). The overall accuracy of T-staging for rectal cancer varies between 70 % and 90 % (Marone et al. 2000; Gualdi et al. 2000; Glaser et al. 1993; Herzog et al. 1993; Cho et al. 1993; Boyce et al. 1992; Yamashita et al. 1988; Beynon 1989; Feifel et al. 1987). When ERUS is incorrect for T-stage, it is typically due to overstaging rather than understaging. ERUS tends to overstage cancers because ultrasound can detect, but not separate, inflammation adjacent to the malignancy from the tumor itself. Understaging is attributed to the undetected microscopic invasion of cancer cells beyond that observed by ERUS. Accuracy is generally lowest for lesions classified as T2 by ERUS, which may be overstaged as T3 lesions. Overstaging is apt to occur when imaging tumors located on a haustral fold, owing to artifact induced by tangential imaging. Filling the rectal vault with water will improve technical results and probably enhances T-stage accuracy.

The overall accuracy of N-staging by ERUS is 73–83 % (Marone et al. 2000; Gualdi et al. 2000; Glaser et al. 1993; Herzog et al. 1993; Cho et al. 1993; Boyce et al. 1992; Yamashita et al. 1988; Beynon 1989). This lower accuracy of nodal staging is attributed to the observation that up to 50 % of malignant nodes are less than 5 mm in diameter, and the EUS detection rates of these nodes may be as low as 20 % (Feifel et al. 1987).

Nonetheless, ERUS has been reported to be equal or superior to computed tomography or T- and N-staging. In several comparative studies, ERUS has shown a greater accuracy than computed tomography scanning for staging rectal cancer: 67–93 versus 53–86 % for T-stage, and 80–87 versus 57–72 % for N-stage (Herzog et al. 1993; Rifkin et al. 1989; Pappalardo et al. 1990). Magnetic resonance imaging (MRI) with an endorectal surface coils is similar to but not better than ERUS in terms of accuracy (Waizer et al. 1991; Thaler et al. 1994; Schaefer et al. 1996; Hunerbein et al. 2000; Meyenberger et al. 1995). MRI is more expensive than transanal ultrasound, and endorectal MRI is not widely available and easy to perform.

Although there is little published experience of ERUS FNA in rectal cancer, experience extrapolated from other malignancies (Faigel et al. 1997) has suggested that the performance of FNA cytology can markedly increase the accuracy and specificity of ERUS nodal classification. Management may be altered when nodal metastasis is identified in a patient in whom the T-classification would otherwise suggest the possibility of local endoscopic or transanal resection as a curative option. This applies to the 10 % of patients with T1 lesions who have positive lymph nodes.

2.5 Restaging After Neoadjuvant Therapy

Preoperative neoadjuvant chemoradiotherapy is commonly used to downstage rectal cancers. In addition to improving long-term survival and local recurrence, this approach allows sphincter-preserving low anterior resection in many patients who would require APR based on findings at initial presentation. Neoadjuvant therapy of rectal cancer results in tumor regression/necrosis and inflammatory and fibrotic changes in the rectal wall. These changes may be sonographically indistinguishable from viable tumor. As such, the accuracy of T- and N-staging after chemoradiation therapy is considerably compromised (Napoleon et al. 1991). Therefore, we do not apply TNM staging when inspecting lesions for their response to preoperative chemoradiotherapy. Instead, we assess for evidence of tumor regression from the surrounding organs, in particular the anal sphincters, vagina, and prostate. In this way, ERUS can direct therapy in patients who have undergone neoadjuvant therapy as a prelude to possible sphincter-sparing surgery (Loren et al. 2002).

2.6 ERUS for Local Recurrence of Colorectal Cancer

A local recurrence of rectal cancer after presumed curative resection occurs in 10–15 % of cases, usually within the first 2 years after surgery. It is hypothesized that an early detection of recurrent local tumor, prompting early retreatment, would improve survival, but although this seems logical, it remains unproven. ERUS may be useful in the detection of suspected local recurrence when no mucosal lesions are seen during surveillance sigmoidoscopy. Preliminary data obtained using blind/rigid ultrasound probes suggested that transrectal ultrasound is highly sensitive for the detection of anastomotic recurrence (Beynon et al. 1989; Feifel and Hildebrandt 1992). A more recent study using a radial scanning echoendoscope reported ERUS to be highly sensitive (90 %) in detecting local rectal tumor recurrence (Muller et al. 2000). However, the sonographic changes of local tumor recurrence are not specific as postoperative and postradiation inflammatory/fibrotic changes have a similar appearances (Hunerbein et al. 1996). ERUS should be used to complement sigmoidoscopy when local recurrence is suspected (Fig. 16.5). In these instances, extraluminal local recurrence suspected by EUS can be confirmed by ERUS-guided FNA.

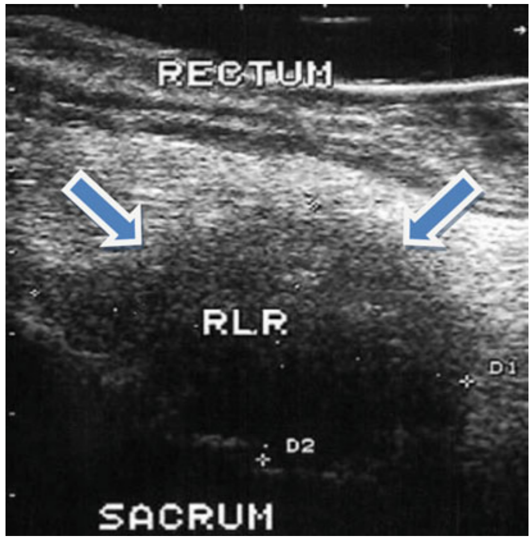


Fig. 16.5 ERUS aspect of the local recurrence of a rectal cancer (RLR) *arrows*

3 Three-Dimensional ERUS

Several studies in the field of endoscopic ultrasound (EUS) technology have reported advantages for three-dimensional (3D) EUS

(Kallimanis et al. 1995; Odegaard et al. 1999; Kanemaki et al. 1997; Hunerbein and Schlag 1997; Ivanov and Diavoc 1997; Tokiyama et al. 1999; Gold et al. 1999; Calleja and Albillos 1998; Marusch et al. 2002; Chung et al. 2000; Hünerbein et al. 1997; Hünerbein et al. 1999; Liu et al. 2000). However, most 3D EUS studies have been performed using catheter-type miniature probe systems (Kanemaki et al. 1997; Tokiyama et al. 1999; Marusch et al. 2002). Some studies have previously reported the benefits of a 3D EUS system using a linear-array echoendoscope for the 3D guidance of interventional procedures, but the scanning method employed in this system was limited, and because the ultrasound probe was not positioned at the tip of the endoscope, it was difficult to obtain clinically sufficient images without geometrical distortion in the stomach (Liu et al. 2000; Tamura et al. 2002; Sumiyama et al. 2002; Molin et al. 1999). A study has tried to resolve this problem and maximize the performance of the 3D EUS using a linear echoendoscope with a miniature electromagnetic position sensor attached to the tip of the scope, which can be used in free-hand scanning in any position (Sumiyama et al. 2003). The problem with this technique, however, is that this electromagnetic sensor increases the size of the probe.

We have today a new method of 3D EUS working without any electromagnetic sensors, which can be used even with electronic radial or linear rectal probes. Two types of system have been developed, making use of either a series of two-dimensional (2D) images produced by one-dimensional arrays, or 2D arrays to produce 3D images directly. Two criteria must be met to avoid inaccuracy: the relative position and angulation of the acquired 2D images must be known accurately; and the images must be acquired rapidly and/or gated to avoid artifacts from respiratory, cardiac, and involuntary motion.

3.1 Tracked Free-Hand Systems

The operator holds an assembly composed of the transducer and an attachment, and manipulates it over the anatomy. The 2D images are digitized as the transducer is moved while meeting two criteria: the exact relative angulation and position of the ultrasound transducer must be known for each digitized image, and the operator must ensure that no significant gaps are left when scanning the chosen area.

3.2 3D Reconstruction

The 3D reconstruction process refers to the generation of a 3D image from a digitized set of 2D images. The approach used is the voxel-based volume. The 2D images are built into a 3D voxel-based volume (3D grid) by placing each digitized 2D image into its correct location in the volume. The main advantages are that no information is lost during 3D reconstruction, and a variety of rendering techniques are possible, although large data files are generated.

3.3 Visualization of 3D Ultrasound Images

The ability to visualize information in the 3D image depends critically on the rendering technique. Three basic type of technique are used:

1. Surface-based viewing technique. An operator or algorithm identifies the boundaries of the structures to create a wire-rame representation. These are shaded and illuminated so that surfaces or structures or organs can be visualized.
2. Multiplane viewing techniques.
   – Orthogonal views: three perpendicular planes are displayed simultaneously and can be moved or rotated.
   – Polyhedron: the 3D images are presented as a multisided volume (polyhedron). The appropriate ultrasound image is "painted" on to each face of the polyhedron, which can be manipulated.
3. Volume-based rendering techniques. The 3D image is projected on to a 2D plane by casting rays through the 3D image. The voxel values intersected by each ray can be multiplied by

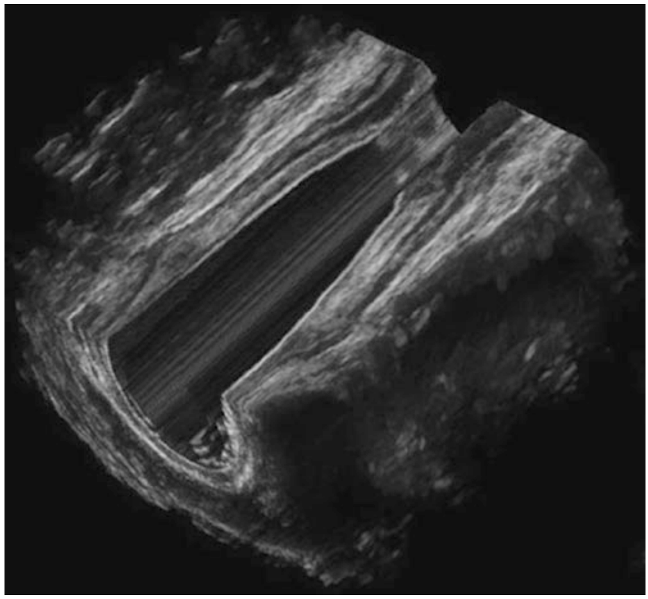


Fig. 16.6 3D reconstruction of a normal rectum

various factors and summed to produce different effects: multiplied by 1 and then added to form a radiograph-like image; multiplied by factors to produce translucency; or display of only the voxel with the maximum intensity along each ray. 3D ERUS is a new technique and is still under development (Fig. 16.6). Sumiyama et al. have recently reported their experience using 3D EUS and an electronic linear probe.

They concluded that 3D EUS using a linear-array echoendoscope was accurate and represented a consistent method. They also claimed that 3D EUS facilitated the anatomical interpretation of sonographic images and reduced the procedural difficulty of scanning. Previous experience using 3D EUS using mechanical miniprobes has been reported for cardiovascular procedures (Klingensmith et al. 2003) and, using rigid electronic probes, for gynecological tumor assessment (Ayoubi et al. 2002; Liu et al. 2002). 3D EUS using mechanical miniprobes has been described for pancreaticobiliary (Kanemaki et al. 1997; Tokiyama et al. 1999; Marusch et al. 2002) and anal disease (Gold et al. 1999). Our experience is slightly different as we use 3D ERUS with new software allowing the use of a linear-curved or radial electronic probe because the software is integrated into the computer of the ultrasound machine and does not rely on an external sensor attached to the tip of the EUS scope.

The most important question, however, relates to the usefulness of 3D EUS (Yoshimoto 1998; Yoshino et al. 1999). With regard to the locoregional staging of rectal cancer, several studies have shown important benefits in terms of better parietal staging (Hunerbein and Schlag 1997; Ivanov and Diavoc 1997) with the possibility of accurate staging even with stenotic lesions, and the more precise performance of an EUS-guided biopsy (Hunerbein et al. 1996). Our results showed that the mesorectal margins are defined better using 3D ERUS than 2D ERUS, allowing more accurate parietal staging. This precise definition of mesorectal involvement has a direct impact on therapeutic decision-making because cancer reaching the margins of the mesorectum is considered to be a T4 lesion even if no pelvic organ is involved (Fig. 16.7) (Heald and Ryall 1986). Such lesions must be treated by preoperative radiochemotherapy.

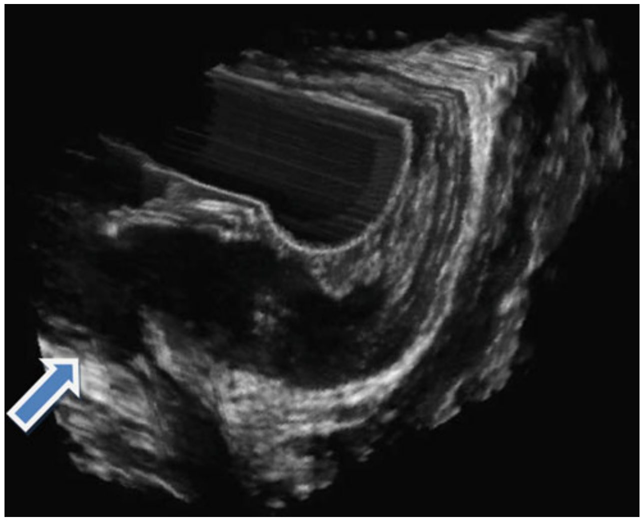


Fig. 16.7 3D reconstruction of a rectal cancer with invasion of the esorectum (*arrow*)

4 Colonic EUS

The place of colonic EUS is less well define, main indication is the use of linear echoendoscope to reach difficult lesions for a biopsy (Giovannini et al. 1998) or the use of mini-probes to access submucosal lesion or extrinsic compressions. But, some studies evaluating the role of neo-adjuvant chemotherapy in advanced colonic cancer (T3N1 or T4 on Ct-scan imaging) are ongoing. If these studies show a real impact on the survival rate, the place of the colonic EUS will be more important in the future.

5 Conclusion

EUS is the most accurate tool for the local staging of rectal carcinoma. In addition to providing accurate T- and N-staging, accurate endosonographic staging guides the choice of the optimal method of management of rectal carcinoma, type of resection, and candidacy for neoadjuvant therapy. Repeat sigmoidoscopic and endosonographic imaging may be considered in selected patients following neoadjuvant therapy. EUS-guided FNA can be used to detect suspected local recurrence.

5.1 Practice Points

- The accurate staging of rectal and anal carcinoma is crucial for planning surgery and indication for adjuvant therapy.
- EUS and MRI remain the most accurate methods for staging rectal and anal cancer.

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Radiologic Imaging of Colo-Recto-Anal Dysfunctions: Procedures and Indications

17

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Contents

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Table 17.1 Rome III diagnostic criteria chronic functional constipation

1. The presence of two or more of the following characteristics during the previous 3 months but with onset at least 6 months previously
(a) Straining associated with at least 25 % defecations
(b) Hard or lumpy stools associated with at least 25 % defecations
(c) Feeling of incomplete evacuation associated with at least 25 % defecations
(d) Sensation of anorectal obstruction associated with at least 25 % defecations
(e) Manual maneuvers to facilitate at least 25 % defecations (e.g., digital evacuation, manual raising of the pelvic floor)
(f) Fewer than three bowel movements per week
2. Loose stools are rarely present without laxatives
3. Insufficient criteria for a diagnosis of irritable bowel syndrome (IBS)

The primitive functional constipation is the main colorectoanal dysfunction and is clinically characterized by reduced weekly frequency of bowel movement, difficult evacuation, and increased stool consistency. The patient reports long straining, hard stools, sensation of incomplete evacuation, anorectal obstruction, and use of manual maneuvers (digital evacuation, lifting of the perineum). The diagnostic criteria of Rome III define this type of constipation (Table 17.1) (Bharucha and Pemberton 2013; Bharucha et al. 2006). Its mechanisms are slow intestinal transit and/or altered defecation. The imaging techniques dedicated to the study of evacuative alterations are barium proctography and magnetic resonance proctography; more recently, the perineal ultrasound of the posterior pelvic compartment was introduced. The study of the colonic transit time is executed when there is a suspicion of a functional constipation due to slow transit (Corazziari 2013).

1 Unit of the Pelvic Floor

The pelvic floor, home of the anatomical structures devoted to defecation, is schematically divided into three compartments: anterior, middle, and posterior. The first compartment includes the bladder; the second, uterus and vagina; the third, anus and rectum. Fascial and muscular structures hold together and support these three sectors. The fascial structures consist of the endopelvic fascia with its different locations and functions (Stoker et al. 2001).

The muscular structures include the urethral muscles, the bulbospongiosus and ischiocavernous muscles, the superficial and deep transverse muscles, the anal sphincters, and the main muscle that keeps together the pelvic floor for its insertions and functions: the levator ani (Strohbehn 1998).

The female pelvic cavity in the course of evolution had to adapt to three fundamental requirements: acquiring a functional conformation at orthostatic position, being able to accommodate the development of the fetus, and allowing the performance of labor with a capacity space much more limited compared to that of other mammals (Schimpf and Tulikangas 2005). It follows that a failure of the supporting structures may determine a competition between prolapsing organs to occupy the limited pelvic space (crowded pelvis syndrome) (Kelvin and Maglinte 1997). For this reason some prolapses are clinically silent and/or hardly appreciable by clinical examination. A surgery that repairs the clinically manifest alterations can, by freeing up space, unmask the silent ones. Anatomical defects that are not repaired for being silent can become symptomatic over a relatively short time (Withagen et al. 2010). The frequency of alterations in other compartments associated with difficult evacuation is high: 95 % of patients have multiple compartmental defects (Maglinte et al. 2011). Clinical evaluation and physical examination are the first approach to pelvic floor disorders, but physical examination is insufficient for understanding the causes of the evacuative alterations, and the prolapses can be underestimated or not diagnosed at all (Kelvin et al. 1999). Whatever the method of approach adopted, be it clinical or instrumental, it must take into account the influence of gravity on the descent of the pelvic floor and on the expression of each structural alteration. The examination in supine position can restrict or prevent such events and make it difficult to assess them. The sitting position instead allows for the complete relaxation of the pelvic muscles during the thrust phase, allowing the full expression of the prolapse (Maglinte et al. 2011, 2013; Ribas et al. 2014).

2 Barium proctography

The introduction of barium proctography dates back to the 1960s, but its standardization took place in the early 1980s thanks to P. Mahieu et al. (1984a, b).

This technique began with the study of rectoanal dysfunctions and evolved progressively extending to the study of the bladder, the vaginal axis, and the prolapse of the small bowel and sigma (dynamic cystoproctography) (Maglinte et al. 1997). With this technique it is possible to obtain a single view of all the organs supported by the pelvic floor and to identify clinically silent prolapses.

Barium proctography without opacification of the bladder, but with opacification of the small intestine, the vagina, and the anorectal region, is currently considered the gold standard for the study of the morphological and dynamic anorectal dysfunction (Maglinte and Bartram 2007).

2.1 Clinical Indications

1. *Obstructed defecation*: rectoanal obstacle to evacuation, incomplete emptying, prolonged defecation effort, use of manual maneuvers to aid the expulsion of the feces.
2. *Light incontinence* (in serious incontinence the retention of barium is impossible). The exam is indicated especially in cases where ultrasound and endoanal manometry do not detect significant sphincter damage (Terra and Stoker 2006). In about a third of the cases, obstructed defecation and incontinence are both present (Siproudhis et al. 2006; Shorvon and Marshall 2005).
3. *Operated patients:* the exam is indicated in patients who have received surgery for anorectal reconstruction (e.g., ileal pouch) and have evacuative difficulties or in patients operated for obstructed defecation who, because of failure or complications of surgery, need reassessment (Shorvon and Marshall 2005).
4. *Chronic or frequently recurrent proctalgia*. Dyssynergic defecation is the main cause of pain in most patients (Shorvon and Marshall 2005). Besides the dyssynergia that can be present even in absence of constipation (Chiarioni et al. 2011), if the cause is a solitary rectal ulcer an intussusception can be found (Womack et al. 1987).

2.2 Technical Aspects

The examination requires a comfortable dedicated radiolucent commode on which the patient sits and performs the defecation act (Mahieu et al. 1984a) (Fig. 17.1).

Fig. 17.1 Barium proctography. The commode for the proctography is located on the platform of the upright radiological table. Below the bolster of the commode is placed a rubber donut filled with water to increase the contrast and improve the quality of the images

The evening of the day before the exam the patient carries a rectal cleaning enema. Some authors prefer the introduction of a laxative suppository about an hour and a half before the exam (Maglinte et al. 2011).

An hour before the examination the patient drinks a diluted barium suspension.

In our center, in this hour of waiting, medical history is collected, and the examination and its purposes are explained to the patient; a clinical examination of the pelvic floor is performed in order to be compared with the results of the barium proctography. A pelvic ultrasound is performed to highlight the presence of a cystocele, of a uterine prolapse (when the uterus is clearly visible), or of a peritoneocele containing or not bowel loops; an initial assessment of the posterior pelvic compartment by ultrasound is also performed.

About an hour after the barium was ingested, we proceed to the opacification of the vagina and rectum, respectively, with 20–30 and 200–300 cc of density semisolid barium pasta (by analogy with the fecal material) that we prepare with a mixture of 100 g powder of barium sulfate, 100 ml of water, and 100 cc of ultrasound gel. Some authors resort to a semisolid mixture of barium and potato starch (Mahieu et al. 1984a). For administration we use 50–60 cc syringes and the cannula attached to the barium bag. Some authors, and we among them, prefer not to stick to a standard quantity but proceed with the introduction until the patient feels the urge to evacuate; the purpose is to facilitate the stage of evacuation, making it as similar as possible to the physiological one. If a radiological study of the anterior compartment is required, we proceed to the retrograde opacification of the bladder. After the insertion of a Foley, iodinated contrast is introduced. The quantity is about 30 ml if the purpose is only to investigate the presence of a cystocele (Maglinte et al. 2013); in order to study also the voiding phase, the mobility and the morphology of the urethra, between 100 and 150 cc of contrast medium is introduced, so that the patient feels the urge to urinate. To avoid interference between compartments, it's best to run the study of the emptying of the organs separately, filling them in subsequent times: after the rectum-emptying assessment, the bladder is filled with contrast medium for its static and dynamic evaluation.

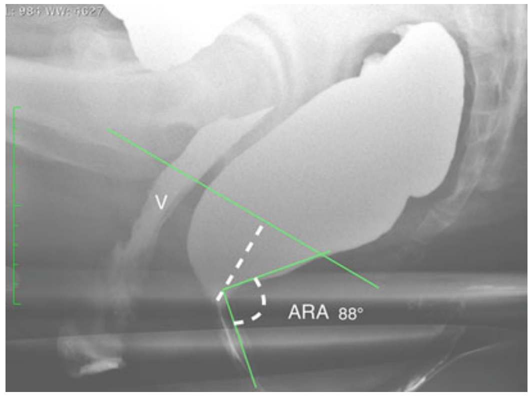

Fig. 17.2 Barium proctography. Normal proctogram. Resting rectal position. The anorectal angle (ARA) is located between the longitudinal axis of the anal canal and the posterior wall of the rectum. The pubococcigeal line (*continuous line*) extends from the bottom edge of the pubic symphysis to the coccyx. The position of the anorectal junction is measured by tracing the perpendicular line (*dotted line*) between the pubococcygeal line and the anorectal junction; *v* vagina

The barium proctography includes three initial radiographs, one at rest (Fig. 17.2), one during voluntary contraction (to assess the contractile ability of the puborectalis sling) (Fig. 17.3), and one in straining (to assess the activities of the external anal sphincter) (Maglinte et al. 2011). Not all authors consider the phase of straining to be useful, because of the difficulty for some patients to push for fear of losing the contrast medium (Maglinte et al. 2011). In the next phase of the evacuation, the patient, at the invitation of the operator, empties the rectum (Fig. 17.4). The descent of the anorectal junction, the opening of the anal canal, the enlargement of the anorectal angle, and the emission time of the contrast medium are evaluated. The evacuation is a complex physiological process based on the interaction between the colonic and rectal activities. In barium proctography it's possible to evaluate only the emptying of the extraperitoneal rectum, showing the passive emptying induced by voluntary abdominal pressure. The retention of barium above the fold cross of the Houston valve (plica border of this part of the rectum) is of no

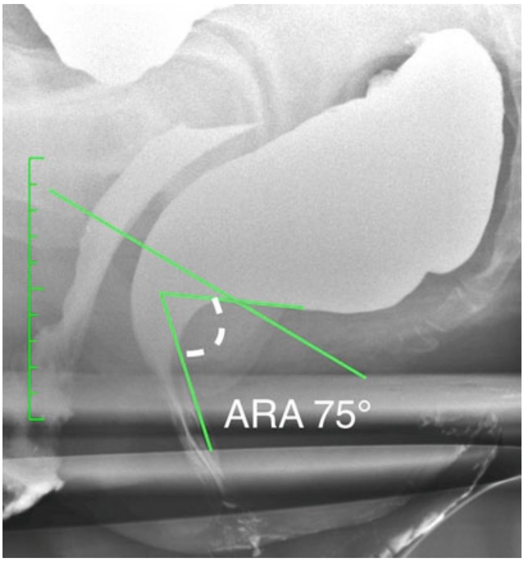

Fig. 17.3 Barium proctography. Normal proctogram. Voluntary contraction phase: (**a**) the amplitude of the angle is reduced and (**b**) the anorectal junction ascends compared to the resting condition

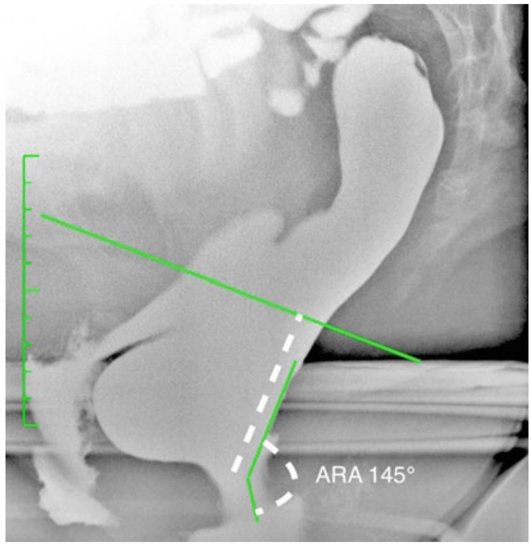

Fig. 17.4 Barium proctography. Normal evacuation. The amplitude of the anorectal angle increases and the anorectal junction descends if compared to the resting condition. The anal canal opens within 5 s since the start of the push and the evacuation is completed within 30 s

significance in the exam assessment (Maglinte and Bartram 2007).

The evacuative or dynamic phase is recorded with a digital system of serial acquisitions (one image per second for 30 seconds) or with video-tape. The system is calibrated to compensate for the radiographic magnification (Maglinte and Bartram 2007).

2.3 Evaluation Parameters

2.3.1 Pubococcygeal Line

In order to identify the location of the anorectal junction at rest and during the emptying phase, a reference line, called pubococcygeal, is drawn between the lower edge of the pubis and the most visible coccygeal articulation. Some authors prefer to use, as extreme tail of the line, the sacrococcygeal articulation, in order to prevent the mobility of the coccyx. In both cases the line is chosen because it closely approximates to the position of the levator ani, which is the plane of reference for the assessment of the descent of the pelvic organs (Maglinte et al. 2013; Kelvin and Maglinte 2000).

The descent of the posterior pelvic floor is measured by drawing the perpendicular line between the anorectal junction (boundary between the rectum and the anus) and the pubococcygeal line. The normal value at rest does not exceed 3–4 cm (Kelvin and Maglinte 2000). Later this measurement is performed on the radiogram of the evacuation phase. The descent of the anorectal junction normally does not exceed 3–4 cm (Kelvin and Maglinte 2000). There are authors who prefer to use, as a reference line, the floor of the commode or the line through the ischial tuberosities (Maglinte and Bartram 2007; Shorvon and Marshall 2005); the reason is that, before the introduction of digital radiology, the pubis was not always identifiable. The anorectal junction at rest normally does not drop below the ischial tuberosities; the evacuative excursion does not exceed 3–4 cm (Shorvon et al. 1989).

The radiologically detected mobility on healthy subjects in position of defecatory excursion is greater than the one detected clinically (Shorvon et al. 1989).

2.3.2 Anorectal Angle (ARA)

It is the angle between the longitudinal axis of the anal canal and the tangent to the posterior margin of the rectum. It is due to the activities of the levator ani; indirectly it provides an assessment of the function of this muscle, by reducing during the voluntary contraction and widening during the emptying (phase that corresponds to the complete relaxation of the muscle complex). The normal and pathological values of this angle largely overlap, making, according to some authors, its measurement of little use (Felt-Bersma et al. 1990; Bartram et al. 1988).

The normal values are at rest (80°–120°), emptying (115°–150°), and squeezing (75°–104°) (Habib et al. 1992).

2.3.3 Length of the Anal Canal

Normal values at rest are between 10 and 38 mm in men and from 6 to 26 mm in women (Shorvon et al. 1989).

2.3.4 Diameter of the Anal Canal

The closing state of the anal canal is evaluated in resting conditions. The normal opening diameter during evacuation is between 8 and 18 mm (Piloni et al. 1993).

2.3.5 Transverse Diameter of the Rectum

It corresponds to the distance between the front and the rear wall of the average rectum. Normally it does not exceed 6.5 cm (Piloni et al. 1993).

2.3.6 Rectosacral Space

It is the space between the rear wall of the rectum and the sacral concavity at S3 level; it does not exceed 10 mm in conditions of rest (Shorvon et al. 1989).

2.3.7 Evacuation Times

They include the time of opening of the anal canal and the time between the beginning of the act of expulsion and the end of the discharge; the latter is determined by the complete emptying of the ampoule, by the perception of completeness of the evacuative act, or by a hindrance in the rectoanal region arisen during the expulsion. The average latency in the opening of the anal canal does not exceed 3–5 s. The actual time of evacuation is less than 30 s with an average value of 11 s (Bartram et al. 1988).

2.3.8 Evacuative Residue

The evacuative residue does not exceed one-third of the normal released quantity. It is calculated with planimetric measures. The difference between the radio-opaque rectal area before and after the emptying is comparable to the difference between the weight of the introduced barium and that of the expelled barium (Halligan et al. 1994).

In a recent study (Palit et al. 2014) conducted in healthy volunteers, the average total time of evacuation was 88 s in men and 128 s in women. The average percentage of total evacuation was 71 % in men and 65 % in women. The study shows a broader range of efficiency of evacuation than in previous reports. The probable explanation is that the administration of contrast medium was not a quantitative standard but varied in relation to the perception of a sustained desire to defecate. It is likely that this diversity caused the wide variability (Palit et al. 2014).

2.3.9 Pattern of Evacuation

In normal subjects, three evacuative patterns have been described: type 1 characterized by the rapid opening of the anal canal and rapid evacuation of the contrast medium, type 2 defined as "pulsating" (emission of small amounts of contrast medium during short and successive time intervals), and type 3 characterized by continuous but slow evacuation (Palit et al. 2014).

3 Functional and Structural Anorectal Abnormalities

Anorectal abnormalities, mainly appreciable during evacuation, are both structural and functional.

3.1 Functional Abnormalities

The beginning of defecation takes place with the voluntary contraction of the abdominal muscles, the voluntary relaxation of the pelvic muscles, the descent of the anorectal junction, the expansion of the anorectal angle, and the opening of the anal canal. When the patient stops the straining, restores the normal tone of the anal sphincters, and lifts the anus, the anal canal closes, and the anorectal angle decreases. The anorectal junction returns to its resting position (postdefecation reflex) (Maglinte et al. 2011).

Functional abnormalities represent an alteration of the physiological evacuative function.

According to the Rome III criteria they include

1. Dyssynergic defecation

   This is the inappropriate contraction of the pelvic floor muscles (anal sphincter and/or puborectalis muscle) or the partial reduction of the basal pressure of the anal canal during the expulsive push (Fig. 17.5).
2. Impaired defecation by inadequate propulsion

   This is the increase of the intrarectal pressure caused by an inadequate contraction of the muscles of the abdominal wall, with or without signs of dyssynergic defecation.

   These alterations can be detected by manometry, some from electromyography, and partly by imaging techniques (RAO).

3.1.1 Dyssynergic Defecation

A plethora of terms were used in the past to indicate dyssynergic defecation, namely, inappropriate contraction, failure or insufficient relaxation of the puborectalis muscle and/or of external anal sphincter, anismo, dyssynergia of pelvic floor muscle, paradoxical contraction of puborectalis muscle, puborectalis syndrome, and spastic pelvic floor syndrome (Rao and Meduri 2011).

The term "dyssynergic defecation" is better fit to describe the plurality of the muscles involved and the alteration of their coordination (Rao and Meduri 2011).

Signs of Dyssynergic Defecation

Initially the defecographic signs indicated as diagnostic elements were the accentuation of the puborectalis impression and the reduction of the ARA during the evacuation (Mahieu et al. 1984b).

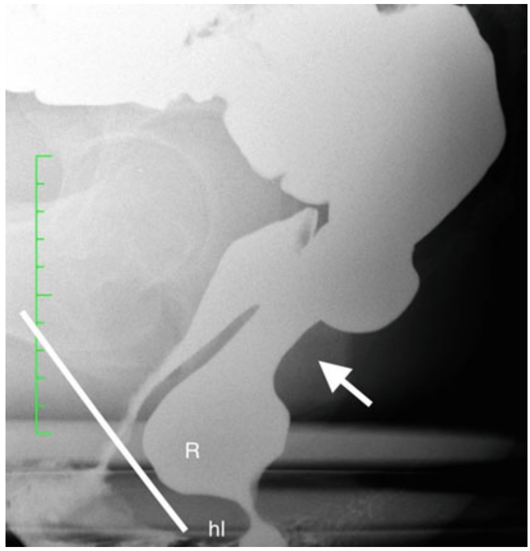

Fig. 17.5 Barium proctography. Dyssynergic defecation. The lateral radiogram shows the accentuation of the puborectalis impression (*white arrow*) during the maximum thrust and the reduced opening of the anal canal (*ac*). The evacuation time is longer than 30 s. There is a rectocele (*r*) from distension. It is positioned above the hymen plane

The puborectalis impression is the indentation that the puborectalis muscle forms immediately above the anorectal junction, on the rear side of the rectum (Shorvon et al. 1989). Normally, with the beginning of the expulsion phase, in conjunction with the relaxation of the muscle, this impression disappears (Mahieu et al. 1984b).

In some cases of dyssynergic defecation, the persistence of this footprint has been observed (Mahieu et al. 1984b). Halligan et al. subsequently found that this sign is not useful to differentiate individuals with functional alterations from healthy ones (Halligan et al. 1995a, 2001). The parameters that have proved to be more sensitive to this diagnosis are the prolonged evacuation (>30 s) and the partial removal of the contrast medium (<60 %). The association of the prolonged evacuation with the incomplete emptying has a positive predictive value of 90 %. Another sign that's present, although not constant, is the reduced opening of the anal canal (Halligan et al. 2001).

Of the various methods used for the diagnosis of dyssynergic defecation (imaging techniques, balloon expulsion test, manometry, electromyography), none can be considered the gold standard for diagnosis. The reliability of the diagnosis is given by the concordance of more techniques (Rao and Meduri 2011).

3.1.2 Altered Defecation by Inadequate Propulsion

A normal evacuation requires an adequate intrarectal pressure and anal muscle relaxation. The pressure increase achieved by the voluntary contraction of the diaphragm and of the abdominal muscles causes an increase of the intrarectal pressure. Halligan et al. described an altered defecation characterized by prolonged emptying time, descent of the anorectal junction below 3 cm, and intrarectal reduced pressure in the absence of other changes (Halligan 2008). Faucheron et al. described a very similar form of evacuative alteration, defined as rectal akinesia (Faucheron and Dubreuil 2000).

In the same patient a reduced intrarectal pressure and a paradoxical contraction or insufficient relaxation of the pelvic muscles can coexist (Bharucha and Pemberton 2013).

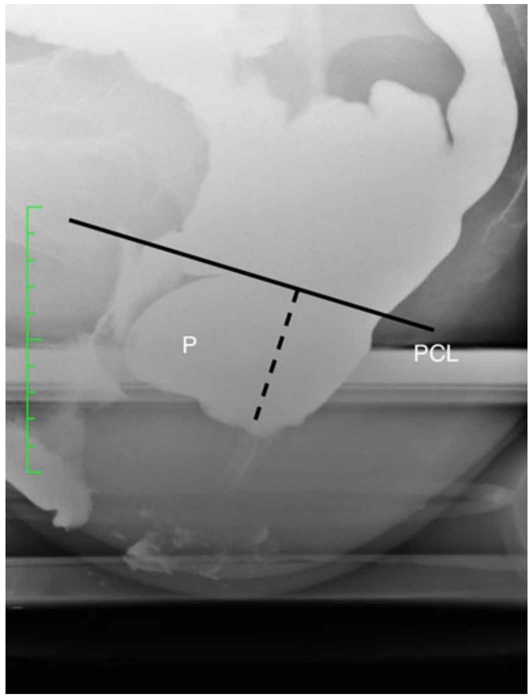

Fig. 17.6 Barium proctography. Lateral proctography at rest. Descended perineum: the anorectal junction appears to be located 8 cm below the pubococcygeal line (nv < 4). It's a sign of weakness of the muscular-fascial supports of the pelvic floor. A protrusion (*p*) of the rectal anterior wall is also present, indicating that the rectovaginal septum had already failed in resting conditions

3.2 Structural Anorectal Abnormalities

3.2.1 Descending Perineum

It is the condition where, during the evacuation, the anorectal junction descends more than 3–4 cm compared to the resting position.

3.2.2 Descended Perineum

The anorectal junction at rest appears positioned more than 3–4 cm below the pubococcigeal line or the ischial tuberosities (Fig. 17.6).

The descended perineum is considered to be the consequence of a structural failure of the muscle-fascial supports of the pelvic floor, as a result of protracted efforts during the evacuation. These efforts can also cause the stretching of the pudendal nerve with secondary neuropathy (Halligan 2008).

With aging, the frequency of the descended perineum grows and the frequency of the descending perineum decreases, due to the loss of elasticity of the muscle-connective tissue support. The pudendal neuropathy can evolve into the sphincter denervation, resulting in incontinence (Pemberton 1990). Other factors that can damage or weaken the pelvic floor are childbirth, high BMI, nutritional deficiencies, and diseases of the collagen (DeLancey et al. 2007).

3.2.3 Megarectum

Megarectum is an alteration in which the transverse medium rectal diameter is greater than 6.5 cm (Piloni et al. 1993). It is a condition rarely seen in healthy subjects. A transverse diameter greater than 8.1 cm in an adult male and 6.9 cm in an adult woman should lead to further investigation (Palit et al. 2014). It can be associated with an increased threshold in the rectal sensitivity,

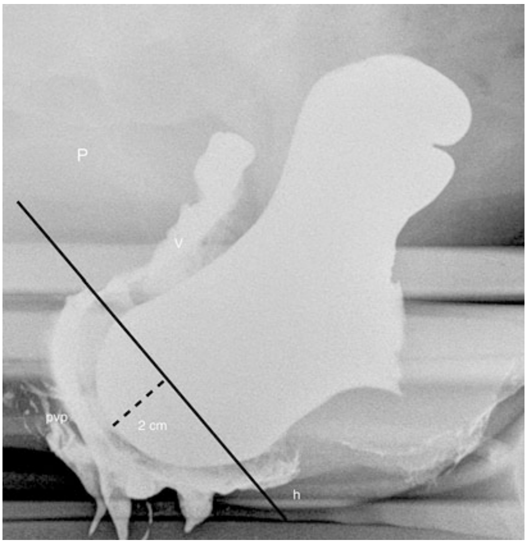

Fig. 17.7 Barium proctography. Lateral radiograms during the evacuation. Rectocele: herniation into the vagina (*v*) of the anterior rectal wall. Rectocele from dislocation: the rectocele protrudes 2 cm beyond the hymen plane (*parallel line* to the longitudinal axis of the pubis (*P*), passing through the posterior margin of the vaginal fork) (*h*); an excessive descent of the vaginal apex can be observed. This type of rectocele is the result of damage to the rectovaginal septum after childbirth. In anterior vaginal axis dislocation corresponding to posterior vaginal prolapse (*pvp*)

which can cause evacuative difficulties (Meunier et al. 1979).

3.2.4 Increase of the Rectosacral Space

A rectosacral space greater than 1 cm can be a sign of a large mesorectum and can be associated with an external rectal prolapse (Mackle and Parks 1986).

3.2.5 Rectocele

Rectocele is the herniation into the vagina of the anterior rectal wall; it is associated with herniation of the posterior vaginal wall (posterior colpocele). In addition to the rectocele, the posterior colpocele can be associated with enterocele, sigmoidocele, or peritoneocele.

The rectoceles can be classified according to position (low, medium, high), size (small <2 cm, medium 2–4 cm, large >4 cm), or degree (type I with bulging in the upper vagina, type II extending to the vaginal introitus, type III extending beyond the vaginal introitus) (Zbar et al. 2003). Rectoceles smaller than 2 cm are a normal finding in nulliparous women (Shorvon et al. 1989). A useful subdivision for the clinical diagnosis is the distinction in rectocele by distention, rectocele by displacement, and mixed rectocele (Maglinte et al. 2013). The first type is associated with dyssynergic defecation (Pucciani et al. 1996). The vaginal apex and the uterus are in the normal position. This type of rectocele is the consequence of a chronic evacuative effort. It is positioned above the hymen. The second type, rectocele from dislocation, is the result of damage to the rectovaginal septum at childbirth. It is located below the hymen and associated with failure of the pelvic floor muscles and of the perineal body (Marti et al. 1999) (Fig. 17.7). It results in excessive descent of the vaginal apex and of the uterus. The third type, the mixed type, is caused by a failure of muscle tissue caused by dyssinergic defecation (Fig. 17.8). The type 2 rectocele shows a broader anorectal angle at rest and during evacuation, greater descent of the anorectal junction, and a more frequent association with intussusception: 61 % of the cases (Pomerri et al. 2001).

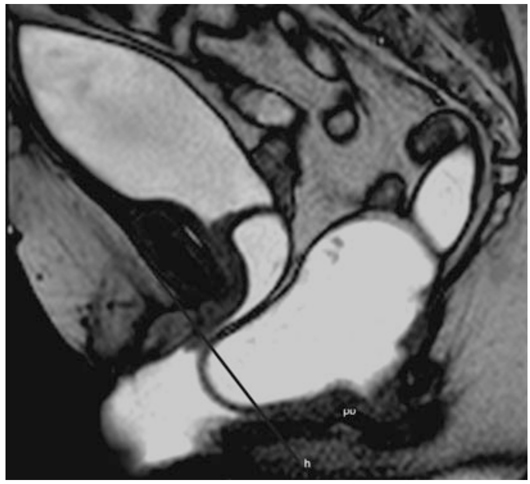

Fig. 17.8 MR proctography. Fast T2 sequence weighted on the midsagittal plane, obtained in patients with dyssynergic defecation. A large rectocele filled with contrast pushes forward the posterior vaginal wall and stretches of the perineal body without emptying. The anterior wall of the rectocele extends beyond the plane of the hymen. It's a mixed rectocele

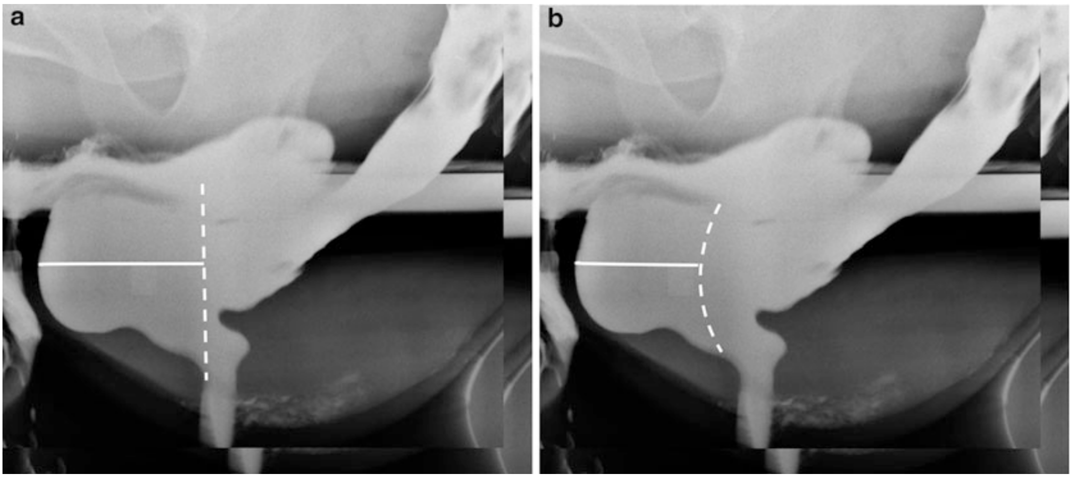

Fig. 17.9 Barium proctography. Measurement of the rectocele: (**a**) distance between the extended line of the anterior border of the anal canal and the tip of the rectocele. This measurement is easier to achieve, but it overestimates the true depth of the rectocele (**b**) distance between the ideal line demarcating the normal position of the anterior rectal margin and the tip of rectocele

Symptomatic Rectoceles and Their Measurement

Rectocele occurs most frequently during evacuation. A large rectocele is more likely to be symptomatic. The size of the rectocele is measured as the distance between the extended line of the anterior border of the anal canal and the tip of the rectocele (López et al. 2001) or, according to other authors, from the ideal line demarcating the normal position of the anterior rectal margin (Shorvon et al. 1989) (Fig. 17.9a, b). The first measurement is easier to achieve, but it overestimates the true depth of the rectocele. The evaluation is completed by measuring the width of the rectocele, that is, its height starting from its basis (Palit et al. 2014). The percentage of emptying is measured by the difference of its area before and after the evacuation. A normal retention should not exceed 10 % of the initial area (Halligan et al. 1994). The amount of the retained contrast medium is considered, however, extremely variable even in asymptomatic subjects; the evacuative difficulties cannot be evaluated only on the basis of this retention. Also, there is no consensus on the relationship between the size of rectocele and the emptying (Greenberg et al. 2001).

An interesting pathophysiological observation is that rectoceles which do not retain barium do not show any difference in pressure compared to the proximal rectum; on the contrary, rectoceles with trapping in 64 % of the cases show a sharp drop in pressure compared to the proximal rectum. They behave as inert rectal pouches. However, no differences have been found in evacuative difficulty between the two groups (Halligan et al. 1995b). Regardless of the depth of the rectoceles, it was detected that an attempt of evacuation in the toilet, after the end of the examination, significantly increases the percentage of emptying (Maglinte et al. 2011).

Symptoms of the Rectoceles

A rectocele may determine a feeling of incomplete evacuation, use of digital maneuvers (introduction of the finger into the vagina and pressure on its posterior wall or lifting and thrust back of the perineum) to facilitate emptying. The surgery can improve the emptying of the rectocele and reduce the associated vaginal bulge (Van Laarhoven et al. 1999). The rectocele-dyssynergia association reduces the chances of success of the surgery (Karlbom et al. 1996).

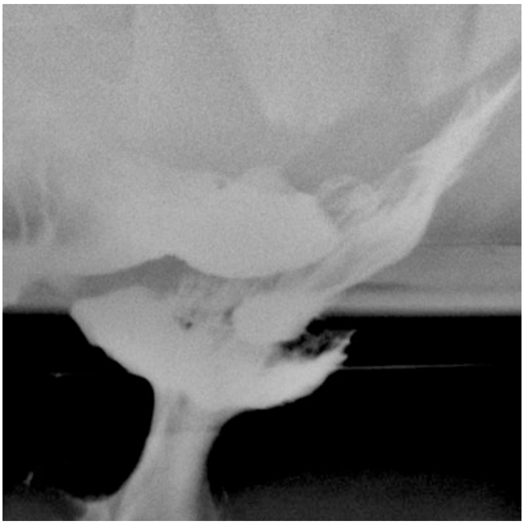

Fig. 17.10 Barium proctography. Rectorectal intussusception. Lateral proctography reveals circumferential rectal infolding during evacuation. The intussusception is limited to the rectum and does not go below the upper rectoanal margin

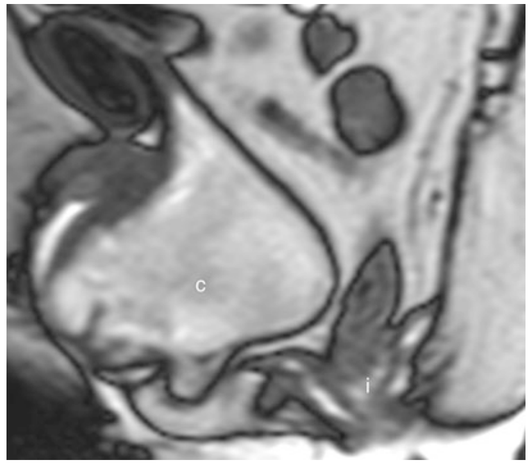

Fig. 17.11 MR proctography. Fast T2 sequence weighted on the midsagittal plane, obtained at the end of the evacuation. Large cystocele (*c*) and rectorectal intussusception *i*) (the rectal walls fold into the rectum but do not enter the anal canal)

Rectoceles in Males

Small rectoceles may be present also in men, especially after total prostatectomy (Cavallo et al. 1991).

3.2.6 Posterior-Lateral Herniations of the Rectum

They may result from a failure of the iliococcygeal muscle during childbirth. They are defined as ischiorectal hernia if they have a depth greater than 4 cm (Grassi et al. 1995).

3.2.7 Intussusception

Definition, Classification, and Morphology of the Intussusception

The intussusception is the introversion at full thickness of the rectal wall into the lumen of the rectum. The diagnosis of the intussusception, unlike that of the rectocele, happens at the end of the evacuative phase when the intussusception of the rectum is maximum and it is possible, thanks to the rectal emptying, to distinguish the invaginated segment from the segment that surrounds it (Kelvin et al. 1994). The invagination limited to the rectum is called intrarectal (Figs. 17.10 and 17.11); it's called rectoanal if it enters the anal canal (Fig. 17.12). A more detailed classification of the intussusception is the one made by the Oxford Radiological Grading System. It includes five degrees: grade I corresponds to high rectorectal intussusception (the top of the invagination stops above the proximal edge of the associated rectocele); grade II, when the apex of the invaginated segment descends to the entrance of the rectocele; grade III, when the apex of the invaginated segment descends to the upper margin of the anal canal; grade IV, when the apex of the invaginated segment enters the anal canal; and grade V, when the invaginated segment protrudes outside the anal canal (it corresponds to the external rectal prolapse) (Shorvon et al. 1989).

This classification is useful to follow the evolution over time of the intussusception. Sometimes the rectum appears collapsed after emptying and still under pressure, and this can lead to suspect an intussusception. In these cases, to settle the question, the frontal radiogram is useful. It will show that a collapsed rectum looks like an inverted "3" that can be mistaken for an intussusception (McGee and Bartram 1993).

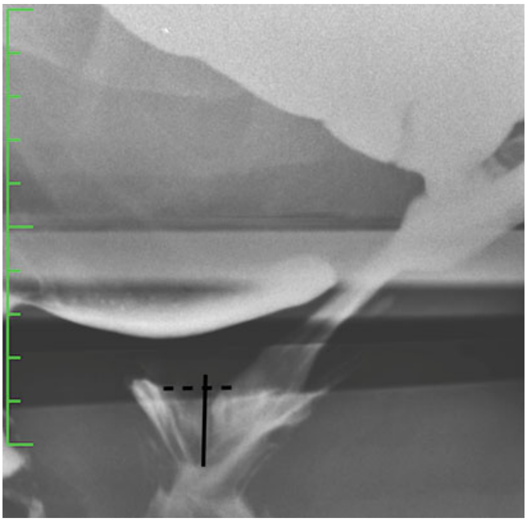

Fig. 17.12 Barium proctography. Rectoanal intussusception. The lateral proctography toward the end of the evacuation reveals the circumferential infolding of the rectal walls during evacuation. The apex of the intussusceptum enters the anal canal. Measurement (**a**) of the thickness (*dashed line*): distance between the edge of the intussuscipiens and the contour of the intussusceptum. The depth of the intussusception (**b**) (*continuous line*): distance between the starting point of the intussusception and the anorectal junction

The intussusception is by definition circumferential. It can be predominantly front or rear. The thickness between the edge of the invagination and the surrounding segment is 3 mm or more.

Pathogenetic Hypothesis

The most credited pathogenetic hypothesis is that the intussusception is secondary to a lack of, or incomplete, relaxation of the levator ani resulting in straining during defecation and subsequent failure of the muscle-fascial structures of support (Pucciani 2008).

The evacuative difficulties may damage the anal sphincters over time. In some rectoanal intussusceptions, an excessive opening of the anal canal is observed (Pomerri et al. 2001).

Symptomatic Intussusceptions

The intussusception is present in 50 % of the asymptomatic population. In symptomatic subjects its thickness is significantly greater: the average anterior thickness is 8 mm versus 4 mm in asymptomatic subjects; the average posterior thickness is 5 mm versus 3 mm in asymptomatic subjects. There are no significant differences in the depth of the intussusception (Dvorkin et al. 2005a). The ratio between the diameter of the invaginated segment and the surrounding intestinal wall appears higher than 2.5 in symptomatic subjects (Pomerri et al. 2001).

In a study evaluating the predictability of the symptoms in relation to defecographic alterations, the pain and the feeling of prolapse are highly predictive of isolated intussusception; the vaginal bulging, of rectocele; and the straining, of rectocele and intussusception in association (Dvorkin et al. 2005b). The authors speculate that the cause of pain in patients with isolated anorectal intussusception may be linked to the presence of solitary rectal ulcer. The genre does not alter the predictive ability of the symptoms. Regarding the morphology of the intussusception (rectorectal or rectoanal), the "toilet revisiting" symptom (the patient after defecating continues to feel the need to defecate and returns in the toilet for a further attempt to empty the rectum) is the only one that's predictive of rectoanal intussusceptions (Dvorkin et al. 2005b).

No symptom is predictive of the defecographic sign of mechanic obstruction.

The fecal incontinence is described as a symptom at times associated with the rectoanal intussusception. It is presumed that it develops after a long history of rectoanal intussusception that can cause a widening of the anal canal, thus altering its sensitivity (Weiss and McLemore 2008; Wijffels et al. 2010; Collinson et al. 2009; Collison et al. 2008).

The enterocele may promote the formation or the aggravation of an intussusception, considered as a peritoneocele of the rectal wall inside which the enterocele descends. In a thick posterior intussusception, the penetration of a particularly large mesorectum is likely (Wijffels et al. 2010; Hoffman et al. 1984).

Evolution of the Intussusceptions

There is an increase of intussusceptions in relation to age. In males, a progression of 2.5 years for each degree of prolapse was calculated. For

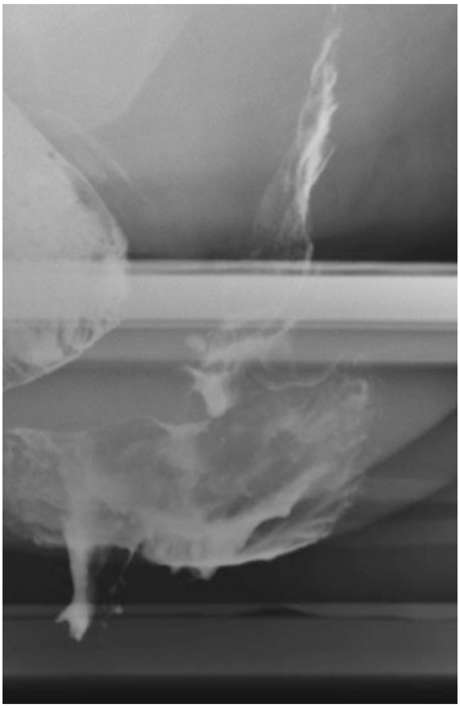

Fig. 17.13 Barium proctography. The lateral radiogram at the end of the evacuation shows a complete rectal prolapse represented by a complete extrusion of the entire rectal wall through the anus

nulliparous women, the progression is of 2.6 years per degree of prolapse. For women with one or more childbirths there is a gradient of progression of 5.4 years per degree of prolapse. The study was conducted on a sample of 60 men and 471 women (Wijffels et al. 2010).

Mellgren et al. and Choi et al. report low percentages (6.7 % and 3.8 % respectively) of evolution of the intussusception into external rectal prolapse (Mellgren et al. 1997; Choi et al. 2001).

3.2.8 External Rectal Prolapse

The rectum is extruded through the anus. All of the rectal wall layers are involved (Fig. 17.13).

Anatomical changes that favor the formation of an external rectal prolapse include particularly deep Douglas, redundancy of the rectosigmoid, posterior elongation of the anchor structures of the rectum with subsequent increase of the presacral space, laxity of the lateral ligaments, and diastasis of the levator ani. The weakness of the external sphincter, the presence of a dilated anal canal with a reduced tone, and the reduced contractile capacity of the puborectalis muscle are frequently detectable characteristics in the external rectal prolapse. The complete rectal prolapse occurs mostly in the elderly, and 80–90 % of those affected are females, with a peak between the sixth and seventh decade. Up to 40 % of the women are represented by nulliparas. In males this condition is distributed more uniformly among different ages, peaking in the second and third decade (Lowry and Goldberg 1987; Parks et al. 1966).

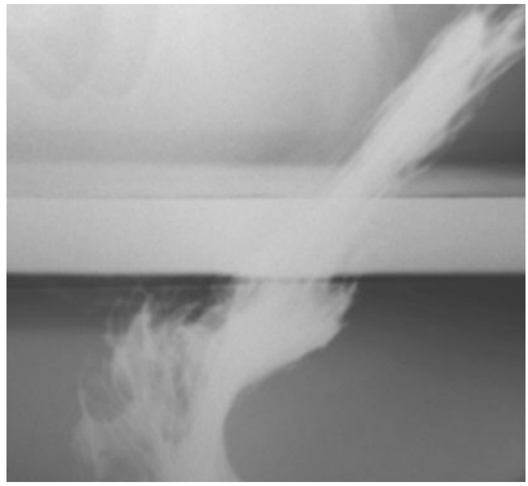

Fig. 17.14 Barium proctography. The lateral radiogram during evacuation shows an anterior mucosal prolapse: introversion of the anterior rectal mucosa into the lumen of the rectum. The thickness of the invaginated mucosal folds is less than 3 mm

3.2.9 Anterior Mucosal Prolapse

Mucosal prolapse is the introversion of the rectal mucosa into the lumen of the rectum or the anal canal (Fig. 17.14). The thickness of the invaginated segment is less than 3 mm. The anterior mucosal prolapse is frequently found during the emptying of a rectocele; it remains intrarectal and does not enter into the anal canal. It is interpreted as a reversal of a redundant rectocele wall. However, it can also be seen in asymptomatic subjects (Parks et al. 1966; Unger et al. 2011; Allen-Mersh et al. 1987).

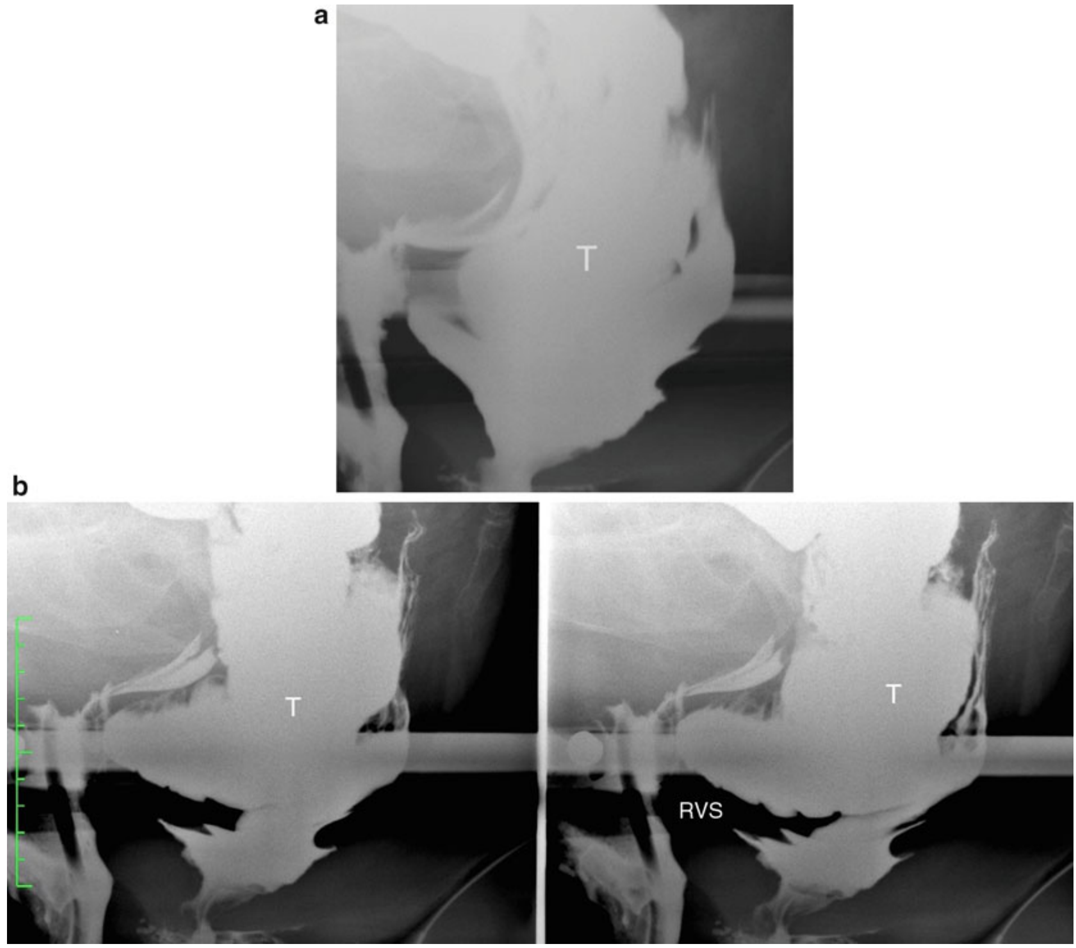

Fig. 17.15 Barium proctography. Enterocele: a herniation of small bowel loops (*T*) in the rectovaginal space (*rvs*) can be observed. The enterocele is evident only at the end of the evacuation, when the rectocele is completely emptied and the rectovaginal space is free to accommodate the loops of the small intestine

3.2.10 Solitary Rectal Ulcer

It is an ulcer or a polypoid formation usually localized on the anterior wall of the rectum, at 6–8 cm from the anal verge. It can be found in patients with rectal prolapse or dyssynergic defecation and/or ultrasound evidence of abnormal thickening of the internal anal sphincter, which is significantly associated with rectoanal intussusception (Womack et al. 1987; Chiang et al. 2006).

3.2.11 Enterocele

Enterocele is the herniation of the small bowel loops in the peritoneocele. The peritoneocele is the extension of the rectouterine sac under the upper third of the vagina. Unfortunately, this definition does not take into account the considerable vaginal apex mobility (Maglinte et al. 1999).

The enterocele can remain confined in the rectovaginal septum; it may extend into the posterior colpocele preceding the entry of a rectocele or occupying it after the emptying of the rectocele (Fig. 17.15, 17.16, 17.17). The enterocele can herniate into an anterior colpocele competing with a cystocele, so that it can be observed only after the emptying of the latter (Fig. 17.18). It can herniate into an intussusception. Rarely, it can herniate posteriorly into the rectum (Maglinte et al. 2011, 2013; Bremmer 1998). In 50 % of the cases the peritoneocele does not contain loops of small

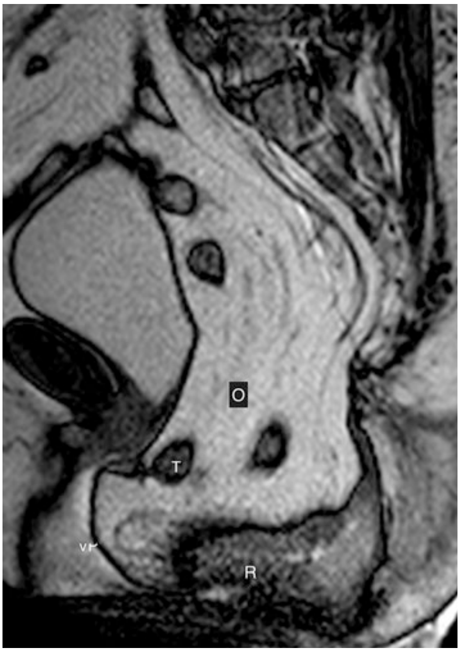

Fig. 17.16 MR proctography. Fast T2 sequence weighted on the midsagittal plane, obtained at the end of evacuation. Descent of omentum and bowel loops (peritoneocele with enterocele) into the rectovaginal space. Also, evidence of rectocele (*r*) and moderate posterior vaginal prolapse (*vp*)

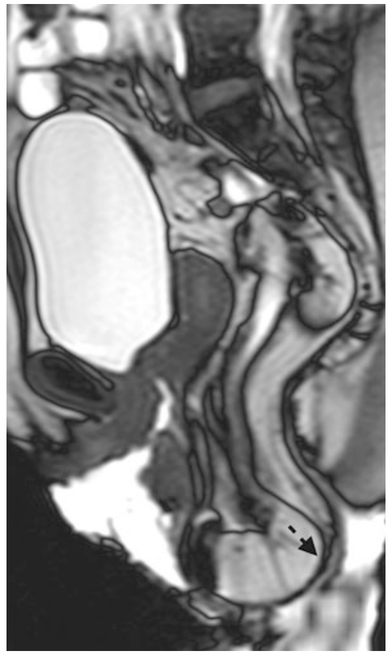

Fig. 17.17 MR proctography. Fast T2 sequence weighted on the midsagittal plane, obtained during evacuation. The enterocele widely occupies the rectovaginal space and hinders the complete emptying of the rectum by pressing on its lower third

intestine but omental fat and, less frequently, sigmoidal loops (Bremmer 1998).

The enterocele is more frequently evident at the end of the evacuation, after the rectum and bladder are empty and there is a release of pelvic space. In up to 50 % of the cases the enterocele occurs after the patient has completed the test in the toilet where, with further evacuative effort, the emptying ends (Maglinte et al. 2011, 2013). There are conditions such as hysterectomy and urethropexy that, by expanding the rectovaginal space, increase the incidence of enterocele. It occurs in fact in 64 % of the patients who have undergone a hysterectomy and 27 % of those undergoing cistopexy (Maglinte et al. 2011, 2013).

Obstructive and Nonobstructive Enterocele

In relation to the effects on the rectum, the enterocele can be obstructive or nonobstructive. Morandi and colleagues distinguished three types of enterocele. Type A is the one that does not reach or does not compress the rectal ampoule during emptying and returns to the previous position after the thrust ends. Type B compresses the rectal ampoule at the end of the evacuation. Type C is called obstructive: it compresses the rectal ampoule at the beginning of the evacuation, determining an obstacle to the expulsion of the barium (Morandi et al. 2010).

3.2.12 Sigmoidocele

The occupation of the peritoneocele by loops of a redundant sigma takes the name of sigmoidocele. It is found in about 5 % of defecographic exams (Jorge et al. 1994; Fenner 1996) (Fig. 17.19a, b).

3.2.13 Vaginal Prolapse: Anterior Colpocele, Posterior Colpocele, Prolapse of the Apex

The vaginal axis opacification is useful if properly implemented. It allows the measurement of the width of the rectovaginal space that normally does not exceed 1 cm (Bremmer 1998). It is

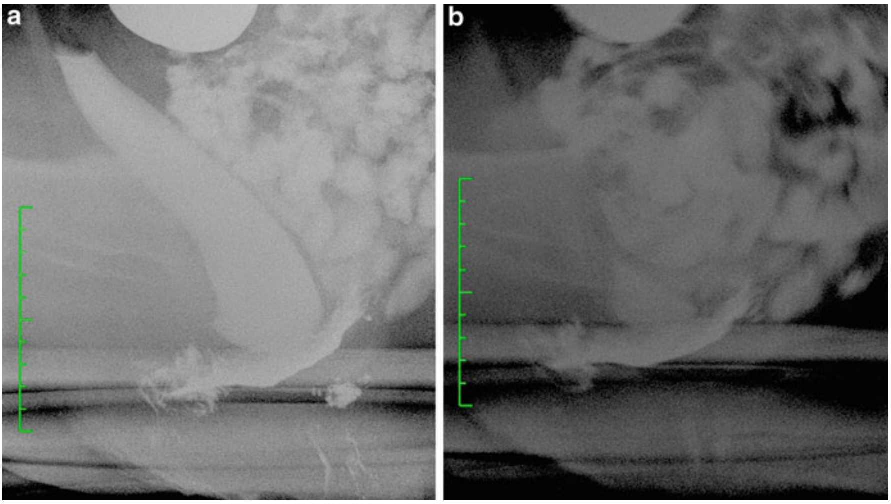

Fig. 17.18 Barium proctography. Intravaginal enterocele. After emptying the bladder, under straining, the entry of small bowel loops into an anterior colpocele is observed (v) (**a**, **b**)

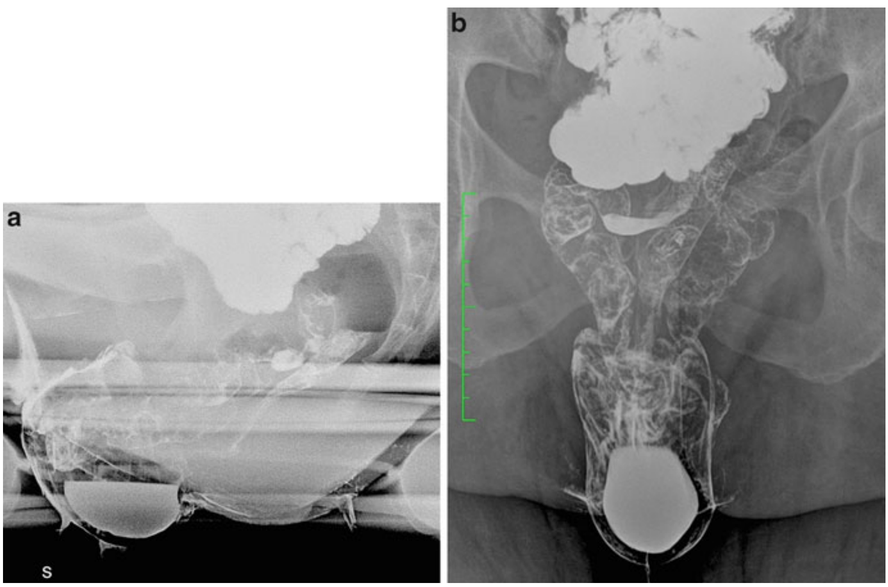

Fig. 17.19 Barium proctography. Sigmoidocele (**a**) lateral radiogram (**b**) anterior-posterior radiogram. At the end of the evacuation, after the partial emptying of the rectocele, the space in the vaginal bag, where the sigma loop enters, is created. Evidence of large posterior vaginal prolapse (*pv*)

possible to measure the vaginal apex descent and compare it with that of other pelvic organs. It's possible to perform a measurement of the vaginal length (anterior vaginal wall: nv 6–8 cm; posterior wall: 7–10 cm) (Bremmer 1998). The excessive descent of the vaginal apex and the excessive stretching of the vagina are respectively expression of damage to the apical support structures (uterosacral and cardinal ligaments) and degeneration of muscular-elastic tissue of the vaginal walls (DeLancey 1993). The anterior displacement of the vaginal axis is equivalent to the vaginal posterior prolapse (posterior colpocele) favored by the pressure exerted on the posterior vaginal wall by a rectocele, a enterocele, a sigmoidocele, or a peritoneocele (Fig. 17.7). The posterior inferior dislocation (anterior colpocele) is associated instead with a cystocele and/or an intravaginal enterocele (Low et al. 1999).

Hymen Plane

The opacification of the vaginal axis is useful to track the plane of the hymen; in this way a common framework of reference for the clinical environment and the imaging can be achieved, with the aim of measuring the position of the pelvic organs and their possible displacement. The clinical system of measurement of prolapses is POP Q; it was approved by the International Continence Society (ICS), the American Urogynecologic Society (AUGS), and the Society of Gynecologic Surgeons (Maglinte et al. 2011, 2013; Weber et al. 2001). This system uses the hymen as a reference plane to assess the position of the organs. The hymen plane in the living is located right behind the external urethral orifice. The latter is placed in the same plane of the vaginal fork represented by the anterior and posterior leaks of the contrast medium from the vagina. In the sagittal radiograms, a line, parallel to the longitudinal axis of the pubis, passing through the posterior margin of the vaginal fork, corresponds to the position of the hymen plane in the living (Figs. 17.5 and 17.7). With this transposition it is possible to measure how much a rectocele and a cystocele overflow from the hymene or are confined within it; with the same criteria the uterus lowering and the descent of an enterocele or sigmoidocele can be measured (Maglinte et al. 2011).

3.2.14 Cystocele

Cystocele is defined as the descent of the bladder base below the pubococcygeal line (Figs. 17.11, 17.18, and 17.20). It's due to a failure of the support constituted by the anterior vaginal wall. The vaginal axis shows, in cystocele, a concavity facing up (anterior colpocele) (Low et al. 1999). The magnitude of cystocele is greater after emptying the rectum, because the freed-up space allows for its maximum descent. A large cystocele can prevent the formation of an enterocele or the complete manifestation of a rectocele (Kelvin and Maglinte 1997). For this reason, it is useful to ensure the complete emptying of the bladder before beginning the study of the rectoanal region; the same is true for the rectum, when studying the anterior compartment (Maglinte et al. 2013). A cystocele typically becomes symptomatic when the bladder base has reached the hymen plane (Maglinte et al. 2013). The symptoms are difficulty to begin urination and incomplete emptying, sometimes complicated by recurrent cystitis. An initial incontinence can be masked by the progressive lowering of the cystocele, determining the folding of the urethra (kinking); this can appear after the repair of cystocele that involves the resolution of the kinking; it can be prevented by anti-incontinence procedures (Maglinte et al. 2013). There is a close relationship between cystocele and descent of the vaginal apex that has to be taken into account in the repair of the cystocele. The bladder neck hypermobility occurs only when the urethrovesical junction, under pressure, descends more than 1 cm compared to the resting position (Maglinte et al. 2011).

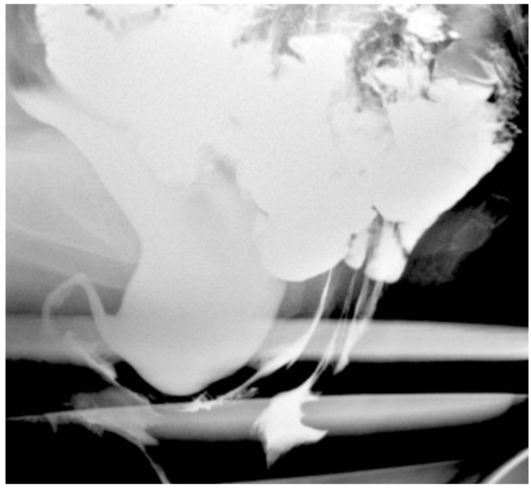

Fig. 17.20 Barium proctography. Cystoproctogram obtained at the end of rectal evacuation. The lateral radiogram shows a moderate cystocele with urethral kinking

Radiographic findings of postoperative double stapled transanal rectal resection (STARR) (Fig. 17.21):

The following radiographic signs after STARR surgery have been defined:

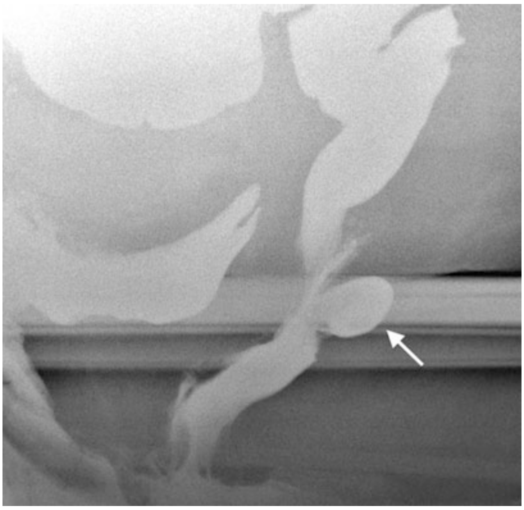

Fig. 17.21 Barium proctography. Outcomes of STARR (stapled transanal rectal resection). Lateral radiogram. The posterior rectal diverticulum (*white arrow*) is observable during evacuation at the anastomotic suture. The latter is identifiable by the reduction in caliber of the rectum at the same level

- Size of the residual rectocele, if present
- Any residual intussusception, if present
- Distance of the suture line from the anorectal junction
- Diameter of the lumen at the level of the rectal suture
- Caudal-cranial dislocation of the anorectal junction

The distance between anorectal junction and the suture line ranges between 4 and 8 cm. The dislocation of the ARJ compared to the radiological examination pre-intervention varies from 1 to 4 cm. Anterior and/or posterior diverticular formations at the level of the anastomosis may be present (Grassi et al. 2005) (Fig. 17.21).

4 Exposure to Ionizing Radiation

SIRM (Italian Society of Medical Radiology), in line with the national guidelines, inserts the barium proctography in class III, which includes exposure to an effective dose between 5 and 10 mSV (SIRM et al. 2004). The International Commission on Radiological Protection (ICRP), by analyzing the epidemiological data related to irradiated populations, derived the approximate risk of cancer induced by ionizing radiation. The conclusions are that the value of the increase in fatal cancer extended to the life of a person of average age is about 5 % for a whole-body dose of 1 Sv (1000 mSV), much higher than that to which most of the diagnostic procedures expose. A statistically significant increase in cancer has not been detected in populations exposed to doses below 50 mSV (ICRP SG2 2001).

5 Dynamic Pelvic Floor MRI (or MR Proctography or Dynamic MR)

The dynamic pelvic floor MRI is an excellent technique for studying the anatomy of the pelvic organs and the surrounding soft tissues. It provides a view of the three pelvic floor compartments and the identification of all its possible prolapses. It detects even levator ani alterations (from focal attenuation to partial and total avulsion), the distortion of the vaginal configuration, the scarring of the rectovaginal septum, the thickening and partial obliteration of the vaginal lumen, fluid collections in the hollow of Douglas, the discontinuity of urethral and paraurethral support structures, and pudendal nerve disorders (Piloni et al. 2013; Faucheron et al. 2014).

5.1 Preparation

The preparation for the dynamic pelvic floor MRI includes

- Cleaning of the rectum before the exam (Dulcolax suppository an hour before)
- Patient lying supine with bent legs
- Distension of the rectum with 300–350 cc of ultrasound gel
  - Vaginal distension with 20 cc of US gel
  - No opacification of the bladder due to its natural contrast
  - No opacification of the small intestine due to its anatomical resolution

5.2 Technical aspects

The pelvic floor is initially examined at rest, during contraction and straining, using static and dynamic sequences. The anatomy of the pelvic floor is studied through high-resolution static sequences at rest on the axial, sagittal, and coronal planes. These provide images of soft tissue with good contrast that allow for a proper observation of fascial and muscular damages and alterations of the pelvic organs. It's possible to draw the pubococcogeal line, to measure its distance from the anorectal junction and the anorectal angle, and to draw the hymen plane for comparison with the clinical evaluation of prolapses (Maccioni 2013; Silva and Maglinte 2013).

The contraction phase, the straining phase, and the evacuation phase (followed by a postdefecation sequence at rest and during maximum effort) are studied with dynamic FSE (fast spin-echo) sequences on a sagittal plane (including pubic symphysis, coccyx, urethra, vagina, rectum, and anal canal).

In the evacuation phase the descent of the bladder floor, of the uterus, of the vaginal apex, and of the anorectal junction and the depth of the rectoceles are measured; the morphology of the intussusception, the emptying mode and times, and the formation of enteroceles/sigmoidoceles are observed.

A possible further increase in the descent of the organs can be evaluated during the postevacuative push. The patient is also asked to perform any maneuver (perineal lifting, digital vaginal maneuver, digital anal maneuver) that usually occurs at home, to complete the evacuation. The post-evacuative residue is estimated and, if urination was possible, the bladder residue. Some authors repeat sequences under maximum stress after bladder catheterization and additional rectum emptying into the toilet. The purpose is to highlight and stage the enteroceles and the sigmoidoceles in their maximum expression (Silva and Maglinte 2013).

5.3 Gradation of Prolapses

The gradation of prolapse in reference to the pubococcigeal line follows the rule of three. The organ prolapse below the pubococcigeal line with a value of 3 cm or less is defined mild, 3–6 cm is considered moderate, more than 6 is defined severe (Kelvin and Maglinte 2000).

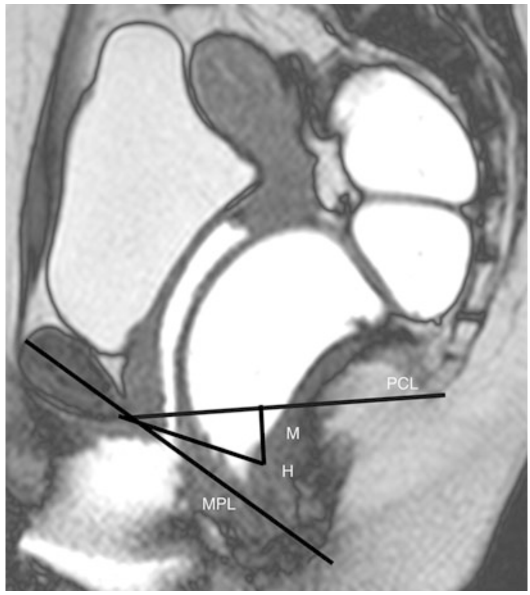

Fig. 17.22 MR proctography. Sagittal image at rest T2 weighted. Reference lines used to evaluate the descent of the pelvic organs and the anorectal junction (*PCL* pubococcygeal line, *MPL* middle-pubic line; hymenal line); H line: it corresponds to the plane of the levators and measures the hiatus of the levator ani; line M: it measures the distance between the PCL and the H line

Other lines of reference adopted in MR are (Fig. 17.22)

Line H: it extends from the bottom edge of the pubic symphysis to the anorectal junction; it is the amplitude of the diaphragmatic hiatus; normally it does not exceed 5 cm.

Line M: it is the hiatal descent and extends perpendicularly from the pubococcigeal line to distal ends of the H line.

In reference to the H line, any descent of an organ under said line is considered prolapse; it is measured as the shorter distance of the most caudal of the organs to H line during maximum thrust. It is considered as small or grade 1 if it extends up to 2 cm below the line H, moderate or grade 2 if ranging from 2 to 4 cm, and significant or grade 3 if >4 cm (Broekhuis et al. 2009; Comiter et al. 1999).

A further reference line was introduced by Singh (Singh et al. 2001).

It's the MPL (medium-pubic line) or hymenal line. It extends along the longitudinal axis of the pubis passing through the average equatorial plane. In corpses, it corresponds to the hymen plane. This is not true in the living, where the hymen plane is placed under the pubis.

The different lines were introduced with the intention to develop a measurement system comparable with the clinical evaluation of prolapses.

The few studies that have effectively compared the physical examination with measurement of dynamic MR prolapse showed that, regardless of the selected line, the evaluations significantly agree about the anterior and the medium compartment; the agreement between the two methods about the posterior compartment is lower.

The pubococcigeal line has the advantage of being the more widely used (Broekhuis et al. 2009).

5.4 Advantages and Disadvantages of MR

The advantages of MR in the study of pelvic floor dysfunctions include minimal invasiveness; multiplanar features and capacity to study more compartments; opportunity to study support structures, uterus, omentum, and pudendal nerves; and absence of ionizing radiation (Maccioni 2013).

The disadvantages are high costs, limited available equipment, performance of the examination in supine position, and the meshes not being displayed (Dietz and Cartmill 2013).

The disadvantage of the supine position does not exist in MR equipments that allow to perform the evacuation in a sitting position. However their number is very limited and they are very expensive (Hilfiker 2002).

On the other hand, defecography (or barium proctography) has the following advantages: the sitting position, the low cost, and the greater availability of the equipment. Its disadvantages are the greater invasiveness, the study being limited to the organs and not including the support structures, and the exposure to ionizing radiation.

The sitting position, a gravitational position allowing the maximum relaxation of the pelvic musculature, is the greatest advantage of classical defecography compared to MR proctography. Consequently, MR proctography has less sensitivity in the intussusception demonstration; an underestimation of the size of cystoceles, rectoceles, and enteroceles; less sensitivity in the diagnosis of the descending perineum syndrome; and greater difficulty in completing the rectal emptying that would best display the presence and depth of intussusceptions and enteroceles (Maglinte et al. 2013; Piloni et al. 1993; Silva and Maglinte 2013).

Because of the limitations associated with the supine position, some operators have returned to barium proctography after the acquisition of extensive experience with the MR proctography. The choice of a traditional defecography is reserved to those cases that require the evaluation of defecatory disorders; after the barium proctography, the opportunity to study the levator ani and other support structures with MR is evaluated. MR is used as a first approach where the clinic involvement is predominant with regard to the anterior and middle compartment or where the exposure to ionizing radiation in not advised (e.g., women in fertile age) (Maglinte et al. 2011, 2013; Silva and Maglinte 2013).

6 Relationship Between Imaging and Pelvic Floor Surgery

Despite their limits, both traditional defecography and dynamic MRI can positively influence treatment decisions in anorectal dysfunctions and entire pelvic floor prolapses. On the other hand, the relationships between physiological and anatomical causes are complex and in many ways still unknown, especially in posterior pelvic floor dysfunctions; a confirmation of this is the evidence that the anatomical correction of a structural alteration (e.g., rectocele, intussusception) does not necessarily restore the function (Altomare and

Giuratrabocchetta 2011; Van Dam et al. 1997; Ott et al. 1994; Harvey et al. 1999; Hetzer et al. 2006; Jayne and Finan 2005; Barber et al. 2009).

In the surgical field, it is difficult to transpose the whole mass of information provided by the imaging because of the lack of intraoperative verification criteria of everything that it describes, of the insufficient knowledge of the pathophysiological mechanisms, but also because of the specialized compartmentalization of the pelvic floor: urethra and bladder are in the exclusive competence of urologists, vagina and reproductive organs in the exclusive competence of gynecologists, and colon and rectum under the management of gastroenterologists and colorectal surgeons (Wall and DeLancey 1991; De Lancey 2001). This separation of responsibilities, which is still widespread, with the exception of a few centers dedicated to the entire pelvic floor, contributes to the underestimation of the relationships between the various sectors and, together with the other reasons stated above, the absence of gold-standard interventions for the alterations of the posterior compartment (Jayne and Finan 2005). In light of these considerations, surgery should be considered as the last option for obstructed defecation, to be taken into account only in selected cases that did not respond to any conservative treatment (Altomare and Giuratrabocchetta 2011).

7 MR of the Levator Ani

The levator ani muscle consists of three main components: pubococcigeus, ileococcigeus, and puborectalis (Fig. 17.23a, b). Each component can be examined with ultrasound and MR (Lawson 1974; Margulies et al. 2006; Shobeiri et al. 2009).

The puboccigeus muscle is medially located to the puborectalis, originates near the superior pubic ramus, and is part of the vagina, the anus, and the perineal body. The puborectalis muscle originates in proximity of the inferior pubic ramus, crosses the anorectal junction to the cranial external anal sphincter, and continues on, inserting near the inferior pubic ramus on the opposite side. It is located laterally to the ileococcigeus and the pubococcygeus (Kearney et al. 2004). The pubococcygeus and puborectalis appear as a single body on the side of the vagina.

The puborectalis muscle forms the anorectal angle and closes the urogenital hiatus, creating a high-pressure zone in the vagina. The ileococcigeus muscle originates from the tendon arch of the levator ani (a condensation of the bundle of the internal obturator muscle). It inserts on each side on the ileococcigeus raphe located along the median line that connects the sacrum

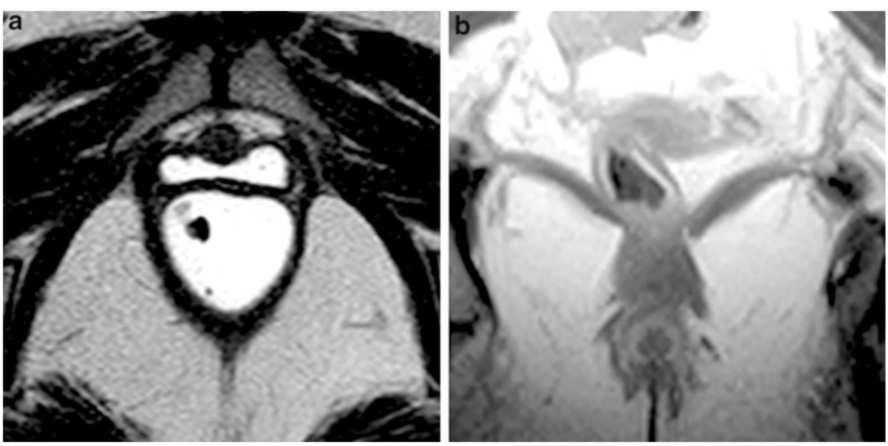

Fig. 17.23 MR proctography. Axial (**a**) and coronal (**b**) image T2 weighted at rest: (**a**) normal aspect of the puborectalis muscle originating in the pubic symphysis (*sp*) (**b**) normal ileococcigeus muscle (*ic*) originating laterally to the obturator internus muscle (*o*)

Fig. 17.24 MR proctography. Axial image at rest T2 weighted. Unilateral right defect in individual with a normal contralateral pubococcigeus muscle. The pubococcygeus right bundle looks hypotrophic, with a partial tear in the vicinity of the pubic insertion

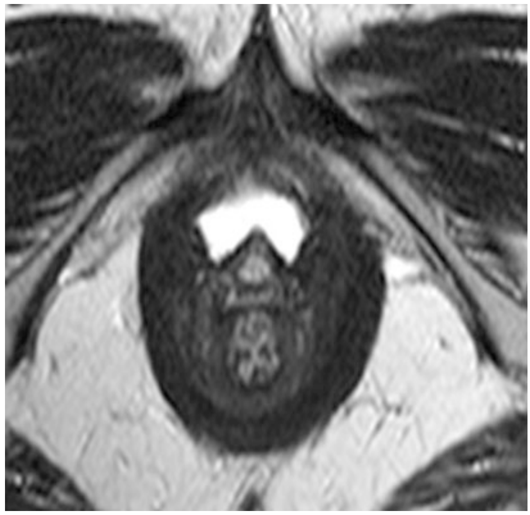

Fig. 17.25 MR proctography. Axial image at rest T2 weighted. Hypertrophy in compensation of the puborectalis dorsal portion in patient with damage to the ventral portion of the puboccigeus muscle

and the coccyx. In coronal scans it is well appreciated and is located in the dorsal part of the pelvis. In the sagittal scans it has a shelf-like appearance for which it is referred to as levator plate (Margulies et al. 2006).

The weight of the abdominal and pelvic organs and the increase of the abdominal pressure are the forces that act on the pelvic floor; the muscles and the endopelvic fascia resist to them. If the load exceeds the resistance of the pelvic tissues of support, a prolapse takes place. The women with levator damage experience higher failure rates with pelvic floor surgery (De Lancey and Hurd 1998; De Lancey et al. 2003).

The most frequent cause of damage to the levator ani are traumas at childbirth (De Lancey et al. 2003).

In these injuries the most frequently affected muscle is the pubococcygeus (Fig. 17.24). The ileococcigeus follows, and lastly the puborectalis.

It was found that when the anterior portion of the levator ani is damaged, there is a compensating hypertrophy of the posterior portion, in particular that of the puborectalis (Fig. 17.25). This hypertrophy explains why a loss of muscle tissue below 50 % is not significantly associated with organ prolapse; instead this happens in case of a loss above 50 %, because the ability of compensation of the remaining muscle is surpassed (De Lancey et al. 2012).

The muscle damage was gradated with a score that considers the left and right beam of the pubococcygeus muscle separately. The score is equal to 0 in the absence of defects, 1 if less than half of the muscle bundle is damaged, 2 if more than half is damaged but the damage is not complete, and 3 in case of a full loss of the muscle. The total score is given by the sum of the scores of each side. The scores ranging from 4 to 6 are the most serious; the serious scores include the complete unilateral loss of the muscle bundle (Berger et al. 2014).

A woman without alteration of the levator ani has a chance of less than 2 % to form a pelvic prolapse; this probability increases to 30 % in women with complete loss of muscle. Women with anterior and/or posterior colpocele show, compared to controls, a greater descent of the pelvic floor in the thrust phase, a greater expansion of the hiatus of the levators and of the urogenital hiatus, and a wider angle of the plane of the levators (all indirect signs of failure of the levator ani) (De Lancey and Hurd 1998; Hsu et al. 2006; Chen et al. 2006; Lewicky-Gaupp et al. 2010).

8 Anal Incontinence and Imaging

Anal incontinence (inability to control the anal muscles, with involuntary loss of stools and/or gas) is a multifactorial disease that can be associated with other alterations of the evacuation (Bharucha 2004).

Traditional defecography and dynamic MR may show signs of incontinence and of the morphofunctional alterations associated with it (Terra and Stoker 2006; Hetzer et al. 2006; Nielsen et al. 1993; Brennan et al. 2008). The loss of contrast medium at rest indicates internal sphincter damage; its loss while coughing (stress incontinence) can be due to alterations of the external sphincter (Pemberton 1990). Other signs that may be present are length of the anal canal at rest below 2 cm, evacuative opening of the anal canal above 2 cm, and excessive amplitude of the ARA (Piloni et al. 1993). Associated structural alterations can be rectocele, intussusception, or external rectal prolapse. The mechanism involved in intussusception is believed to be an intermittent activation of the rectoanal inhibitory reflex The external rectal prolapse, in addition to activating this reflex, causes a chronic dilation of the sphincter because of the external displacement of the rectum (Pfeifer et al. 1996; Harmston et al. 2011). With the imaging it's possible to detect the presence of an enterocele that, by wedging into the intussusception, favors its descent, thus increasing the damage to the anal muscles.

Incontinence can coexist with obstructed defecation syndrome. It is known that the descending perineum syndrome is initially characterized by predominant symptoms of evacuative difficulty followed sometimes, because of the straining and the consequent chronic pudendal neuropathy, by incontinence, that can become predominant (Pemberton 1990).

Rectocele alone is not responsible for anal incontinence, unless it is associated with pudendal nerve entrapment or injury of the sphincter. In these cases, there is prior history of obstructed defecation and/or childbirth trauma (Parks et al. 1966; Hetzer et al. 2006).

MR by intraluminal coils is a valid method in the diagnosis of the lesions of the anal sphincters (Terra and Stoker 2006).

Compared to endoanal ultrasound, MR seems to be less accurate in the diagnosis of lesions of the internal anal sphincter. Thanks to the large difference in contrast with the surrounding adipose tissue, MR is the most indicated technique for the diagnosis of atrophy of the external anal sphincter (Dobben et al. 2007).

9 Ultrasound of the Pelvic Floor

The approach to pelvic floor ultrasound can be perineal, endovaginal, or transanal (Kleinubing et al. 2000; Sarnelli et al. 2003; Santoro et al. 2011). The less invasive approach is the perineal ultrasound. It is practiced by placing the probe between the vulvar rhyme and the perineum. A convex probe, between 3 and 6 MHz and with a field of vision of at least 80°, is used (Santoro et al. 2011). The probe is rotated in order to view the bladder, the vagina, the distal part of the rectum, the anal canal, and the puborectalis muscle on a sagittal plane; the latter presents itself as a hyperechoic area with convex anterior surface in close contact with the external anal sphincter and the anorectal junction. The convexity of this area is evaluated at rest, during voluntary contraction and expulsive thrust. Its increase during the voluntary contraction and its reduction in the expulsion phase are considered as indexes of normal puborectalis muscle function. A quantitative evaluation of the activities of this muscle can be made by measuring the angle formed by the tangents at the edge of the two beams of the puborectalis sling. This angle corresponds, in location and meaning, to the one described for the defecography (Viscardi et al. 2012). All of this can be displayed using the 2D technique. The use of 3D and 4D techniques, routinely used in obstetrics, allows obtaining tomographic images that provide an overview of the entire puborectalis muscle, including its pubic insertion

(Santoro et al. 2011). It is possible in this way to measure the diameter and the area of the hiatus of the levator ani, to determine the degree of the hiatal distension in the Valsalva maneuver, and to obtain a real-time acquisition of the images (Dietz 2004a, b). The introital ultrasonography is a variant of the perineal ultrasonography. In the introital access, the probe rests on the vaginal entrance without passing the hymen's plane. The main difference with the perineal approach is represented by the fact that the used probe is an "endfire" endocavitary transducer, with a microconvex surface with short radius of curvature and wide viewing angle (Sarnelli et al. 2003; Santoro et al. 2011).

Vaginal access involves placing the transducer inside the vaginal cavity. This position represents a factor that interferes with the dynamic assessment of the organs, because it limits their mobility (Sarnelli et al. 2003; Santoro et al. 2011). The transrectal access takes place with mechanical or electronic 360° rotational probes. This method allows for the optimal evaluation of the thickness and the structure of the internal and external anal sphincter. It is the gold standard technique to study the morphology of the anal canal, the evaluation of which is of primary importance in fecal incontinence (Santoro et al. 2011).

10 Levator Ani Injuries

The study of the levator ani is carried out with perineal or endovaginal approach using the 3D technique that allows a complete view of the muscle (Dietz 2004b). The 4D technique also allows a dynamic evaluation. With these techniques it's possible to detect major muscle traumas in the same way as with MR. The complete avulsion is distinguished from the partial. An avulsion is intended as the disconnection of the muscle from its location on the branches of the pubis. The complete avulsion is diagnosed when the slice identified by the plane of minimum hiatal size and the two slices that are immediately cranial to it show an avulsion of the muscle. If only one slice shows any abnormality, there is the framework of the partial avulsion (Dietz 2007).

The effect of the avulsion is a widening of the hiatus of the levators. A widening of the hiatus above 25 cm^2 during the Valsalva maneuver is defined as pathological (Dietz and Steensma 2005a). The degree of relaxation is closely related to the severity of the prolapse and its clinical manifestations.

A highly significant relation between the intussusception and the increase of the hiatal area of levator ani during the Valsalva maneuver has been found (Rodrigo et al. 2011). It is likely that structural or functional abnormalities of the levator ani have a role in the pathogenesis of the intussusception.

11 Ultrasound Assessment of the Anterior Compartment

The perineal ultrasound proved to be a valuable technique for the study of the anterior compartment, to detect the presence of a cystocele (and to make an assessment of the urethra, which presents itself like a linear hypoechogenic image with an anterior concavity (Dietz 2008)) (Fig. 17.26). An evaluation of its mobility is useful because the

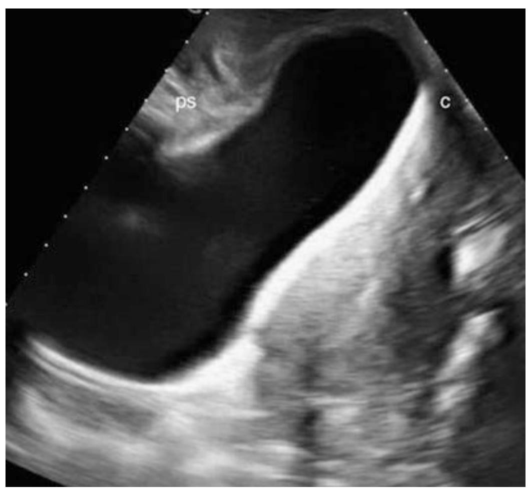

Fig. 17.26 Transperineal ultrasound. Exam performed with convex 2D probe with natural contrast (air and feces). Sagittal scan during straining. Moderate cystocele: bladder floor located 5 cm below the lower border of the pubic symphysis (*ps*); mild hysterocele: the cervix (c) descends at the level of the pubic symphysis

hypermobility of the urethra is connected to stress-related urinary incontinence. The urethral mobility by definition is the measurement of the descent of the bladder neck during the Valsalva maneuver, starting from the condition of rest (≤10 mm). The average value in women with stress-related incontinence is around 30 mm. In hypermobility the retrovesical angle opens beyond its normal limit of 120° (Dietz 2008). The cystocele with abnormal retrovesical angle is probably associated with a central defect of the endopelvic fascia (Dietz 2008).

A cystocele may present itself with a normal retrovesical angle. This second type of cystocele is frequently seen in patients with prolapse and continents (Dietz 2008). It seems to be particularly common in women with abnormalities of the levator ani; the implication of this observation is that cystoceles with normal retrovesical angle can be due to lack of paravaginal support (Dietz 2008).

Ultrasound assessment of the anterior compartment seems to agree with clinical prolapse assessment (Dietz 2008).

12 Ultrasound Assessment of the Central Compartment

The evaluation of the descent of the uterus and, after hysterectomy, of the vaginal vault is possible if a rectocele or enterocele do not obstruct their visibility. The ultrasound measurement of their descent seems to agree with the clinical measurement.

13 Ultrasound Assessment of the Posterior Compartment

The ultrasound assessment of the posterior pelvic floor is more recent. In the literature there are studies comparing it to barium proctography (Viscardi et al. 2012; Beer-Gabel et al. 2004; Grasso et al. 2007; Steensma et al. 2010; Perniola et al. 2008; Regadas et al. 2011). These studies reflect the limitations imposed by the lack of standardization of the technique. Some authors perform an examination of the rectum after filling it with contrast medium consisting of ultrasound gel; others use air and/or naturally present feces (Fig. 17.27) (Beer-Gabel et al. 2004; Grasso et al. 2007; Steensma et al. 2010; Perniola et al. 2008; Regadas et al. 2011). Some distinguish the true rectocele from the perineal hypermobility. They define the true rectocele as a herniation of the rectum through a discontinuity in the anterior contour of the anorectal musculature. This herniation must have a depth of no less than 10 mm. The perineal hypermobility is defined instead as a dislocation of the content of the ampoule (hyperechoic feces or air) under a reference line passing through the bottom margin of the pubic symphysis, without any detectable discontinuity of the anorectal muscle (Dietz and Steensma 2005b). Other authors consider any herniation of the anterior wall of the rectum as a rectocele, regardless of the presence of discontinuities (Beer-Gabel et al. 2004; Grasso et al. 2007; Perniola et al. 2008) (Fig. 17.28).

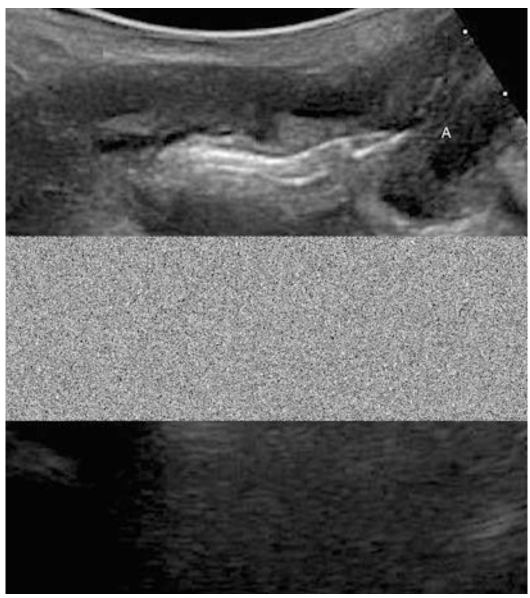

Fig. 17.27 Transperineal ultrasound. Exam performed with convex 2D probe with natural contrast (air and feces). Sagittal scan during straining. Rectorectal intussusception: the rectal walls fold on themselves, entering into the lumen of the rectum; the apex of the intussusceptum facing the anal canal can be observed

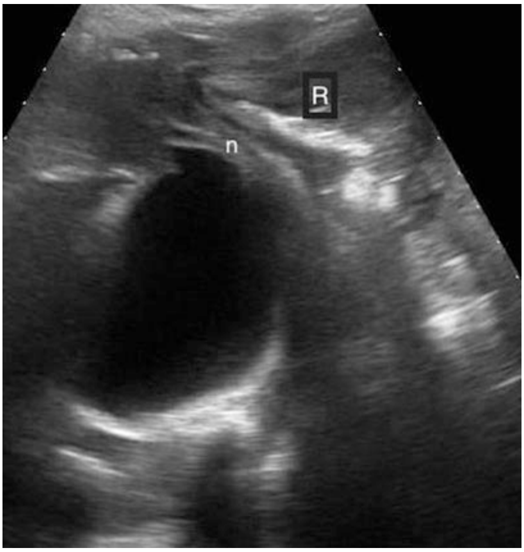

Fig. 17.28 Transperineal ultrasound. Exam performed with convex 2D probe with natural contrast (air and feces). Sagittal scan during straining. Large rectocele located under the floor of the bladder. Bladder neck hypermobility (*n*): the bladder neck descends more than 1 cm below the resting position

Fig. 17.29 Transperineal ultrasound. Exam performed with convex 2D probe with natural contrast (air and feces). Sagittal scan during straining. Enterocele (*E*) that largely fills the rectovaginal space (*rvs*). The contour of the wall of an intestinal loop is visible (*s*)

For some authors, the enterocele is the dislocation of the abdominal contents (omentum or bowel loops, whether mixed or not with intraperitoneal liquid) beyond the abovementioned line (Dietz and Steensma 2005b). Others define the enterocele as the descent between the rectum and the vagina of bowel loops previously contrasted with gastrographin (Beer-Gabel et al. 2004) (Fig. 17.29).

In studies comparing the ultrasound technique and defecography the correlation is greater when the survey is preceded by the introduction of contrast media inside the rectal ampoule (Beer-Gabel et al. 2004). Typically, the more advanced the posterior pelvic floor disorders are, the greater the correlation between the two techniques is.

Currently the transperineal ultrasound can act as a useful method of initial approach to the patients with clinical signs and symptoms of pelvic dysfunction, since it provides an overview and a dynamic assessment of the pelvic floor. It can be complemented by defecography whenever a more accurate evaluation in patients with evacuative difficulty is required (Viscardi et al. 2012).

14 Use of Ultrasound in the Assessment of Meshes

Pelvic ultrasonography is the only method currently available to highlight the propylene meshes used in the surgery of the pelvic prolapse. The reason for this exclusivity is due to the fact that only the ultrasound shows such meshes thanks to their hyperechogenicity. Partial or complete detachments of the meshes can be easily diagnosed (Dietz 2012).

15 Colonic Transit Times

The study of the transit times allows to identify the presence and the location of a possible slowing of the colonic transit (Corazziari 2013). The mode of transit can be classified in four types:

1. Normal intestinal transit
2. Colonic slowing
3. Rectal slowing
4. Rectocolonic slowing

Fig. 17.30 Assessment by radiopaque markers. Seven days after the administration, no marker was expelled, and all markers are stationed inside the ascending colon (slow colonic transit)

The measurement of the transit times requires the oral administration to the patient of radiopaque markers and the subsequent execution of direct radiograms of the abdomen to count the residual markers (Rao and Meduri 2011). The persistence of the markers in a quantity above the established maximum standard limit indicates slow transit; the place, or places, of storage allow to identify whether the slowing is local or global (Corazziari 2013) (Fig. 17.30). Several methods have been proposed but with very different results because of the variability of the transit in normal subjects, the diversity of the used methodologies, and the difficulty in differentiating the subtypes of constipation. One example is the predominant localization in the rectosigmoidal segment; it is suggestive but not diagnostic of obstructed defecation. The delayed rectosigmoidal transit from dyssynergia, in fact, can slow the transit down in the proximal colon, thus determining a broader distribution of the markers in 60 % of the patients with dyssynergia. In these patients, the entire transit improves with therapy; hence the importance of excluding this diagnosis, before concluding for constipation due to slow colonic transit (Rao and Meduri 2011). Apart from the differentiation between subtypes of constipation, a simple method to evaluate the colonic transit is the one by Hinton et al. (1969). A gelatin capsule containing 24 radiopaque markers is administered and a direct examination of the abdomen performed after 5 days. Normally, less than five markers remain in the colon at this time.

16 Summary

Imaging techniques used for the study of colorectoanal dysfunctions include barium proctography, MR proctography, and time studies of colonic transit. Ultrasonography of the pelvic floor, transperineal or introital, can be considered a method of initial approach that can integrate the barium proctography with the study of the anterior pelvic floor. The advantage of barium proctography with opacification of the bowel, vagina, and rectum, compared to MR proctography, is the sitting position that allows the full expression of the pelvic floor dysfunctions, especially those related to the posterior compartment. The advantages of MR is the multiplanar and multicompartmental study of the pelvic floor, including the support structures, and the absence of ionizing radiation. It is preferable if the main clinical indication is urogynecological. The colonic transit time assessment is indicated if slow transit constipation is suspected, once the obstructed defecation has been ruled out.

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Ultrasound of the Colon and Rectum: Procedures and Indications

18

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Contents

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Abstract

This chapter demonstrates the technique of 2-D and 3-D anorectal ultrasound to evaluate the benign and malignant diseases of the anal canal and rectum. It discusses precisely the anatomic configuration of the anal canal and rectum and mentions the indications of ultrasound. This method evaluate benign disease such as *fecal incontinence*, demonstrating sphincter and pubovisceral muscles defects and the extension of the injury; *abscess and anal fistula*, identifying the primary and secondary tracts, the exact position of the internal opening in relation to the anal margin and adjacent cavities, classifying the entire extension of the fistulous tract and its relation to the sphincter muscles, and the percentage of sphincter muscle to be transected during the surgical procedure. Regarding to *malignant disease*, it is an important method for evaluating malignant rectal and anal canal tumors as it allows quantifying the extent of tumor invasion into the rectal layers, sphincter muscles, and adjacent tissues and to identify compromised lymph nodes, making the exam as the mainstay for evaluating response to chemoradiotherapy. The rectal US is very effective to follow tumors after chemoradiotherapy and constituting an examination of great importance to choose the best therapeutic option. The *dynamic 3-D ultrasound* is currently used in many centers as the first option for evaluation of the anatomic and functional disorders of the posterior and middle compartment of pelvic floor.

Anorectal ultrasonography has been part of the pretreatment assessment of the benign and malignant diseases of the anal canal and rectum (Deen et al. 1993; Cheong et al. 1993; Gavioli et al. 2000; Kim et al. 2002). The detailed anal canal anatomy and rectal wall layers can be clearly identified detecting abnormalities using this method of imaging. The increasing availability of ultrasound in the clinical setting has renewed interest in using this modality for the assessment of pelvic floor anatomy and dysfunctions. Many types of ultrasound probes have been developed and different approaches are used to assess the pelvic floor dysfunctions, such as the transanal, transperineal/translabial, and endovaginal, with 2-D, 3-D, and 4-D overview.

1 Types of Ultrasound Approaches to Assess the Anorectal and Pelvic Floor Diseases

It is recommended cleaning the rectum with a rectal enema (2 h prior the scanning) and perform a digital rectal examination for the assessment of anal canal, rectum, and pelvic floor organs.

1.1 Transperineal/Translabial Ultrasound

The exam is performed with the patient placed in the dorsal lithotomy position, with hips flexed and abducted.

The transperineal ultrasound is performed with the transducer positioned on the perineal area while the translabial with the probe kept between the labia. Images are acquired with the patient at rest, during maximal Valsalva maneuver and during pelvic floor muscle contraction (Dietz and Steensma 2005; Dietz et al. 2005). All perineal structures can be assessed in an acceptable extent in real-time using 2-D, 3-D, and 4-D modalities.

1.2 Endoanal/Endorectal Ultrasound

Patients are examined in the left lateral position with their knees and hips flexed.

Equipment used for endoluminal ultrasonography includes an endocavitary probe with rotating transducer which acquires a 360° images with 2-D and 3-D overview. The 3D modality is performed with 360° rotating anorectal transducer, high frequency (between 6 and 16 MHz), focal distance between 2.8 and 6.2 cm, and automatic image acquisition without manual movement of the transducer. Images up to 6.0 cm long are captured along the proximal-distal axis during up to 55 s by moving two crystals on the extremity of the transducer. The images are acquired as a series of transaxial microsections up to 0.20 mm thick, producing consequently a high-resolution digitalized volumetric image (cube). The 3-D volume can be saved, exported, reviewed, and manipulated, visualizing the lesions at different angles and in different planes. It is possible to have spatial information to better understand the anatomy, sequence of axial image, and the whole length of the anal canal and the distribution of the muscles.

– *Endoanal Ultrasound* – A condom containing ultrasonic gel is placed around the transducer and lubricated externally. After digital rectal examination, the transducer is then inserted into the anal canal, as far as the low rectum to scan the whole anal canal length.
– *Endorectal Ultrasound* – For rectal scanning, the transducer is used with a balloon placed around it, which is filled with degassed water. Air bubbles should be completely removed from the balloon to produce high-quality images. A rigid proctoscope is initially inserted into the rectum past the level of the tumor. Following that, the endoprobe is inserted into the rectum through the proctoscope. The balloon is filled with 40–60 mL of degassed water, adjusting to the rectal diameter and in the whole length of the scan area, providing an acoustic interface.

1.3 Doppler

Various Doppler techniques have been used to demonstrate the distribution of vascularity in benign and malignant disorders (Ogura et al. 2001; Drudi et al. 2003; Kobata et al. 2008; Miyamoto et al. 2013). Currently, a 360° color Doppler anorectal ultrasonography system is able to assess the vascularity of structures at different levels of the anal canal, rectum, and mesorectal fat. The system automatically calibrates distances and color hues as flow velocities and calculates the color pixel area and flow velocity – encoded by each pixel – inside each region of interest. The advantage of this technique is that it makes possible to measure the vascularity circumferentially instead of by quadrant. The 360° endoluminal probe clearly identifies and enables quantitative assessment of vascular parameters for inner and outer anatomic structures at upper, mid, and low levels of the anal canal, and at middle and low levels of the rectal wall and mesorectal fat.

2 Anal Canal Anatomy

The normal anal canal US images are clearly demonstrated in five layers, from inner to outer:

1. Interface of the transducer with the anal mucosal surface (hyperechoic layer)
2. Subepithelial tissues (moderately reflective) – Between the transducer and the internal border of the internal anal sphincter
3. Internal anal sphincter (IAS) (hypoechoic layer)
4. Longitudinal muscle (LM) – It is an heterogeneous muscle (hypoechoic and hyperechoic) although it is mainly smooth muscle but with conjoins with striated muscular fibers from the Levator Ani.
5. External anal sphincter (EAS) – It is described as having three parts: subcutaneous, superficial, and deep. The deepest portion is integrated with the puborectalis muscle (PR).

Due to its different conformation, the anal canal is divided into three levels, with different anatomical structures:

– **Upper anal canal:** PR, the deepest part of the EAS, LM, and the IAS (complete ring) (Fig. 18.1);
– **Middle anal canal:** Superficial part of the EAS (complete ring), LM, IAS (complete ring), and the transverse perineal muscles (Fig. 18.2). At this position, it is possible to assess the perineal body which is the central portion of the perineum (where the EAS, the

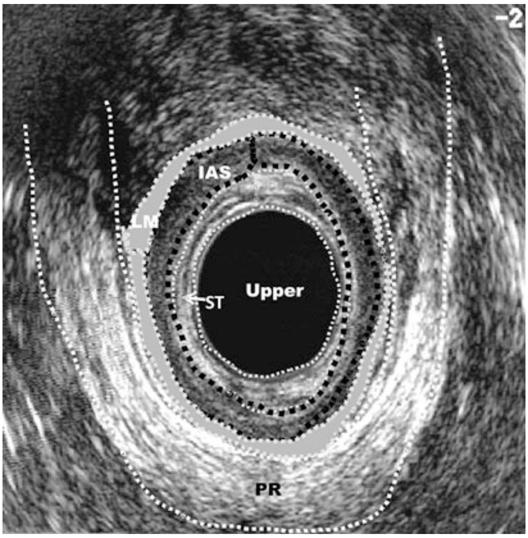

Fig. 18.1 Upper anal canal (female patient). *ST* subepithelial tissues, *IAS* internal anal sphincter, *LM* longitudinal muscle, *PR* puborectalis muscle

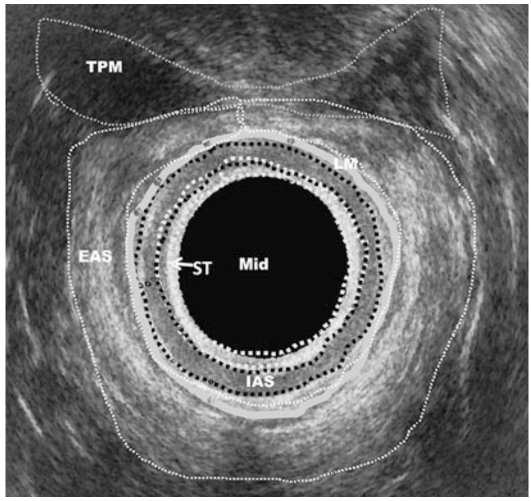

Fig. 18.2 Middle anal canal (female patient). *ST* subepithelial tissues, *IAS* internal anal sphincter, *LM* longitudinal muscle, *EAS* external anal sphincter, *TPM* transverse perineal muscles

bulbospongiosus, and the transverse perineal muscles meet). The anococcygeal raphe is also seen as a posterior hypoechoic triangle.

– **Low anal canal**: Subcutaneous part of the EAS (complete ring).

In a previous study, Regadas et al. evaluated the anal canal anatomy using 3-D modality and demonstrated the asymmetrical shape of the anal canal. The anterior anal canal starts and ends more distally, and it is formed by the external anal sphincter (EAS) and the internal anal sphincter (IAS) while the posterior anal canal starts and ends more proximally and it is included the puborectalis muscle too (Fig. 18.3). They also evaluated the gender-related differences in anal canal anatomy and the muscles lengths, demonstrating that the anterior EAS is shorter (mean = 2.2 cm) and the gap length (mean = 1.2 cm) (the area in the anterior quadrant without striated muscle, measured from the proximal edge of the posterior PR to the proximal edge of the anterior EAS) is longer in females compared with males (EAS = 3.4 cm; GAP = 0.7 cm), providing a possible explanation for the higher incidence of pelvic floor dysfunctions in women (Fig. 18.4). The posterior EAS-PR was significantly longer in men (mean = 3.6 cm) than in women (mean = 3.2 cm). The anterior and posterior IAS was significantly shorter in women than in men (Regadas et al. 2007).

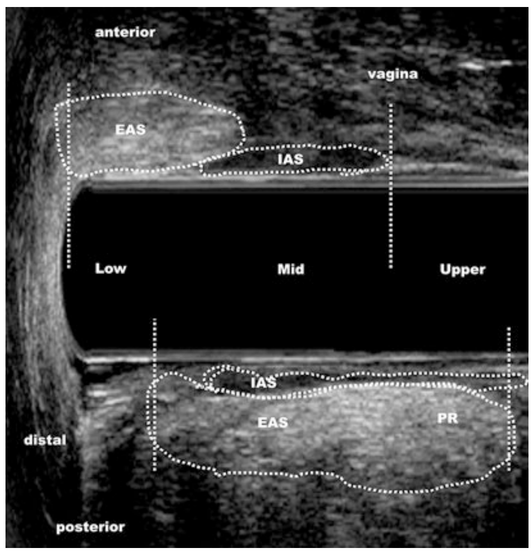

Fig. 18.3 Anatomic configuration of female anal canal (sagittal plane). *IAS* internal anal sphincter, *LM* longitudinal muscle, *EAS* external anal sphincter, *PR* puborectalis muscle

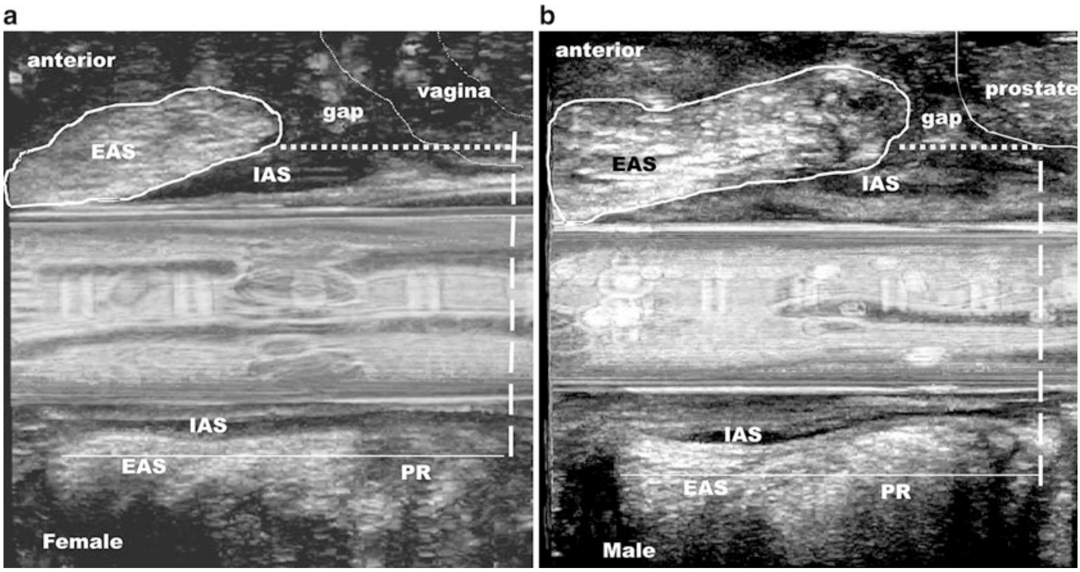

Fig. 18.4 Anatomic configuration of the anal canal, comparing Female (**a**) with male (**b**) (sagittal plane). *IAS* internal anal sphincter, *LM* longitudinal muscle, *EAS* external anal sphincter, *PR* puborectalis muscle

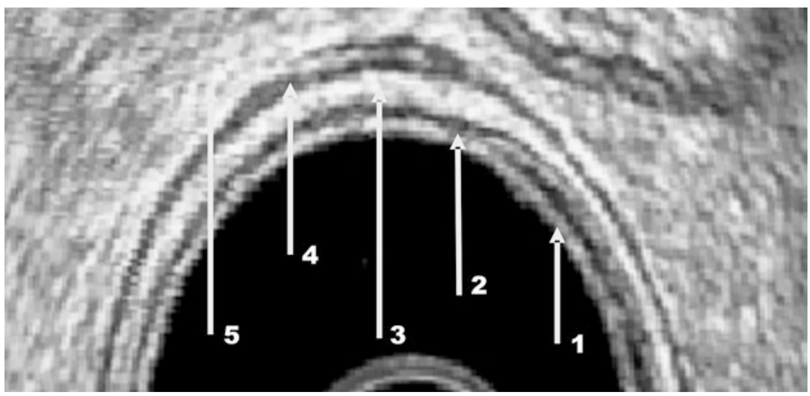

Fig. 18.5 Normal anatomy- Rectal layers *1* Mucosa (hyperechoic); *2* Muscularis mucosa (hypoechoic); *3* Submucosa (hyperechoic); *4* Muscularis propria (hypoechoic); *5* Perirectal fat (hyperechoic)

3 Rectal Anatomy

The rectal wall is made up of five layers (Fig. 18.5)

1. The first hyperechoic layer: the interface of the balloon with the rectal mucosal surface
2. Hypoechoic layer: the mucosa and muscularis mucosae
3. Hyperechoic layer: the submucosa
4. Hypoechoic layer: the muscularis propria, sometimes seen as two layers: inner circular and outer longitudinal layer

All the pelvic organs adjacent to rectum are clearly visualized, identifying its relation with the rectal wall layers in multiple anatomic planes.

4 Fecal Incontinence

The ultrasound clearly identifies the presence of sphincter defects, combined EAS and IAS or if the injury involves the EAS alone. Using a 3-D modality, additional measurements include the following: length of the anterior EAS and radial angle of the EAS defect, length of the anterior and

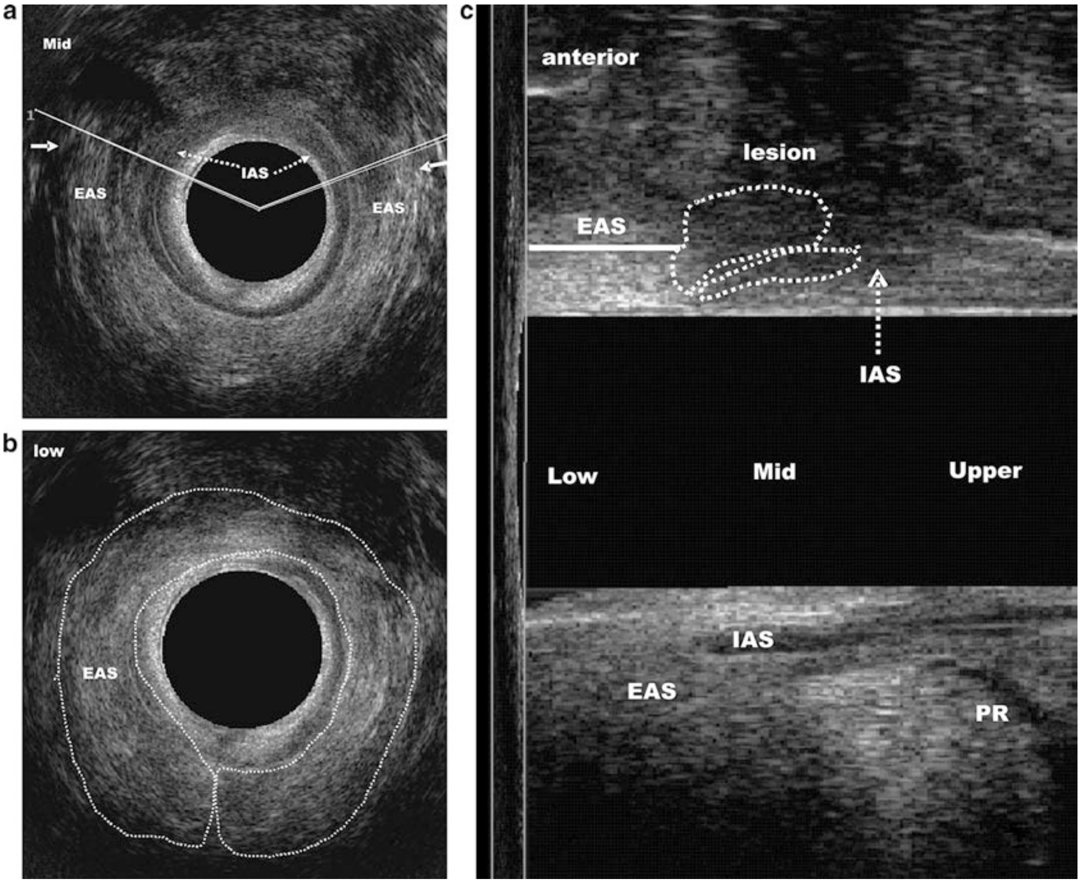

Fig. 18.6 Combined partial EAS and IAS defects (*dotted line*) in the mid anal canal after vaginal delivery: (**a**) Angle of the EAS injury, (**b**) EAS is intact in the low anal canal, and (**c**) 3D modality – measurements of the residual anterior EAS length. *IAS* internal anal sphincter, *LM* longitudinal muscle, *EAS* external anal sphincter, *PR* puborectalis muscle

posterior IAS, length of the posterior EAS plus the PR muscle (EAS + PR), and the gap length (distance from the proximal edge of the posterior PR to the proximal edge of the anterior EAS, corresponding to the area in the anterior quadrant without striated muscle). Those measurements can be correlated with incontinence score (Murad-Regadas et al. 2014a).

The endoanal ultrasound scanning identifies injured muscles and the extension of the injury in relation to the anal circumference and the length of the anal canal. The angle of the lesion is measured by drawing two lines tangentially to the injured muscle and making them converge to the center of the circumference. On ultrasound scans, muscle injuries appear as interruptions (or changes) in the echogenicity of the original musculature. Internal anal sphincter (IAS) injuries appear as lighter-colored single or multiple disruptions of the normal hypoechoic circumferential image, while external anal sphincter (EAS) injuries are characterized as areas of reduced hyperechogenicity, depending on the amount of fibrous tissue formed (Sultan et al. 1993; Felt-Bersman et al. 1995; Karoui et al. 1999; Fig. 18.6). The "septum maneuver," used routinely in the measurement of the perineal body, is helpful in the identification of sphincter injuries of the anterior quadrant as it helps view the extremities of damaged muscles. It consists of measuring the distance between the finger of the examiner held against the posterior vaginal wall and the internal border of the IAS (normal > 10.0 mm) (Zetterstrom et al. 1994; Fig. 18.7). However, this technique cannot be used clinically with patients previously submitted to perineoplasty with sphincteroplasty since the size of the perineal body in these patients exceeds 10 mm, even before muscle repair.

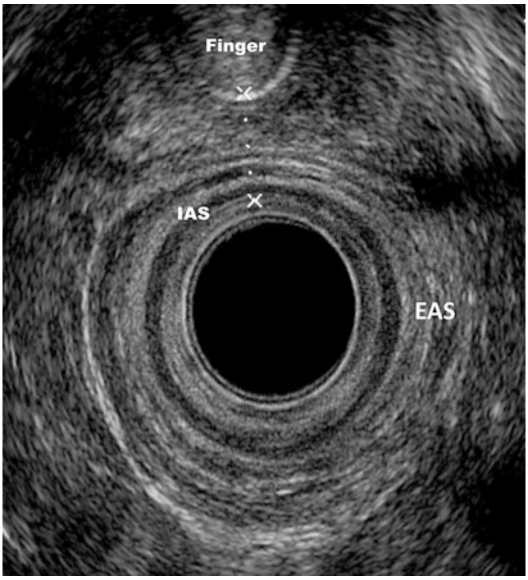

Fig. 18.7 Septum maneuver – normal perineal body thickness (>10.0 mm)

Anal ultrasonography is also particularly useful in the evaluation of the results of surgical repair of the anterior and posterior anal sphincter, identifying adjacent or overlapping muscles or documenting persistent muscle injury (Nielsen et al. 1994; Savoye-Collet et al. 1999; Fig. 18.8).

Changes in muscle thickness may correlate with atrophy and symptoms of fecal incontinence. Atrophy is characterized by generalized sphincter thinning and fatty replacement. The similar results of MRI for the evaluation of atrophy have not been found with EUS. This could probably be explained by the fact that EUS is not able to distinguish fatty infiltration from normal muscle tissue. For that purpose, MRI is the best technique to be used for detecting EAS atrophy (Briel et al. 1999; Williams et al. 2001).

It can be also used the same probe (360° rotating anorectal transducer) in the endovaginal assessment to evaluate the Levator Ani muscle defect. This muscle has a recognized role in pelvic floor–supporting structures. The nomenclature regarding the Levator Ani varies. As proposed by DeLancey, it is used the term puboviceral muscle (PVM) as synonymous with the term pubococcygeus/puborectalis since the two components can't be distinguished on imaging (DeLancey 2001; Fig. 18.9). Anatomic alteration of the insertion of the PVM has been clearly

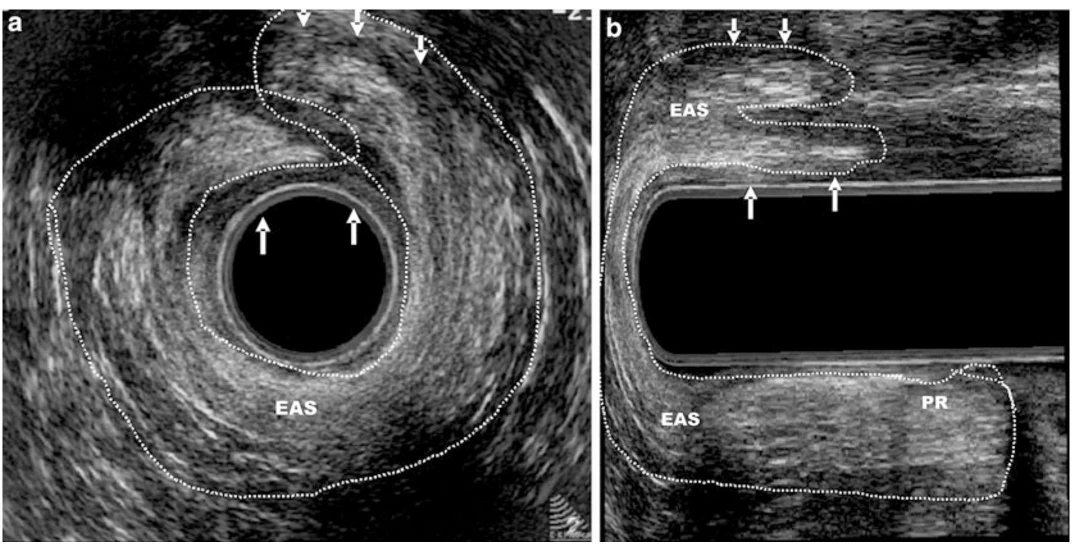

Fig. 18.8 Anterior sphincter repair – overlapping (*dotted line*) (**a**) Axial/ (**b**) Sagittal

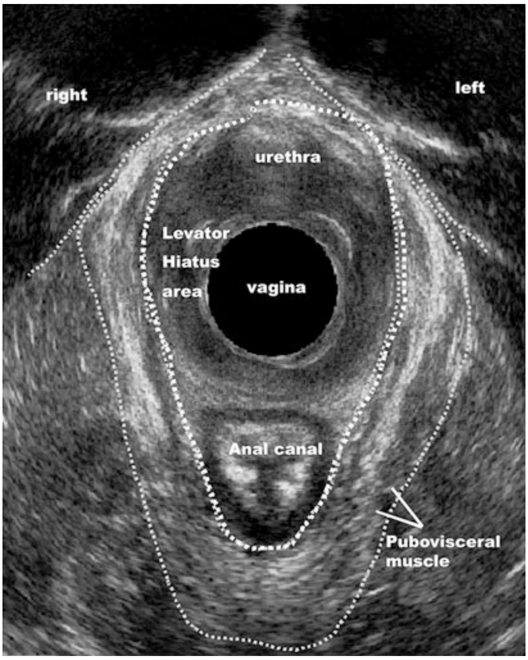

Fig. 18.9 3D Endovaginal ultrasonography – the anatomic configuration of the pubovisceral muscle

demonstrated after vaginal childbirth. The most common form of major levator trauma appears to be an avulsion injury at the insertion of the muscle on the pubic branch, which has been demonstrated by MRI and ultrasonography (DeLancey et al. 2007; Dietz and Simpson 2008; Abdool et al. 2009; Murad-Regadas et al. 2013) in 15–55 % of parous women after vaginal delivery, and which is associated with pelvic organ prolapse and ballooning hiatal dimensions (DeLancey et al. 2007; Dietz and Simpson 2008; Murad-Regadas et al. 2014). The ultrasound examination identifies pubovisceral defects, defined as the detachment (discontinuity) of the PVM from its insertion on the pubic branch. Complete detachment of the PVM involves the entire muscle and partial detachment is either unilateral (involving just one side) or bilateral (both sides). The volume of the defect (sum of microsections at 2 mm) can be measured and the measurements also include the anteroposterior diameter, laterolateral diameter, and the full area. Murad-Regadas et al. demonstrated that severity of FI symptoms is significantly related to extent of the defect as determined by the novel 3-D ultrasound score, including the anal sphincters and pubovisceral muscle defects in women who had undergone to vaginal delivery, and suggested both evaluation of the anal sphincter and PVM to identify the defects and determine a strategy for treatment in women with FI after vaginal delivery (Fig. 18.10). They also evaluated the position of the anorectal junction, as measured from the anorectal junction to the lowest margin of the symphysis pubic (Fig. 18.11), and the position of the bladder neck, as measured from the bladder neck to the lowest margin of the symphysis pubic (Murad-Regadas et al. 2014).

5 Anorectal Abscess

The ultrasound is useful to show the location, extension of the abscess cavity, and relation to the sphincter muscles and to the rectal wall, making possible its classification. Abscesses appear as a non-homogenous hypoechoic area due to the inflammatory process associated with more hypoechogenic area in those cases with liquid content in the cavity. A hyperechogenic area inside the cavity suggests residual air.

The ultrasound image is also able to show early inflammatory processes or the late absorption stage. And these findings cannot be identified by proctological exam alone and are not able to determine if the therapy should be conservative or surgical. The US images are likewise useful to determine the location and extension of large abscesses in relation to the sphincter muscles as well as in the choice of the best treatment approach. It is also well tolerated because the scanning procedure is quick and images may be analyzed posteriorly.

The examination may be difficult or inconclusive in patients with Fournier's syndrome due to the fibrosis and excessive damage muscle.

6 Anal Fistula

The anorectal ultrasonic scanning is able to identify all the fistula complex, facilitating the surgical planning and preventing recurrence and fecal

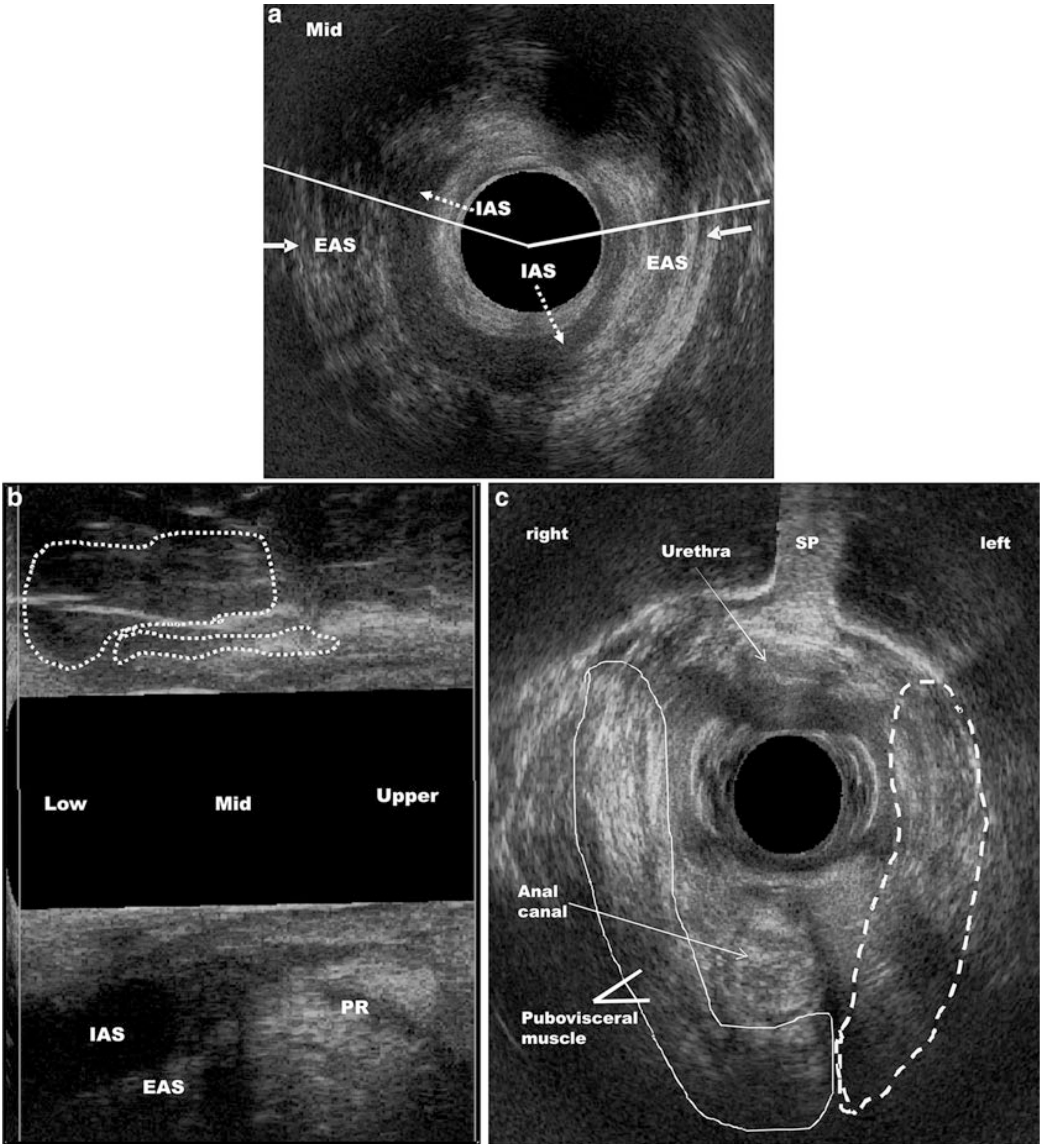

Fig. 18.10 Sphincteric and pubovisceral muscles injuries after a complicated vaginal delivery. (**a**) (Axial plane) – EAS and IAS injuries (9–3 o'clock – *white arrows*). EAS angle measurement (**b**) – (Mid sagittal) Anterior EAS and IAS injured in their whole length (*dotted line*) (**c**) – 3D endovaginal ultrasonography – Pubovisceral muscle defect (*left side*) (*dotted line*). *IAS* internal anal sphincter, *LM* longitudinal muscle, *EAS* external anal sphincter, *PR* puborectalis muscle, *SP* symphysis pubis

incontinence. The 3-D scanning mode allows to accurately view and classify the entire extension of the fistulous tract and its relation to the sphincter muscles, the exact position of the internal opening in relation to the anal margin and any secondary tracts and / or cavities. If the external opening is pervious, hydrogen peroxide (H_2O_2) should be applied through a fine polyethylene catheter to confirm findings and identify any previously secondary tracts (Cheong et al. 1993).

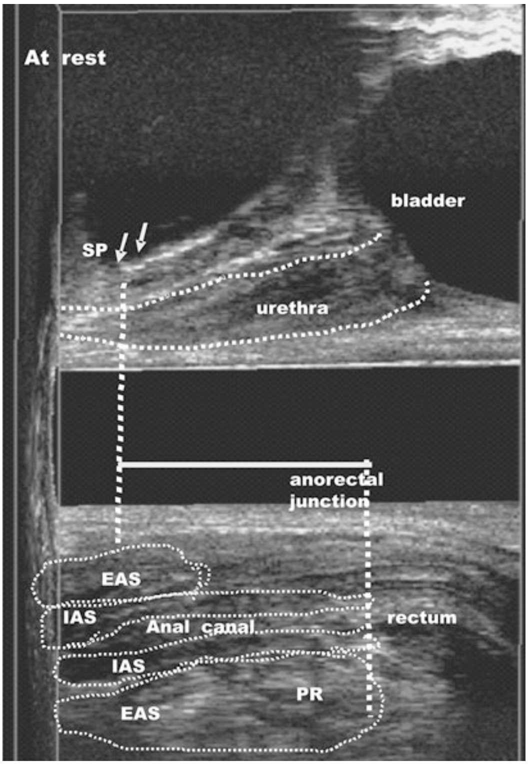

Fig. 18.11 3-D endovaginal ultrasound. Measurements of the anorectal junction position. *IAS* internal anal sphincter, *LM* longitudinal muscle, *EAS* external anal sphincter, *PR* puborectalis muscle, *SP* symphysis pubis

The scan is performed in two steps, administering 0.1–3.0 ml of 10 % H_2O_2 at normal pressure (first step) and heightened pressure (second step) (Murad-Regadas et al. 2010). Fistulous tracts typically appear as hypoechoic areas and the echogenicity increases and then appears as hyperechoic, as a result of bubble formation due to the contact between H_2O_2 and the tissue. The internal fistulous opening appears on the image as a rupture in the IAS (in the absence of previous sphincterotomy) and subepithelium tissue. When the use of hydrogen peroxide is applied, a hyperechoic area is clearly observed in the subepithelium space, crossing the IAS towards the endoprobe. Based on the classification proposed by Parks, the anorectal fistulas are showed with the following ultrasound features (Parks et al. 1976):

1. Intersphincteric – The fistulous tract is located in the intersphincteric space, with the distal part between the EAE and the subepithelial surface (Fig. 18.12);
2. Transsphincteric – The tract crosses the external and internal anal sphincters. According to the point at which the tract crosses the EAS, fistulas may be classified into high, medium, or low (Fig. 18.13);
3. Extrasphincteric – The tract is located in the ischiorectal fossa (laterally to the sphincter muscles) while the internal opening is in the rectum, corresponding to an area of lost uniformity in the rectal layers (Fig. 18.14);
4. Suprasphincteric – The internal fistulous opening may be seen associated with the intersphincteric tract extending toward the rectum and crossing over the puborectalis muscle distally, laterally to the sphincter muscles, through the ischiorectal fossa.

The percentage of sphincter muscle to be sectioned during surgery must be previously determined. To calculate this percentage, the total length of the compromised sphincter is measured as well as the distance from the distal part of each muscle to the point where it is crossed by the fistulous tract (Figs. 18.15 and 18.16). These measures are used in surgical planning and help prevent fecal incontinence (Murad-Regadas et al. 2010).

7 Anorectal-Vaginal Fistula

The ultrasound scanning may show the fistulous tract and its relation to the anal canal or rectum and helps to evaluate the anatomic integrity of the sphincter muscles, improving consequently the surgical planning and preventing recurrence and fecal incontinence.

The identification of the opening in the vagina and the injection of hydrogen peroxide help to visualize the tract, especially at the point where it crosses the perineal body and in cases of associated fibrosis. The transducer may subsequently

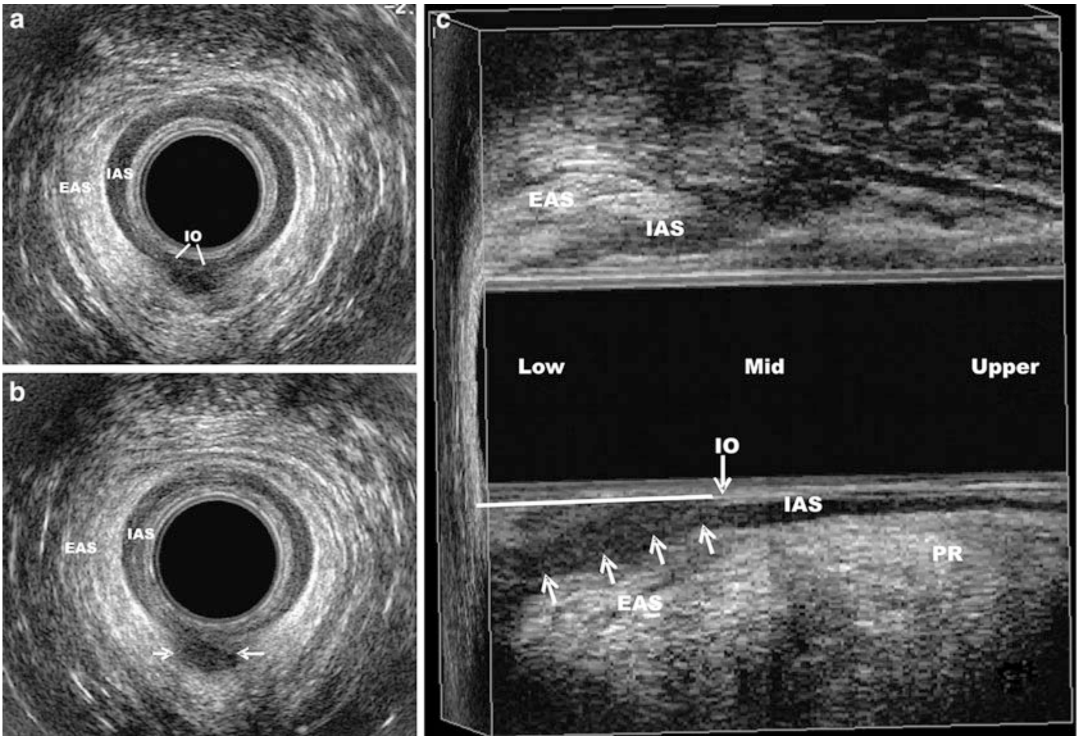

Fig. 18.12 Patient without H_2O_2 injection. (**a**, **b**) – Posterior intersphincteric tract located between 6 and 7 o'clock (*arrow*). IO located at 6 o'clock, 2 cm from the anal verge (**c**) Intersphincteric tract length (sagittal plane) (*arrows*). *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle, *IO* internal opening

be introduced into the vagina to confirm or expand the findings.

8 Benign and Malignant Rectal Neoplasias

The anorectal ultrasonography is particularly useful for staging rectal cancer as it provides accurate information on rectal wall infiltration, anal canal invasion and perirectal lymph node enlargement and select patients for surgery or to neoadjuvant radiochemotherapy (RCT) (Hildrebant and Fiefel 1985; Milsom and Graffner 1990; Katsura et al. 1992; Dattala et al. 2000; Garcia-Aguilar et al. 2002). Using the 3-D modality, it is easily measured tumor length and volume, the distance between the distal margin of the tumor to proximal margin of the anal sphincter muscles and determine the distal margin (Murad-Regadas et al. 2009; Fig. 18.17), and the closest predicted radial tumor-mesorectal margin (Fig. 18.18; Phang et al. 2012). The 3-D anorectal ultrasound (3-DAUS) is also useful to evaluate the response after RCT. Murad-Regadas et al. identified residual tumors in rectal wall and lymph nodes and complete response with a high level of accuracy (Murad-Regadas et al. 2009). In addition, 3-DAUS can help in the choice of surgical approach by providing important information on the distance between the tumor and the anal muscle and select patients for sphincter saving resection (Murad-Regadas et al. 2011b). In the follow up, this

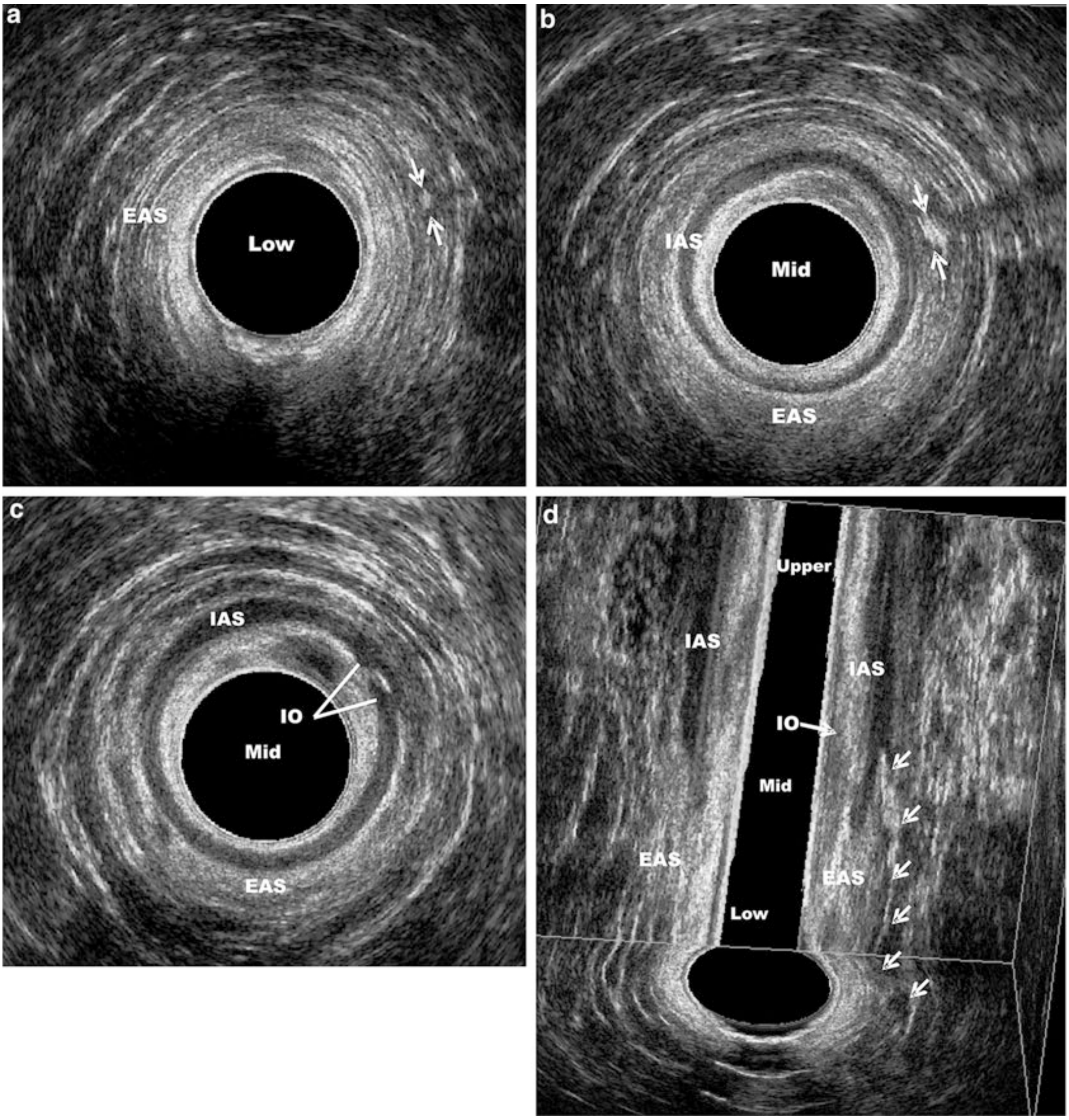

Fig. 18.13 Patient with H_2O_2 injection. Transsphincteric fistula (**a**) Low anal canal – tract outside the EAS (*arrow*) (**b**) Middle anal canal. Tract crossing the EAS (*arrow*) (**c**) Internal opening is located between 2 and 3 o'clock (**d**) Whole length of the transsphincteric tract (*arrows*). *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle

modality detect early local recurrence in the rectal wall or perirectal lymph nodes (Beynon et al. 1986)

Ultrasonographic tumor staging – In 1985, Hildebrand and Fielfel proposed using ultrasound scanning for the staging of rectal tumors, based on the TNM classification (Hildrebant and Fiefel 1985):

uT0 – Noninvasive lesion affecting the mucous membrane and the muscularis-mucosa
uT1 – Submucosal invasion

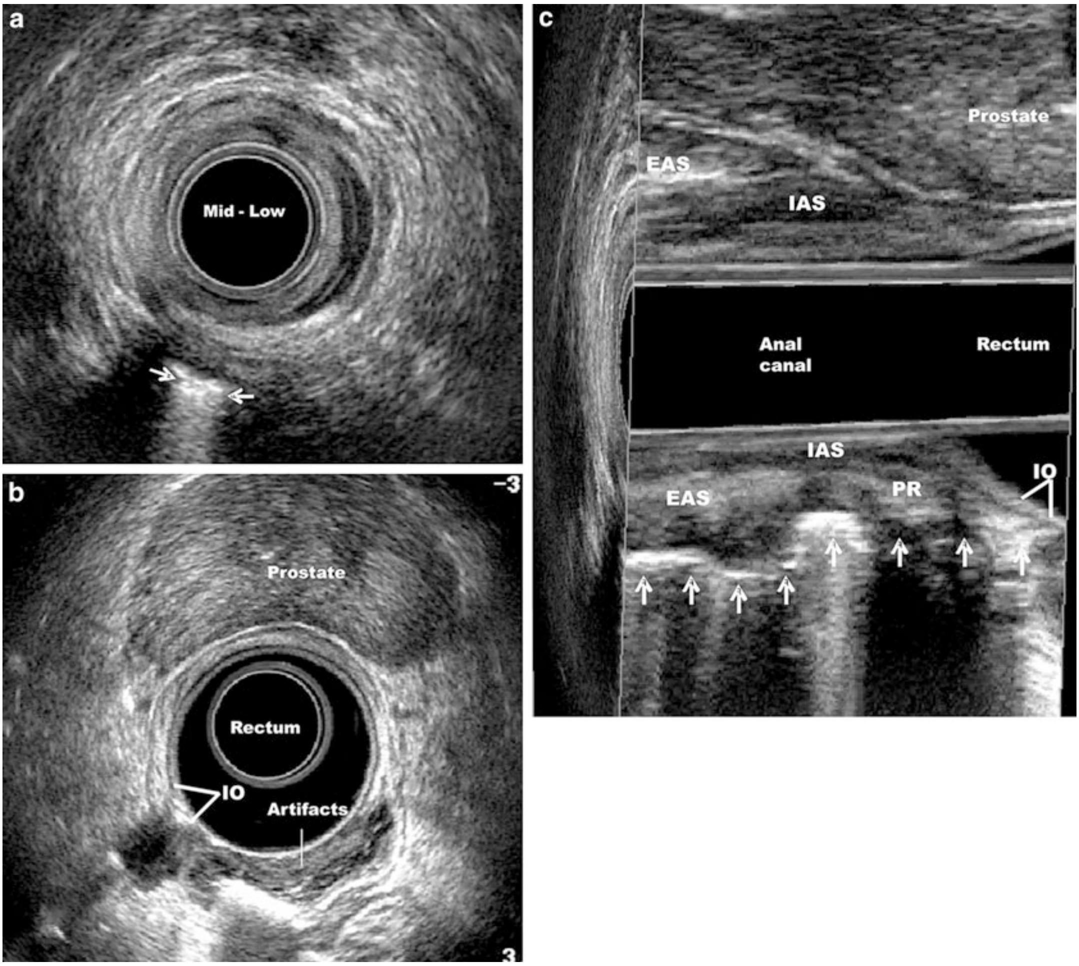

Fig. 18.14 Patient with H_2O_2 injection. Extrasphincteric fistula (**a**) Low anal canal – tract outside the muscle (*arrow*). (**b**) Lower rectum. IO is located at 7–8 o'clock (**c**) Tract is located outside the sphincter muscles, extending from the perianal skin to the rectum lumen (*arrows*). IO is located in the lower rectum. *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle, *IO* internal opening

uT2 – Invasion of the circular and longitudinal muscle layers
uT3 – Invasion of perirectal fat
uT4 – Invasion of adjacent organs
N0 – Uncompromised lymph nodes
N1 – Compromised lymph nodes

– **Muscularis-mucosa and muscle layers** – Represented by hypoechoic (dark) images. Thickened layers suggest tumor invasion or involvement.
– **Submucosal layer and perirectal fat** – Represented by hyperechoic (white) images. Tumor invasion appears as a disruption or irregularity.

(a) Benign neoplasia – Characterized by a thickened muscularis-mucosa layer and an intact submucosal layer.
(b) Severe displasia, adenocarcinoma *in situ* – Hypoechoic areas surrounded by homogenous image, characteristic of adenoma.

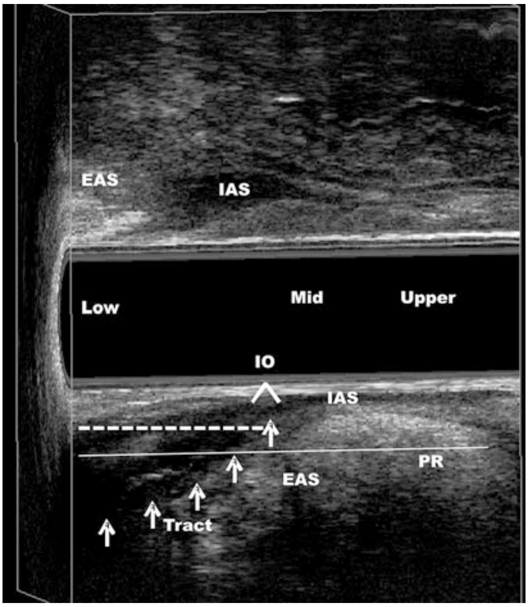

Fig. 18.15 Patient with H_2O_2 injection. Transsphincteric fistula located at 7 o'clock. The percentage of sphincter muscle to be transected during the surgical procedure is measured (*dotted line*). Muscle length (*continuous line*). *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle

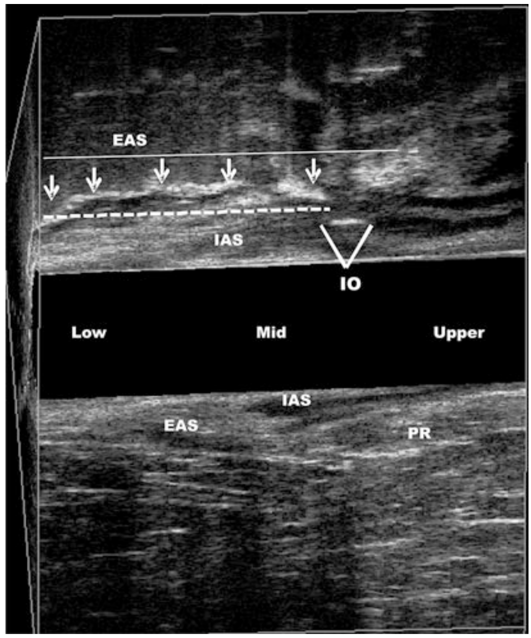

Fig. 18.16 Patient with H_2O_2 injection. Anterior transsphincteric fistula. The percentage of sphincter muscle to be transected during the surgical procedure is measured (*dotted line*). Muscle length (*continuous line*). *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle

(c) uT1-type lesion – A disruption (irregularity) is observed in the second hyperechoic (submucosal) layer.
(d) uT2-type lesion – Complete disruption of the submucosal layer associated with thickening of the musculature and intact perirectal fat.
(e) uT3-type lesion – Irregularities (spicules) in the last hyperechoic layer (corresponding to the perirectal fat).
(f) uT4-type lesion – Characterized by invasion of adjacent structures.

Perirectal lymph nodes – Observed in the perirectal fat proximally or distally to the lesion and measuring over 1.0 mm. They are easily distinguished from blood vessels, because the latter assume a longitudinal or branch-like form when the transducer is moved. The size, echogenicity, and shape help distinguish between inflammatory and metastatic forms. When observed in the perirectal fat in the form of rounded and hypoechoic (or tumor-like) (Fig. 18.19) areas with irregular borders, lymph node metastasis should be suspected. In contrast, oval structures with regular borders and a hyperechoic area in the center (corresponding to the hilum) suggest inflammatory lesions.

8.1 Limitations of Ultrasonographic Staging of Neoplasms

The patient and the transducer must be adequately prepared in order to produce useful images with a minimum of artifacts and the best possible definition of rectal layers and/or perirectal tissues. Other factors may interfere with the accuracy of ultrasonographic staging of neoplasms:

(a) Large lesions can produce attenuated areas such as posterior acoustic shadows (shadows behind tumors) making staging difficult.
(b) In case of very small lesions and the balloon is excessively distended around the transducer, lesions may be compressed and overstaged.

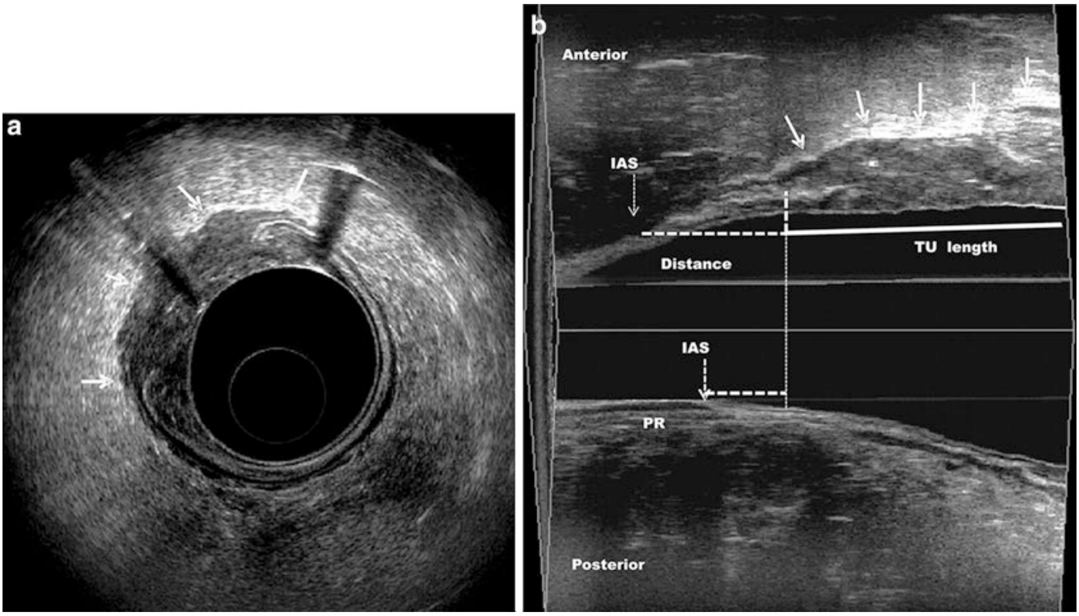

Fig. 18.17 uT3-Rectal cancer (*arrows*) located on the anterior and right lateral quadrant. (**a**) Axial plane (**b**) Measurement of tumor length (*arrows*) and the distance between the distal border of the tumor and the proximal border of the IAS and PR. *IAS* internal anal sphincter/*PR* puborectal muscle

(c) If the air which is often retained on the surface of ulcerous lesions becomes interposed between the lesion and the transducer and the balloon is distended, a reverberation due to the reflection of the echo received by the transducer, leading to a series of densely juxtaposed reflexes which coalesce behind the lesion making it impossible to view it completely.
(d) Severely inflamed lesions make it hard to distinguish between inflammatory parietal thickening and tumor invasion. In addition, the inflammatory reaction can produce attenuated areas (shadows) behind the lesion making staging inaccurate. Peritumoral inflammation from the own lesion often leads to ultrasonographic overstaging, making the invasion seem larger than it is. Understaging is generally observed in cases of minimally invasive lesions and cause retention of tumor tissue, early recurrence and shortened survival. Tumor biopsies can produce inflammatory reactions such as edema and fibrosis and even intratumoral hemorrhage resulting in hypoechoic patches compromising interpretation. The examination should therefore be performed at least 15 days after the biopsy.

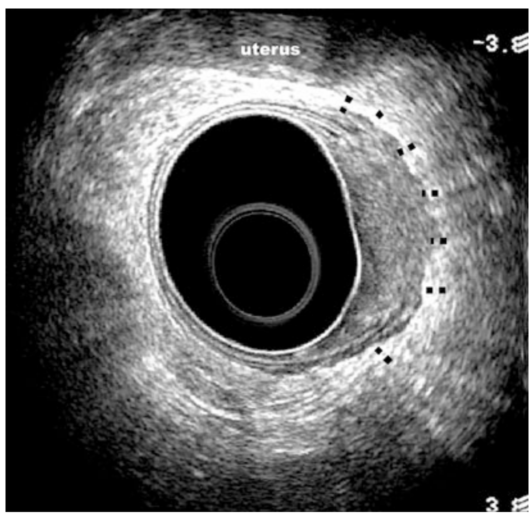

Fig. 18.18 uT3-Rectal cancer (*arrows*) located on the left lateral quadrant. Distance from the tumor to the mesorectal fascia – radial tumor-mesorectal margin. *IAS* internal anal sphincter/*PR* puborectal muscle

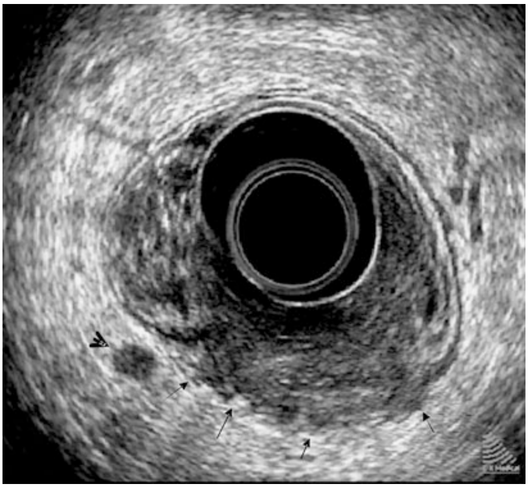

Fig. 18.19 uT3-Rectal cancer (*arrows*) in a male patient located on the posterior, right and left lateral quadrants. Malignant lymph node located on the right quadrant

(e) Stenosing tumors represent a challenge to endorectal ultrasound scanning. Ultrasound is not indicated when it is not possible to bypass them.
(f) Staging lesions located at the rectosigmoid junction can be difficult to keep the transducer in the center of the circumference.

8.2 Limitations of Ultrasonographic Staging of Lymph Nodes

(a) The evaluation of perirectal tissue may be inadequate due to artifacts (shadows and reverberation), insufficient preparation of the rectum, or the endoprobe.
(b) Lateral pelvic lymph nodes cannot be evaluated because they exceed the focal distance of the transducer.

8.3 Staging After Radio/ Chemotherapy

Response to RCT may be evaluated with endorectal ultrasonography, PET scan and magnetic resonance imaging (MRI) (Napoleon et al. 1991; Barbaro et al. 1999; Vanagunas et al. 2004; Capirci et al. 2004; Chen et al. 2005; MERCURY Study Group 2006). One of difficulties is distinguish individual layers and therefore to restage lesions. Studies using ultrasound scanning to evaluate tumor regression in response to radiotherapy, disappearance of lymph nodes or overall response varied 47–75 % (Napoleon et al. 1991; Vanagunas et al. 2004) in relation to other imaging techniques and anatomical/pathological findings. On the other hand, Barbaro et al. reported 95 % post-RCT uTN restaging accuracy for rectal tumors and 61 % for compromised lymph nodes using transrectal ultrasonography (Barbaro et al. 1999).

In a study of rectal tumor patients submitted to RCT and evaluated with 3-DAUS, Murad-Regadas et al. identified residual tumors and complete response with a high level of accuracy. In addition, 3-DAUS is able to evaluate tumor length and total volume and to detect anal canal invasion and can help in the choice of surgical approach by providing important information on the distance between the tumor and the anal muscles (Murad-Regadas et al. 2009). By evaluating patients with malignant rectal neoplasms using high-resolution automatic 3-D ultrasound scanning before and after radiotherapy and comparing the results with anatomical/pathological findings, criteria have been established parameters for postradiotherapy ultrasonographic analysis of such lesions (Murad-Regadas et al. 2009):

(a) *Complete regression of lesions* – Rectal wall layers and/or sphincter muscles are clearly distinguishable where the tumor was previously located (Fig. 18.20).
(b) *Residual lesions* – Certain patterns are observed depending on the extent of tumor regression and the association with the inflammatory process:
    1. Heterogeneous image with hyperechoic areas due to residual tumor associated with hypoechoic areas due to the inflammatory process. Anatomical disorder is observed associated with parietal thickening at the tumor borders and rectal wall layers cannot be distinguished.

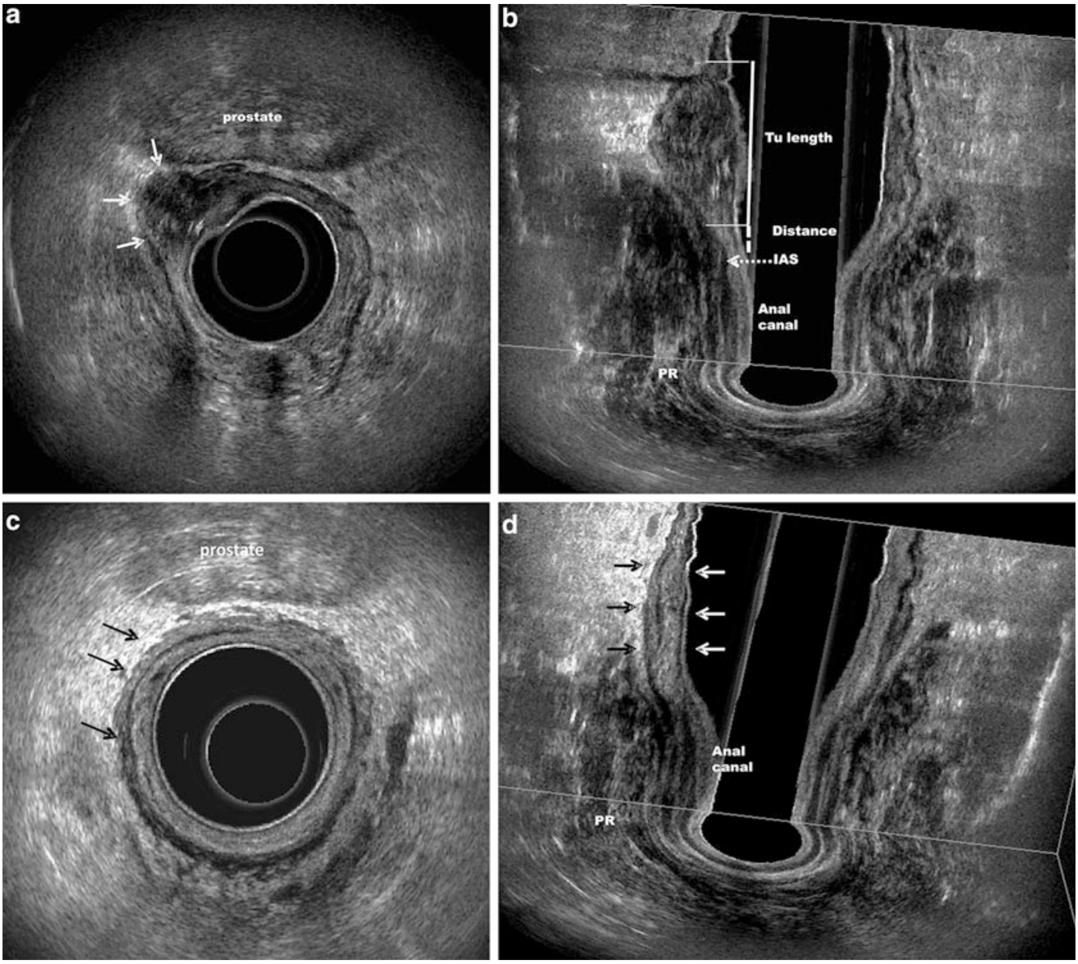

Fig. 18.20 (**a**) uT3-Rectal cancer located on the anterior and right lateral quadrants (*arrows*) before RCT. (**b**) Tu length and the distance between the distal border of the tumor and the proximal border of the IAS posterior before RCT (**c**) Complete response – rectal wall layers is clearly distinguishable where the tumor was previously located (*arrows*) (axial plane) after RCT (8–10 weeks) (**d**) Complete response (*arrows*) (coronal plane) after RCT. *IAS* internal anal sphincter/*PR* puborectal muscle/*RCT* radiochemotherapy

2. Similar to image before radiotherapy, but more hypoechoic due to inflammation. Rectal wall layers are distinguishable at former tumor location and restaging is possible in the absence of inflammation-induced anatomical disorder. The circumference and length of the lesion may be decreased and the distance between the distal border of the tumor and the proximal border of the sphincter muscles may have increased (Fig. 18.21)

(c) *Inconclusive image*: Characterized by parietal thickening with indistinguishable rectal wall layers at former tumor location. The existence of residual tumors cannot be ruled out.

8.4 Identification of Early Recurrence

Characterized by tumor-like hypoechoic image with the widest portion outside the rectal wall. The initial examination becomes an important reference during follow-up, since fibrosis and especially after local sepsis and postsurgical

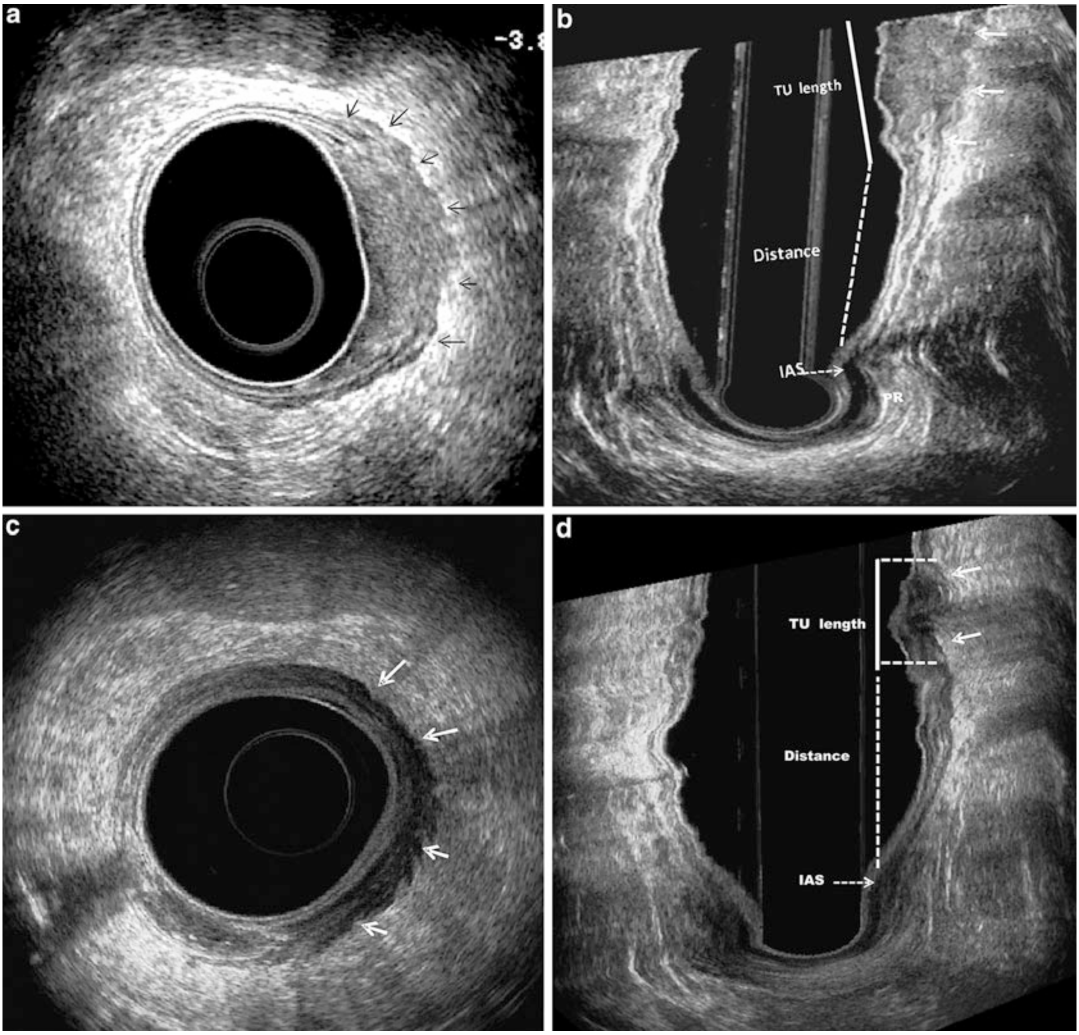

Fig. 18.21 (**a**) uT3-Rectal cancer located on the left lateral quadrant (*arrows*) before RCT. (**b**) Tu length and the distance between the distal border of the tumor and the proximal border of the posterior IAS before RCT (**c**) Partial response after RCT (8–10 weeks) (**d**) The distance between the tumor and the IAS increases. *IAS* internal anal sphincter/*PR* puborectal muscle/*RCT* radiochemotherapy

anatomical distortions can influence interpretation. Tumor recurrence may be detected even when the lesion is enveloped by fibrous tissue.

9 Staging and Follow-Up of Anal Canal Neoplasia

The ultrasound scanning allows to quantify the extent of tumor invasion into the sphincter muscles, adjacent tissues and rectum and to identify compromised lymph nodes, making the examination the mainstay for evaluating response to RCT (Giovanini et al. 2001). The advantages of using 3-D ultrasound are the possibility of evaluation in multiple planes, accurate measurement of circumferential and longitudinal tumor extension, and real-time review in ambiguous cases (Christensen et al. 2004). However, the diagnosis requires detailed clinical examination, evaluation of the risk factors, proctological examination, and the histopathology finding.

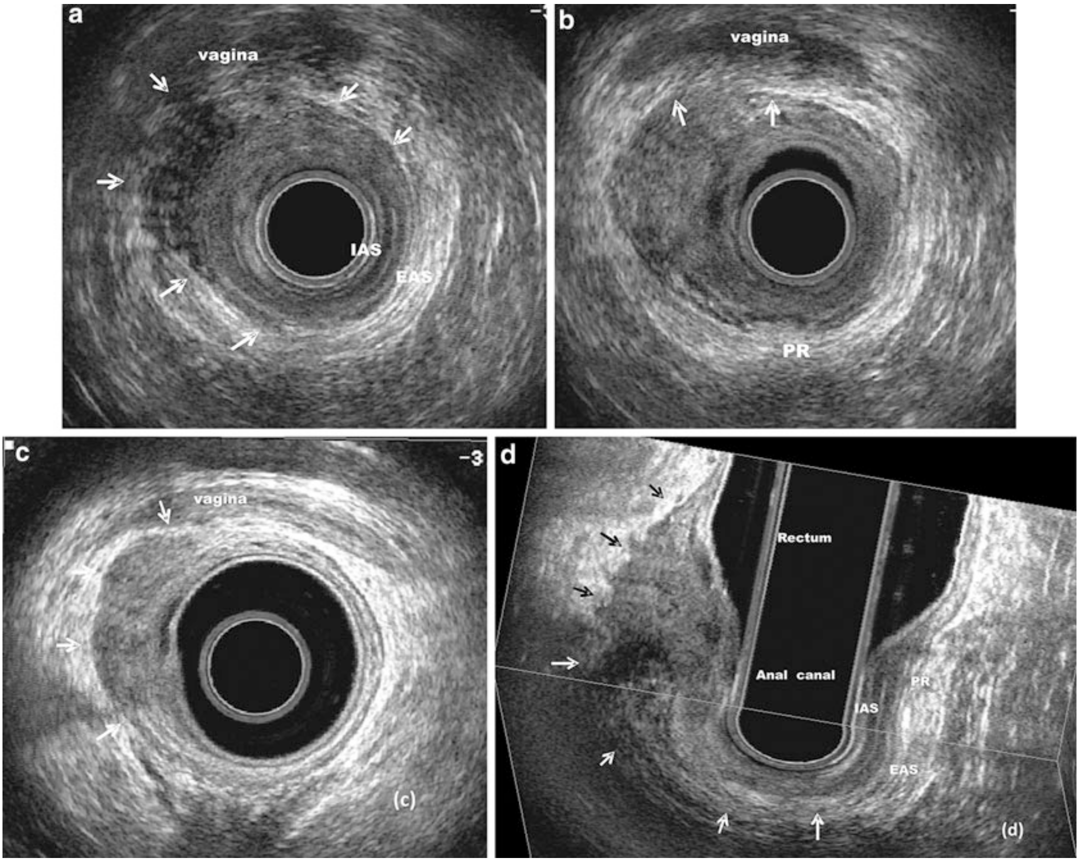

Fig. 18.22 uT4 located at the left and right antero-lateral quadrants, invading the IAS, EAS, perianal fat, and extending to lower rectum. (**a**) Mid-anal canal (Axial plane) – Tumor is invading the IAS, EAS, and perianal fat (*arrows*). (**b**) Upper anal canal (Axial plane) – Tumor is invading the vagina wall (*arrows*). (**c**) Extending to the lower rectum, invading the perirectal fat (outer hyperechoic layer) (*arrows*). (**d**) (Coronal with axial planes) – Lesion is involving middle/upper anal canal and lower rectum (*arrows*). *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle

The most appropriate scanning modality is 3-D anorectal ultrasound because of the possibility of evaluation in multiple planes, accurate measurement of circumferential and longitudinal tumor extension, and real-time review in ambiguous cases. In a study comparing the 2- and 3-D scanning modalities, Christensen et al. demonstrated the superiority of the latter in the staging of lesions due to the possibility of evaluation in multiple planes and real time (Christensen et al. 2004).

Ultrasonographic staging of neoplasms was first proposed by the *Union Internationale Contre le Cancer* (UICC) in 1987, and it is based on the criteria of tumor size and degree of invasion (International Union Against Cancer 1997).

uT1 – Tumor restricted to the mucous membrane
uT2 – Tumor compromising the internal anal sphincter
uT3 – Tumor compromising the external anal sphincter
uT4 – Tumor invading adjacent structures
N0 – Lymph nodes unaffected
N1 – Lymph nodes affected.

Based on the satisfactory responses obtained in 94.5 % of cases, it is now generally held that uT1 and uT2-type tumors under 4.0 cm should be treated with radiotherapy alone. For uT2-type lesions larger than 4.0 cm and for uT3 and uT4-type lesions, treatment should include both radio and chemotherapy (International Union Against Cancer 1997).

Tarantino and Bernstein have been proposed tumor stage based on the degree of invasion alone (Tarantino and Bernstein 2002).

uT1 – Tumor restricted to the submucosal layer. Echographically, this type of lesion is characterized by a hypoechoic area in the subepithelial (mucosal-submucosal) tissue due to thickening, with the IAS preserved
uT2a – Tumor compromising the IAS. Characterized by hypoechoic area representing disruption or thickening of the musculature
uT2b – Tumor compromising the EAS. Hypoechoic area covering the entire width of the IAS and affecting the EAS and the PR
uT3 – Perianal tissue affected
uT4 – Adjacent structures affected (Fig. 18.22)
N0 – Lymph nodes unaffected
N1 – Lymph nodes affected

If the lower rectum has been affected, the exam should include scanning in the rectal mode and the evaluation of the rectal wall layers (Fig. 18.22).

The lymph nodes are located in the perianal or perirectal fat proximally or distally to the lesion.

Post-RCT ultrasound scanning may be performed after 6–8 weeks and then periodically, depending on the case, until response to therapy has been established. During this period, the patient may also need to be submitted to proctological exams and biopsy.

Less invasive tumors may appear to have regressed completely upon first follow-up exam if the anatomical structures are clearly distinguishable at the former tumor location. However, reduced size lesions, ill-defined and slightly hypogenic areas may be observed as a result of the radio/chemotherapy effects (edema, inflammation, and fibrosis) or even presence of residual tumor. However, the continuous size reduction of such image seen during the subsequent exams, defining the anatomic structures and visualizing all the rectal layers represent complete tumor regression. The presence of residual tumor (incomplete response) is evidenced by the persistence of well-defined hypoechoic image or by its enlargement on the subsequent exams and/or in the absence of cleavage planes with affected adjacent structures. When a residual tumor is suspected due to the persistence of a hypoechoic image from the first follow-up examination, an excisional biopsy should be performed for definitive diagnosis. In this case, ultrasound scanning is helpful in the selection of an appropriate biopsy site, preventing muscle injury. Between the fourth and fifth month, the patients have a complete regression of radiotherapy effects (edema and fibrosis). At this point, any change in the image may suggest recurrence. Periodical ultrasound scanning during follow-up can provide early diagnosis of subclinical neoplasms not observed on earlier exams.

When response to radio/chemotherapy is complete, follow-up exams are performed every 6 months, or even more frequently, during the first 2 years, then annually until the fifth year.

Ultrasound scanning makes possible to follow the evolution of neoplasms of the anal canal, choose a suitable treatment approach, establish response to treatment, confirm complete response, or select safe biopsy sites in case of suspicion of early recurrence.

10 Miscellaneous (Endometriosis, Pelvic Cyst, Rectal Solitary Ulcer, Rare Neoplasias)

10.1 Endometriosis

10.1.1 Rectal Endometriosis

Anorectal ultrasound scanning provides the most detailed view of endometriosis infiltration in the rectal wall (Bahr et al. 2006). The 3-D modality allows an accurate measurement of circumferential and longitudinal tumor extension and the distance to the sphincter muscles, thus providing crucial information for the choice of therapeutic approach (Regadas and Murad-Regadas 2008). Lesions appear as heterogeneous hypoechoic images, mostly located in the perirectal fat or serosa

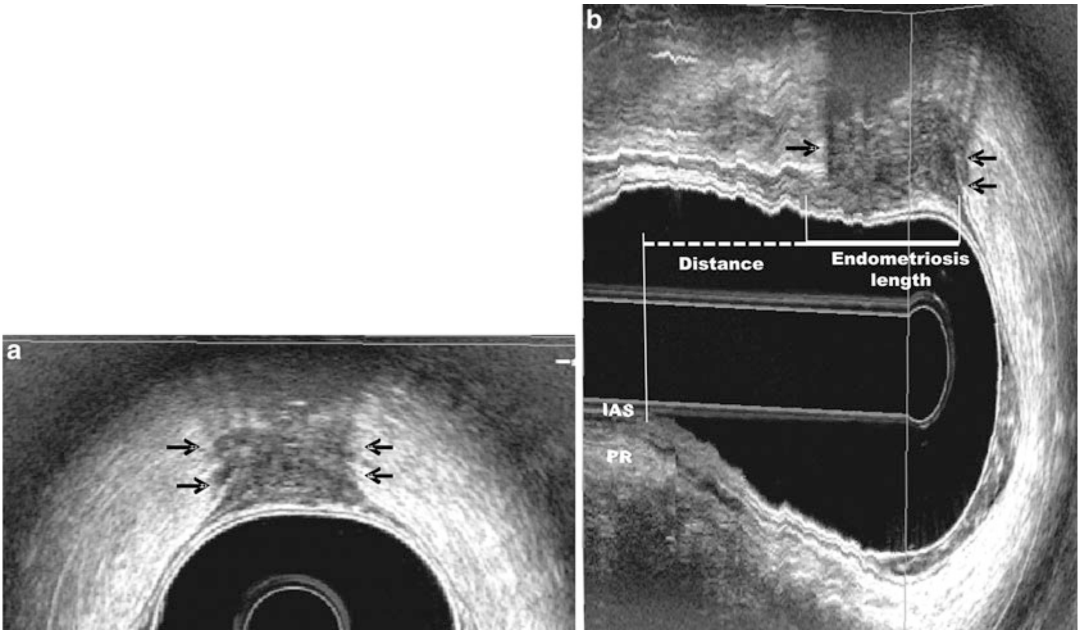

Fig. 18.23 Endometriosis lesion in the anterior quadrant, infiltrating the rectal wall as far as the muscular propria layer (**a**) Axial plane – Heterogeneous hypoechoic image compromising 30 % of rectal circumference (*arrows*). (**b**) Sagittal plane – The length of the endometriosis lesion and the distance between the distal infiltration edge and the proximal edge of the posterior sphincter muscles (*arrows*). *IAS* internal anal sphincter, *EAS* external anal sphincter, *PR* puborectalis muscle

and infiltrating the layers of the rectal wall into the muscular propria or submucosa layers (Fig. 18.23).

10.1.2 Perianal Endometriosis

While proctological exam allows establishing a diagnosis of perianal endometriosis, 3-D ultrasound scanning makes possible to determine the exact circumferential and longitudinal extension of the infiltration into the sphincter muscles and the rectovaginal septum.

Presacral Neoplasia

Perirectal neoplasia is most often located in the retrorectal space and may be of varied etiology. Half the cases are congenital and two-thirds are cystic in nature. They are classified as epidermoid cysts, dermoid cysts, enteric cysts (tailgut cysts and cystic rectal duplication), and neurenteric cysts according to their origin and histopathologic features (Gordon 1999).

Anorectal ultrasound scanning is useful in the evaluation of size, type of lesion and relation with the rectal wall and the sphincter muscles. Perirectal neoplasia appears with different characteristics: as a unilocular or multilocular retrorectal lesion, sometimes a hypoechoic area (cystic) or as an area of mixed echogenicity/heterogeneous image, due to mucoid material or inflammatory debris or solid component, usually with regular outline and not adhering to the rectal wall. It is important to define the evidence of rectal wall infiltration in invasive lesion or a communication between the cyst and the anorectal lumen.

Rare Tumors

It is included in these groups: Schwanoma or neurilenoma and Gastrointestinal stromal tumors.

Schwanoma is a rare neoplasm, originally referred to as malignant schwanoma, currently known as malignant peripheral nerve sheath tumor. Although a definitive diagnosis requires anatomical and pathological examination.

Gastrointestinal stromal tumors are the most common mesenchymal tumors of the gastrointestinal tract but they comprise fewer than 1 % of all GI tumors (Judson and Demetri 2007).

Anorectal ultrasound scanning provides the most detailed view of such lesions and the relationship with the anatomical structures, including perirectal, perianal tissues, anal canal muscle, perirectal wall, and adjacent organs.

11 Pelvic Floor Dysfunctions

Some techniques have been published using dynamic ultrasound with different types of transducers to evaluate pelvic floor dysfunction related to obstructed defecation syndrome, and the results were similar when compared with defecography (Barthet et al. 2000; Beer-Gabel et al. 2004; Steensma et al. 2010). Murad-Regadas et al. developed the echodefecography, a 3-D dynamic anorectal ultrasonography technique using a 360° transducer, automatic scanning, and high frequencies for high-resolution images to evaluate the evacuation disorders affecting the posterior compartment (Rectocele, Intussusception, Anismus) and the middle compartment (grade II or III Sigmoidocele/Enterocele) (Murad-Regadas et al. 2008, 2011). The standardization of the technique, parameters, and values of the echodefecography makes the method reproducible, as demonstrated in a previous prospective multicenter study, comparing with conventional Defecography (Regadas et al. 2011).

11.1 Echodefecography (EDF) Technique

It is performed with a 3-D ultrasound device (Pro-Focus, endoprobe model 2052, B-K Medical®, Herlev, Denmark) with proximal-to-distal 6.0-cm automatic scans. By moving two crystals on the extremity of the transducer, axial and longitudinal images are merged into a single cube image, recorded and analyzed in multiple planes.

Following a rectal enema, the patient is examined in the left lateral position. Images are acquired by four automatic scans and analyzed in the axial, sagittal and, if necessary, in the oblique plane. The result of the exam depends on the degree of cooperation obtained from the patient.

- Scans 1, 2, and 4 used a slice width of 0.25 mm and lasted 50 s each.
- Scan 3 lasted 30 s with a slice width of 0.35 mm.

Scan 1 (at rest position without gel): The transducer is positioned at 5.0–6.0 cm from the anal margin. It is performed to visualize the anatomic integrity of the anal sphincter muscles and to evaluate the position of the PR muscles and EAS at rest. The angle formed between a line traced along the internal border of the EAS-PR muscles (1.5 cm), and a line traced perpendicular to the axis of the anal canal is measured.

All the following scans are dynamic evaluation.

Scan 2 (at rest – straining – at rest without gel): The transducer is positioned at 6.0 cm from the anal verge. The patient is requested to keep at rest during the first 15 s, strain maximally for 20 s, then relax again, with the transducer following the movement. The purpose of the scan is to evaluate the movement of the PR-EAS muscles during straining, in order to identify normal relaxation, non-relaxation, or paradoxical contraction (anismus). The result of the EAS-PR muscles' position (represented by the angle size) is compared between scans 1 and 2. Normal relaxation is recorded if the angle increases by a minimum of 1°, whereas paradoxical contraction (anismus) is recorded if the angle decreases by a minimum of 1°. Non-relaxation is recorded if the angle changes less than 1° (Figs. 18.24 and 18.25).

Scan 3: The transducer is positioned proximally to the PR (anorectal junction). The scan starts with the patient at rest (3.0 s), followed by maximum straining with the transducer in fixed position (the transducer does not follow the descending muscles of the pelvic floor). When the PR becomes visible distally, the scan is stopped.

Perineal descent is quantified by measuring the distance between the position of the proximal border of the PR at rest and the point to which it

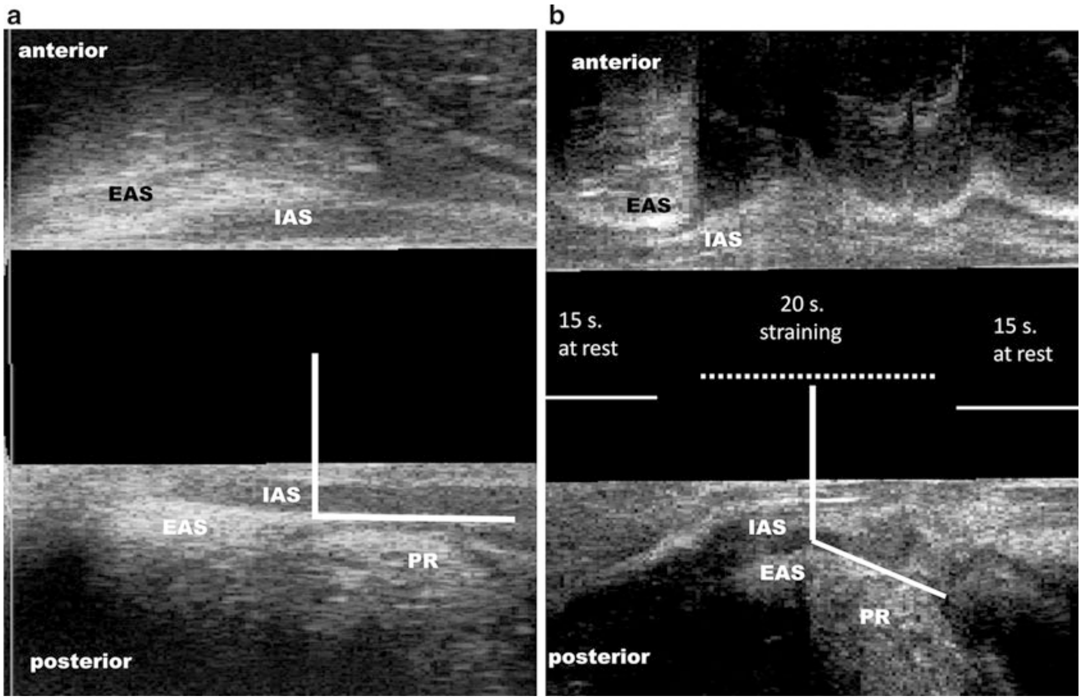

Fig. 18.24 Echodefecography: (**a**) Angle measured at rest position (*lines*). (**b**) Increased angle (normal relaxation) during straining (*lines*). *EAS* external anal sphincter, *IAS* internal anal sphincter, *PR* puborectal

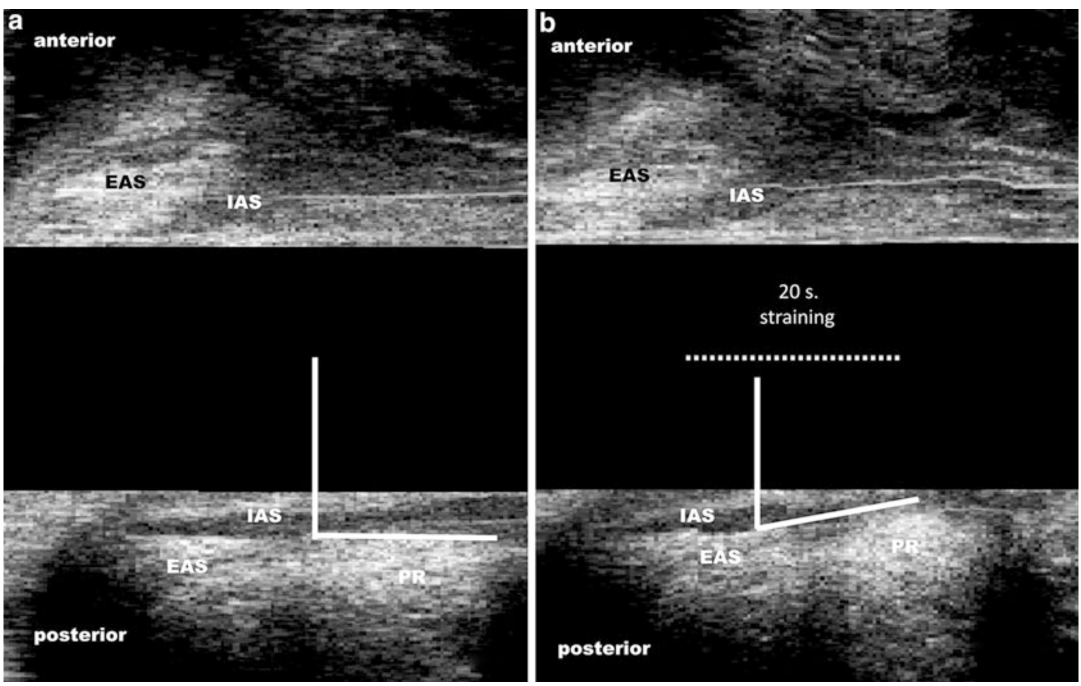

Fig. 18.25 Echodefecography: (**a**) Angle measured at rest position (*lines*). (**b**) Decreased angle (Anismus) during straining (*lines*). *EAS* external anal sphincter, *IAS* internal anal sphincter, PR puborectal

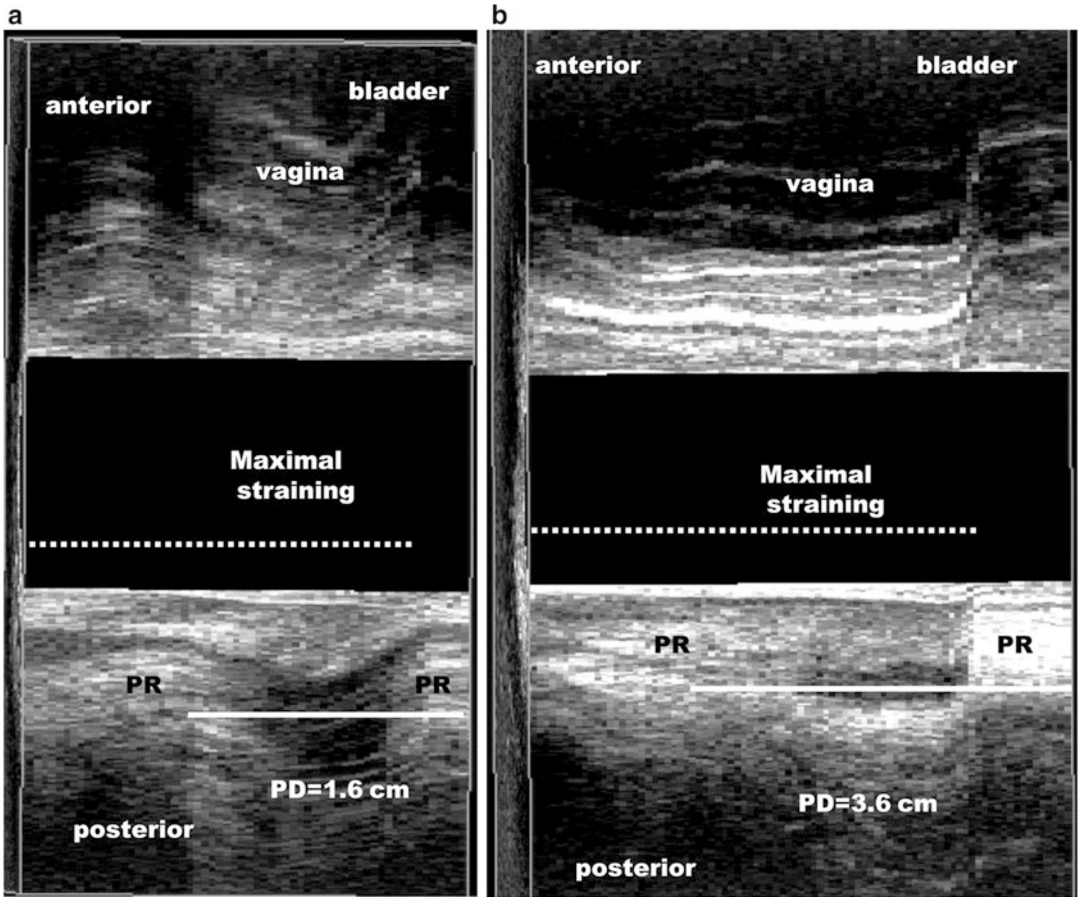

Fig. 18.26 Echodefecography: (**a**) Normal perineal descent – PD ≤2.5 cm. (**b**) Perineal descent – PD >2.5 cm. *PR* puborectal, *PD* puborectal descent

has been displaced by maximum straining (PR descent). Straining time is directly proportional to the distance of perineal descent.

Even with patients in the lateral position, the displacement of the PR is easily visualized and quantified. On echodefecography, normal perineal descent during straining is defined as a difference in PR position of ≤2.5 cm and perineal descent >2.5 cm (Fig. 18.26). The normal range values were established by comparing EDF findings with DF.

Scan 4: Following injection of 120–180 mL ultrasound gel into the rectal ampulla, the transducer is positioned at 7.0 cm from the anal verge. The scanning sequence is the same as in scan 2 (at rest during 15 s, strain maximally during 20 s, then relax again, with the transducer following the movement). The purpose of the scan is to visualize and quantify all anatomical structures and functional disorders (rectocele, intussusception, grade II or III sigmoidocele/enterocele).

In normal patients, the posterior vaginal wall displaces the lower rectum and upper anal canal inferiorly and posteriorly but maintains a straight horizontal position during defecatory effort (Fig. 18.27). If rectocele is identified, it is classified as grade I (<6.0 mm), grade II (6.0–13.0 mm) or grade III (>13.0 mm) (Fig. 18.28). Measurements are calculated by first drawing two parallel horizontal lines along the posterior vaginal wall, with one line placed in the initial straining position and the other line drawn at the point of maximal straining.

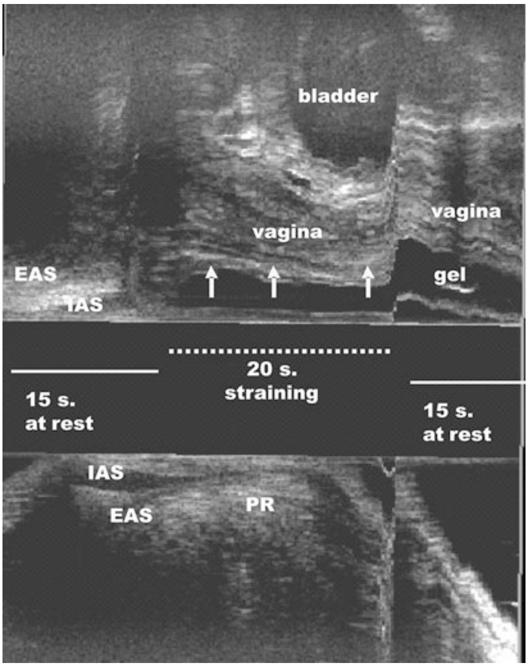

Fig. 18.27 Echodefecography – using gel into the rectum. Patient without rectocele – the vagina maintains a straight horizontal position during defecatory effort. *EAS* external anal sphincter, *IAS* internal anal sphincter, *PR* puborectal

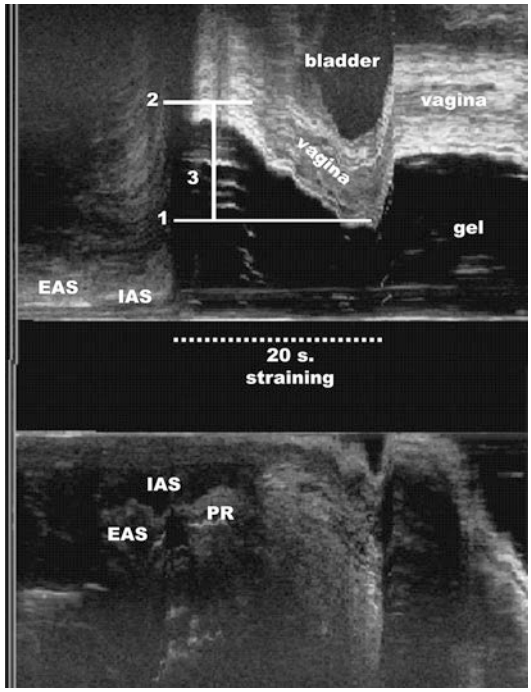

Fig. 18.28 Echodefecography – using gel into the rectum. Patient with grade III rectocele. *Line 1* = Initial straining position; *Line 2* = point of maximal straining. The distance between both lines determines the rectocele size. *EAS* external anal sphincter, *IAS* internal anal sphincter, *PR* puborectal

The distance between the two vaginal wall positions determines the size of the rectocele. Intussusception is clearly identified by observing the rectal wall layers protruding through the rectal lumen. No classification is used to quantify Intussusceptions (Fig. 18.29). Grade II or III sigmoidocele/enterocele is recognized when the bowel is positioned below the pubo-coccygeal line (on the projection of the lower rectum and upper anal canal).

Dynamic ultrasound scanning is a helpful tool in the evaluation of patients with obstructed defecation as it clearly shows the anatomical structures and mechanisms involved on defecation. It also demonstrates the anal canal anatomical integrity and it is able to detect sphincter injury with high spatial resolution. In addition, the cube image acquired during the automatic scan is recorded in real-time for subsequent analysis as may be necessary in many cases. It is quick, inexpensive, and well tolerated by patients without exposure to radiation.

11.2 3-D Transvaginal and Transrectal Ultrasonography (TTUS)

Dynamic ultrasonography combining transvaginal and transrectal approaches and using a 3-D biplane transducer has been used in the diagnosis of anismus, rectocele, enterocele/sigmoidocele, and intussusception in women with obstructed defecation syndrome.

11.2.1 Techniques

TTUS is performed using a biplane transducer (type 8848, B-K Medical, Herlev, Denmark), 22 mm in diameter, with a 60 mm linear array and a frequency of 12 MHz. The transducer has two perpendicular arrays that could be used for either a midsagittal view (linear array) or an axial view (transverse array with a field of view up to 180°) of the pelvic floor. An external mover is

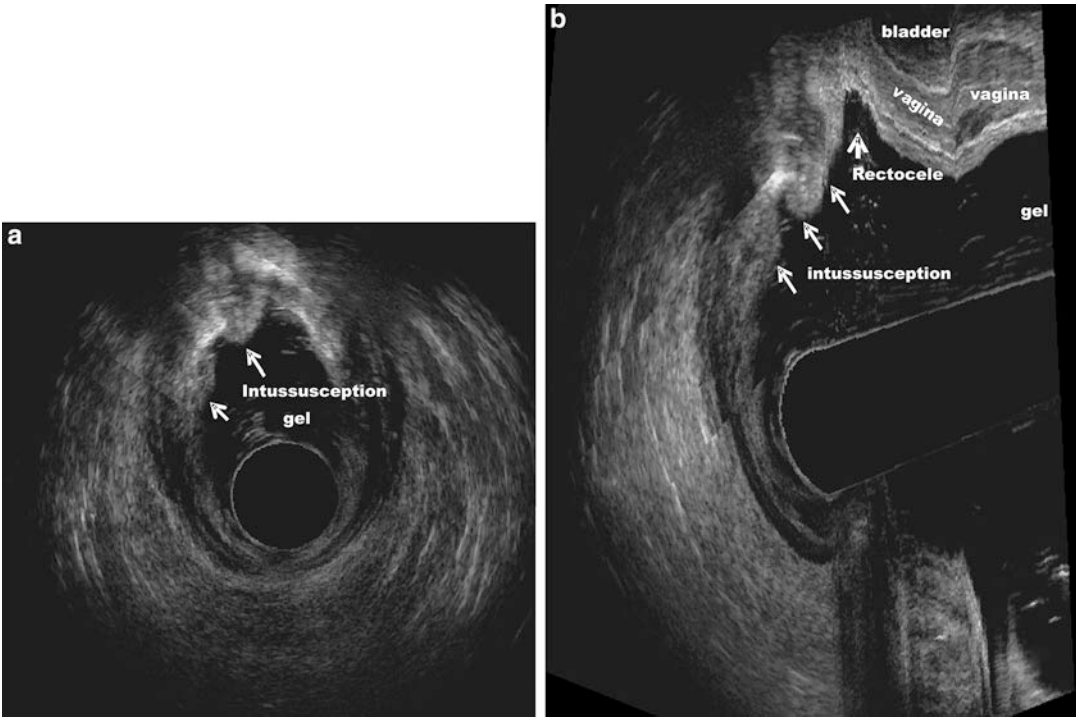

Fig. 18.29 Echodefecography – using gel into the rectum (**a**) (Axial plane) – Anterior intussusception (*arrows*). (**b**) (Sagittal with coronal plane) – Rectocele associated to anterior intussusception (*arrows*). *EAS* external anal sphincter, *IAS* internal anal sphincter, *PR* puborectal

connected to the transducer via a magnetic clip-on collar that, when activating the move, could sample a volume that covers a 180° view in 30 s (Fig. 18.30).

A rectal enema is administered, the patients is placed in the left lateral position and given the instructions required for the examination. After injection of 120–180 mL gel into the rectal ampulla, scanning is performed by means of two combined approaches and three automatic scans, with analysis in the axial and sagittal plane. The scans use spacing of 0.66° and last 30 s each. The duration of the combined procedure is approximately 5 min.

– **Transvaginal approach –** The transducer is placed in the vagina in the neutral position. Longitudinal images of the inferior rectum and entire length of the anal canal are visualized and two scans are acquired. The rotation of 180° is performed from 3 to 9 o'clock position.

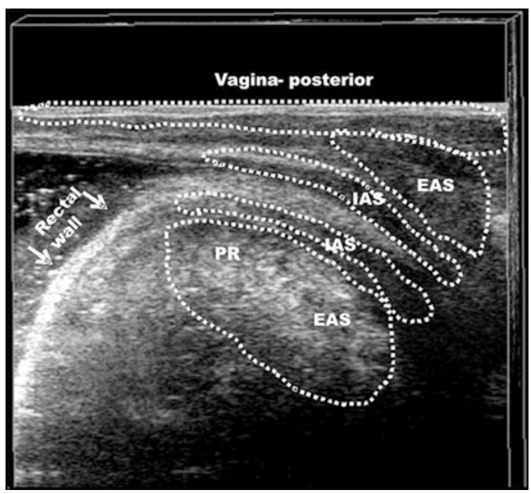

Fig. 18.30 Dynamic 3-D transvaginal and transrectal ultrasonography (TTUS). Using gel into the rectum. Transvaginal approach – normal anatomy. *EAS* external anal sphincter, *IAS* internal anal sphincter; *PR* puborectal

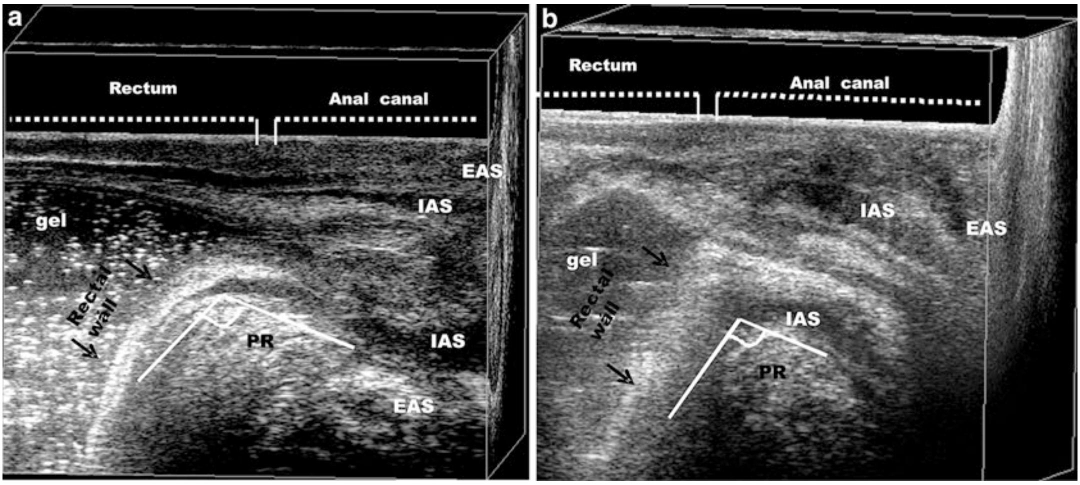

Fig. 18.31 Dynamic 3-D transvaginal and transrectal ultrasonography (TTUS). Using gel into the rectum – Transvaginal approach (**a**) Angle measured at rest position (*lines*). (**b**) Decreased angle (anismus) during straining (*lines*). *EAS* external anal sphincter, *IAS* internal anal sphincter, *PR* puborectal

Scan 1: (at rest) Evaluates the integrity of the IAS and EAS.

Scan 2: the patient is requested to keep at rest during the first 10 s and strain maximally for 20 s. The anorectal angle is measured at the intersection of the longitudinal axis of the anal canal and a line drawn along the posterior border of the rectal wall. The measurements at rest are compared with those made during straining to identify normal relaxation or anismus (including non-relaxation or paradoxical contraction) (Fig. 18.31). Anismus is characterized by a failure to execute the relaxation of the PR-EAS muscles that is required for a successful defecation. Thus, anismus is recorded when the anorectal angle fails to open during straining compared to the anorectal angle at rest. In cases with normal relaxation, the angle increases.

Rectal intussusception is identified by observing the infolding of the rectal wall into the rectum.

– **Transrectal approach –** The rectum is refilled with ultrasound gel whenever the patient eliminates the gel in scan 2. The transducer is placed in the rectum and positioned at 7.0 cm from the anal verge. The patient is requested to keep at rest position during the first 10 s, and then strain maximally for 20 s.

The purpose of scan 3 is to visualize and quantify all anatomical structures and any functional disorders such as rectocele, intussusception, or enterocele/sigmoidocele grade II or III.

The rectocele size is measured by drawing two parallel horizontal lines along the posterior vaginal wall, one in the initial straining position and one at the point of maximal straining, when the anterior rectal wall and upper anal canal are maximally distended, bulging into the vaginal lumen. In normal patients, the posterior vaginal wall displaces the lower rectum and upper anal canal inferiorly and posteriorly but maintains a straight horizontal position during defecatory effort (Fig. 18.32). The distance between the two vaginal wall positions is used to determine the size of the rectocele which is classified as grade I (<6.0 mm), grade II (6.0–13.0 mm), or grade III (>13.0 mm) (Fig. 18.33).

Rectal intussusception is confirmed by observing the infolding of the rectal wall into the rectum (anterior part of the rectal circumference).

Enterocele/sigmoidocele grade II or III is diagnosed if the bowel bulges downward to the pelvis,

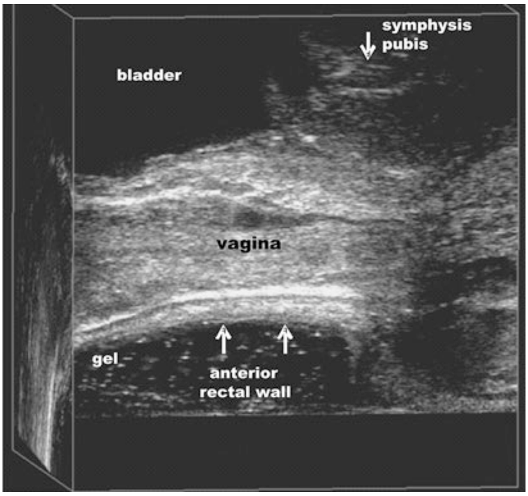

Fig. 18.32 Dynamic 3-D transvaginal and transrectal ultrasonography. Using gel into the rectum – Transrectal approach – the vagina maintains a straight horizontal position during defecatory effort. Patient without rectocele

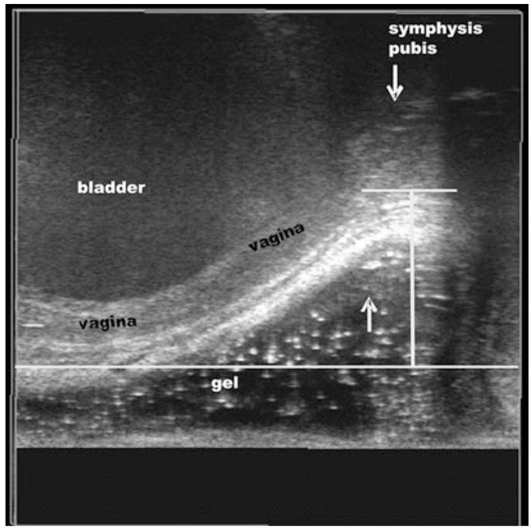

Fig. 18.33 Dynamic 3-D transvaginal and transrectal ultrasonography. Using gel into the rectum – Transrectal approach Patient with rectocele (classified as grade II). Line 1 and 2 = 2 vaginal wall positions; Line 3 = depth of the rectocele

between the posterior vagina and anterior lower rectal wall or anal canal.

Murad-Regadas et al. showed the ability of this new imaging technique to assess the posterior pelvic floor and sphincter muscles in patients with obstructed defecation syndrome (ODS) with high rates of concordance and accuracy compared with echodefecography in the diagnosis of anismus, rectocele, enterocele/sigmoidocele, and intussusception in women with ODS. This type of transducer covers half of the circumference, and it is necessary to combine the transvaginal and transrectal approaches for complete evaluation. However, the technique is simple and quickly performed (approximately 5 min). One limitation of TTUS is that it cannot be used in patients who have never had intercourse or in male patients with obstructed defecation symptoms. As the transvaginal approach is not able to measure the anorectal angle for diagnosing of anismus in such patients, it is not completely effective technique to evaluate the posterior compartment dysfunctions related to obstruction defecation syndrome (Murad-Regadas et al. 2014).

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Anorectal Manometry

19

Filippo Pucciani

Contents

Abstract

At the present time, anorectal manometry is extensively used to identify the pathophysiological mechanisms of obstructed defecation, fecal incontinence, and Hirschsprung disease. Function of anal sphincter complex, rectoanal reflexes, rectal compliance, and rectal sensation is detected. Anorectal manometry generates useful diagnostic data that assist also in the selection of appropriate therapy.

Diagnostic work-up of fecal incontinence and obstructed defecation is based on both imaging techniques, to discover abnormalities of anorectal structural integrity, and on functional instrumental studies, to evaluate the neuromuscular function of anorectum. Anorectal manometry (AM) is used to study function of anal sphincter complex, rectoanal reflexes, rectal compliance, and rectal sensation, but manometric data must be supplementary to those from other anorectal morphological and functional techniques to make a diagnosis. Actually, the clinical utility of anorectal manometry is limited by the relative absence of standardization of test protocols and by the lack of universally accepted normative data from a large number of healthy individuals. Notwithstanding these limitations, anorectal manometry is considered as a useful test for diagnosis and management of fecal disorders.

Routine diagnostic manometry (Azpiroz et al. 2002) explores all continence mechanisms and entails the following:

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– Exploration of the anal sphincter apparatus, including smooth and striated components. **Anal resting pressure** (ARP) reflects the tonic activities of both the internal anal sphincter (IAS) and external anal sphincter (EAS); several studies attribute approximately 55 % of ARP to the IAS, 15 % to the vascular anal cushions, and the remaining 30 % to the EAS (Lestar et al. 1989). **Maximal voluntary contraction** (MVC) is the squeeze pressure obtained by asking the patient to maximally contract the anus; it reflects the contractile activity of the external anal sphincter.
– Evaluation of the **rectoanal inhibitory reflex** (RAIR). RAIR is the reflex inhibition of internal anal sphincter tone that is elicited by distending a rectal balloon with different volumes of air. It is part of the sampling reflex responsible for triggering the impulse to defecate (Martelli et al. 1978; Kumar et al. 1990). Transient relaxation of the internal anal sphincter allows rectal stool contents to come into contact with specialized sensory organs in the upper anal canal; typifying of rectal content alerts the patient to discharge flatus or to defecate.
– Detection of rectal sensation. Volumetric perception of fecal mass is reproduced by distending a rectal balloon with increasing volumes (AGA 1999). **Conscious rectal sensitivity threshold** (CRST) is the lowest volume of air that evokes the first sensation; **constant sensation** (CS) is the volume that calls to stool; **maximum tolerated volume** (MTV) measures the threshold volume for urgency to defecate and for pain.
– Monitoring of **rectal compliance**. Rectal compliance, as determined by the pressure/volume ratio at several different distending volumes, reflects tonic adaptation of rectal wall (rectum distensibility) to the incoming fecal load (Madoff et al. 1990).

Traditional anorectal manometry is performed using perfused or solid state catheters, but none of them can measure circumferential pressures simultaneously throughout the anal canal and in the rectum. High-resolution manometry (HRM) catheters can do so and are increasingly used to evaluate anorectal functions in clinical practice (Noelting et al. 2012). A HRM catheter comprises circumferential sensors, usually at 6-mm intervals along the anal canal, and at least two sensors in the rectal balloon. At each level, 36 circumferentially oriented pressure-sensing elements detect pressure using proprietary pressure transduction technology. Pressures are then averaged to obtain a mean pressure measurement at each level and pressures graphic is displayed on a colorimetric scale ranging from blue, for the lowest pressure values, to violet for the highest pressures. A simultaneous comparison between HRM and anorectal perfused manometry showed that HRM is highly correlated with water perfused manometry measurements and provides greater anatomic detail (Jones et al. 2007; Vitton et al. 2013). A few years ago, 3-D high definition anorectal manometry (HDAM) was introduced, studying pressure topography (Cheeney et al. 2011). HDAM can provide knowledge on the puborectalis, internal and external anal sphincters, and can show how they mediate the rectoanal inhibitory reflex, sensorimotor responses, and the spatiotemporal orientation of these muscles. In addition, anal sphincter defects can be mapped and readily detected using 3-D technology (Lee et al. 2013).

Regardless of monitoring typology, anorectal manometry provides objective information about the mechanisms of fecal continence. For this reason, indications for anorectal manometry are fecal incontinence, obstructed defecation and, in pediatric age, Hirschsprung disease (AGA 1999).

For better understanding of these topics, the use of anorectal manometry will be described according to each individual fecal disorder.

1 Fecal Incontinence

Fecal incontinence is defined as failure to control the elimination of stool and/or flatus recurring for >3 months (Miner 2004; Pucciani 2013). Use of anorectal manometry is mandatory in incontinent patients who undergo work-up studies of fecal incontinence after failure of conservative treatment by drugs, high residual diet, and irrigation systems.

An evaluation of anal pressures is little or no useful because resting and squeezing pressures are generally lower in incontinent patients independently of sphincteric status, with or without sphincter defects. Low anal resting pressure refers to internal anal sphincter dysfunction and may be a manometric marker of passive incontinence; low maximal voluntary contraction is related to external anal sphincter dysfunction and often patients have urge incontinence with loss of stool because of the inability to suppress defecation. Unfortunately, manometry has wide variations in normal pressure that can differ with age and sex so manometric results need to be interpreted carefully along with other findings and symptoms. On the contrary, rectal sensation study and rectal compliance evaluation are decisive in identifying a sensorimotor rectal dysfunction that is the functional report in patients, for example, who are affected by fecal incontinence after sphincter-saving operations (Pucciani et al. 2008). A significant decrease or loss of rectal sensation (> CRST, > CS) may contribute to fecal incontinence by impairing the recognition of impending defecation. When stool enters the rectum, the perception of rectal distension gives the conscious stimulus to contract the anal sphincter to preserve continence; if stool is not perceived, the contractive voluntary response is not elicited and fecal incontinence may occur. Patients with diabetes mellitus (Wald and Tunuguntia 1984) and multiple sclerosis (Caruana et al. 1991) may exhibit this pathophysiological mechanism of incontinence. The rectoanal inhibitory reflex, the transient decrease in resting anal pressure in response to rapid inflation of a rectal balloon, often cannot be elicited when anal pressures are very low (<10 mmHg). Therefore, in some incontinent patients with low ARP it is not possible to judge whether the reflex is present or absent, normal or not. However, there are some reports on RAIR modifications in patients affected by fecal incontinence. One ambulatory manometric study showed that abnormal transient internal anal sphincter relaxation might lead to fecal soiling and pruritus ani (Farouk et al. 1994). The duration of RAIR is longer in patients affected by idiopathic fecal incontinence than in controls; a prolonged contraction time, with a slow return to the prestimulation values, is the typical sign (Pucciani et al. 1997). A longer RAIR impairs the fecal continence mechanism. When small amounts of stool elicit RAIR with a prolonged contraction time in patients with a poor external anal sphincter recruitment and in presence of a conscious rectal sensitivity threshold higher than that of RAIR threshold, fecal passive incontinence may occur. A low MTV, often combined with a defective rectal compliance (>0.50 mmHg/ml), suggests the patient has insufficient rectal volumetric capacity with an impaired adaptation of the rectal wall to endoluminal content. It is the pathological and functional substratum of impaired continence after both low anterior rectal resection and coloanal anastomosis (Pucciani et al. 2008).

Anorectal manometry is a diagnostic testing which may provide information that guides the management of fecal incontinence. Rehabilitative treatment is the first-line conservative therapy of incontinence after the failure of medical treatment (Norton and Kamm 2001) and multimodal rehabilitation may be used as rehabilitative protocol (Pucciani et al. 2003). The algorithm for this rehabilitation management is based on manometric reports. Low anal resting pressures or weak maximal voluntary contraction indicate the patient's need of biofeedback and pelviperineal kinesitherapy. Volumetric rehabilitation (sensory retraining) is indicated for disordered rectal sensation or impaired rectal compliance. Electrostimulation is only a preliminary step when patients need to feel the anoperineal plane. The usual procedural sequence is (1) volumetric rehabilitation, (2) electrostimulation, (3) pelviperineal kinesitherapy, and (4) biofeedback. Their combination is suggested by manometric data. Clinical outcome of multimodal rehabilitation is good: 89 % of patients has significant improvement of Wexner incontinence score. Furthermore, anorectal manometry can help to select candidates who are in need of surgery for fecal incontinence. Low anal resting pressures (<10 mmHg) and low maximal voluntary contraction (<40 mmHg) are considered cut-off values for overlapping sphincteroplasty (Ternent et al. 1997). The same cut-off values identify those patients with rectal prolapse who are at high risk for postoperative incontinence, modifying the surgical strategy of simple correction of prolapse (Yoshioka et al. 1989).

In conclusion, anorectal manometry can be considered an important tool in the diagnostic work- up of fecal incontinence. It can offer decisive data for understanding the pathophysiology of fecal incontinence and can suggest ways in which the therapeutic strategy should be modified.

2 Obstructed Defecation

Obstructed defecation as identified by Bartolo and Roe (1986) is broadly defined as the inability to evacuate contents from the rectum (Khaikin and Wexner 2006) and is accompanied by symptoms of dyschezia and a subjective sensation of anal blockage during defecation (Pucciani et al. 2011). Outlet pelvic obstruction with anorectal dysmotility, caused by organic or functional diseases, is the main pathophysiological mechanism (Andromanakos et al. 2006). Mechanical causes include rectocele, rectoanal intussusception, descending perineum syndrome, solitary rectal ulcer syndrome, mucosal rectal prolapse, enterocele, and sigmoidocele. Disorders of rectal sensation and pelvic floor dyssynergia are the functional diseases. At the present time, AM is extensively used to identify the pathophysiological mechanisms of anorectal obstruction in constipated patients, but its clinical usefulness is debated. Rasmussen et al. reported that standard AM did not show any differences between constipated patients and controls (Rasmussen et al. 1993), whereas others state that AM is a necessary diagnostic step in chronic constipation (Siproudhis et al. 2009; Rao and Singh 2010). AM can provide essential information about the rectoanal function defects involved in the physiopathology of obstructed defecation, including increased pressure in the anal canal, inappropriate increase or less than 20 % relaxation of basal resting pressure during attempted defecation (straining test), rectoanal inhibitory reflex defects, lower rectal sensitivity, and increased rectal compliance (Bove et al. 2012). In any case, manometric data alone do not provide sufficient grounds for the diagnosis. Consequently, a comprehensive evaluation of anorectal function is necessary and should include tests to evaluate various aspects of defecation, including the balloon expulsion test and imaging techniques.

Anal resting pressure measurements can identify those constipated patients (25.3 %) who exhibit a significant impairment of anal tone (Pucciani and Ringressi 2012). For example, some patients (11.6 %) have a low mean anal resting pressure, and lower anal pressure profiles are positively related to grade 3 pelvic organ prolapse quantification (POP-Q) (ρ_s 0.63; $P < 0.01$). Nevertheless, anal hypertonia may be detected in some other patients (5.8 %). These reports confirm the phenotypic variability of OD, suggested by Bharucha et al. (2005). Increased anal resting pressures are the manometric landmark of anal fissure (Opazo et al. 2013). Some neurologic diseases with anal involvement, such as spinal cord lesions, Parkinson disease and generalized dystonia have this manometric sign (Jost et al. 1999). Increased sphincter tone does not differentiate between the different muscles involved, the internal or external anal sphincter, different kind of external anal contractile dysfunction, the effect of primary dyscoordination, the expression of spasticity or manifestation of anal dystonia. Therefore, manometric hypertonia needs to be integrated with other techniques in order to have appropriate diagnostic value. A positive straining test is the manometric sign of pelvic floor dyssynergia but also asymptomatic and incontinent subjects can have this manometric feature. The *type I pattern*, characterized by both adequate propulsive intrarectal forces (intrarectal pressure $\geq$ 45 mmHg) and increased anal pressure, and the *type III pattern*, characterized by increased intrarectal pressure ($\geq$45 mmHg) with absent or insufficient ($\leq$20 %) relaxation of basal anal sphincter pressure, are defined as dyssynergic defecation (Rao et al. 2004). Anorectal manometry can identify a 47.7 % prevalence of dyssynergic pattern when applied in patients affected by chronic constipation (Videlock et al. 2013). Detection of RAIR is a very important manometric tool. The absence of RAIR is considered diagnostic for Hirschsprung disease with 84.6 % positive predictive value and 91.6 % negative predictive value (Emir et al. 1999). For this reason, AM can be used as a screening test in patients in whom HD is suspected during the neonatal period, but for definitive diagnosis it must be combined with other diagnostic tests such as rectal biopsies. Morphologic RAIR impairment can be detected also in dyssynergic defecation: the residual pressure at the lowest point of reflex may be higher while the duration of reflex shorter than in controls (Pucciani et al. 1998). This manometric report could be expression of an exaggerated rectoanal excitatory reflex that overcomes the inhibitory response during anal sampling. In fact, after rehabilitation, a patient's RAIR becomes normal, and the RAIR parameters show no significant differences in relation to those of controls.

Rectal sensory perception is blunted or absent in most patients with obstructed defecation (Gosselink and Schouten 2001); usually, a significantly higher CRST than in controls is detected and the highest CRST values are showed in patients affected by megarectum (Pucciani and Ringressi 2012). A normal perception of a fecal bolus is determinant to triggering and maintaining defecation. CRST values higher than normal imply alterations in coordinated defecation. The subjective sensation is impaired concerning starting the stimulus for defecation and also the perception of complete emptying of the rectum does not occur. Obviously, the coordinated sensory-motor integration of the rectum is distorted and obstructed defecation occurs. The importance of sensorimotor rectal impairment is confirmed by effects of MTV values and compliance distortions. An increased MTV, considered as an indirect expression of increased rectal capacity, is usually combined with increased rectal compliance. These manometric signs are present in about 20.0 % of constipated patients, all of whom have low pressure/volume values (<0.10 mmHg/ml). This means that the biomechanical properties of the rectal wall are defective and, as a result, tonic adaptation to endoluminal volumes is impaired. The distension of the rectal wall is passive, without resistance, impairing the volumetric sensation of the rectal content because of the failure of a generation of forces: deformations in the rectal wall during distension do not stimulate rectal mechanoreceptors. Thus, a secondary rectal hyposensitivity takes place (Scott et al. 2011).

In conclusion, anorectal manometry is an irreplaceable functional diagnostic technique and there are several reasons to use it in fecal incontinence and obstructed defecation: (1) it can identify which pathophysiological mechanisms may be involved; (2) it provides measurements that may be integrated with those of other diagnostic techniques; and (3) it generates useful data that assist in the selection of appropriate therapy.

3 Cross-References

▶ Diagnosis in Coloproctology: From the General Practitioner to the Tertiary Referral Center
▶ Electrophysiological Study of the Pelvic Floor
▶ Factors Affecting Defecation and Anal Continence
▶ Instruments for the In-Office Diagnosis
▶ Integration of Diagnostics in Proctology: Assessment, Choice of Treatment, and Evaluation of Results
▶ Physiology of the Rectum and Anus

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Electrophysiological Study of the Pelvic Floor

20

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Contents

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1 Introduction

Neurophysiologic testing is not routinely used to assess anorectal disorders. However, it can be useful in at least two instances, the first of which is to assess anorectal disorders in patients with central and peripheral neurologic diseases (e.g., trauma, multiple sclerosis, Parkinson's, diabetes, etc.). Anorectal disorders (incontinence and/or constipation) may be the initial symptom of these pathologies (Bardoux et al. 1997; Abbott et al. 2001) or may even be neurologic in origin. Information provided by neurophysiologic testing may help to determine the prognosis of the disease, and thus the treatment and therapeutic follow-up (Lefaucheur 2006). Secondly, in patients with a known neurologic disease, neurophysiologic testing can be used to discriminate between a bowel dysfunction and a neurologic disease since anorectal disorders in such patients are not necessarily due to a neurologic pathology, but may arise from other pathologies such as an anatomical trauma or a dynamic pelvic floor disorder. The exact mechanism of anorectal disorders in these neurologic patients will inform the therapeutic approach.

This chapter summarizes current knowledge about the various neurophysiologic methods used in assessing anorectal disorders in clinical practice, and positions neurophysiologic testing in the anorectal disorder management algorithm.

2 Pelvic Floor Electromyography (EMG)

Conventional methods are limited to the study of the striated pelvic floor muscle (i.e., external anal sphincter, bulbo(clitorido)-cavernosus, puborectalis). Recordings of the EMG activity of the smooth muscle of the internal anal sphincter have been performed in the past (Sorensen et al. 1994), but have only been used for research purposes since EMG activity is variable and recording conditions are difficult to standardize (Lefaucheur 2006).

2.1 Method

2.1.1 Devices Used for EMG Recordings

While the most accurate information on the pelvic floor muscle is obtained using disposable concentric needle electrodes, many researchers have studied alternative approaches such as surface recording electrodes. Multichannel surface EMG allows many signals from different locations to be recorded and provides accurate and non-invasive innervation assessments (Enck et al. 2004a, b). Unfortunately, it is difficult to interpret surface external anal sphincter EMG recordings. Only a few studies have used surface EMG recordings to determine the underlying cause of fecal incontinence (Enck et al. 2005). However, more recently, Nowakowski et al. created a new algorithm using surface anal sphincter EMG to determine the etiology of fecal incontinence based on objective numerical values rather than on subjective graphic EMG interpretations (Nowakowski et al. 2014). Linking this tool to a computer-aided diagnosis system may lead to the wider use of surface EMG for the assessment of anorectal disorders in the near future.

2.1.2 Needle Placement for EMG Recordings

For anorectal disorders, EMG assessments are usually performed on the external anal sphincter, puborectalis, or bulbo(clitorido)-cavernosus muscle. For anal sphincter recordings, the patient lies in the lateral position and the needle is inserted approximately 1 cm lateral to the anal orifice. The four quadrants of the external anal sphincter are typically examined during EMG. Bilateral examinations are required for suspected lesions of the sacral roots, plexus, and pudendal nerves (Lefaucheur 2006). When a neurogenic pelvic floor lesion cannot be confirmed or excluded using the anal sphincter, EMG recordings of other pelvic floor muscles such as the bulbo (clitorido)-cavernosus or puborectalis muscle are indicated. For the bulbo(clitorido)-cavernosus muscle, the patient lies in a gynecological position, and the needle electrode is inserted into the muscle belly on both sides of the vagina in women and the anal median raphe in men. For the puborectalis muscle, the same approach is used as with the anal sphincter, but the needle is advanced parallel to the anal canal until the tip is 4 cm from the skin (Fowler 1995).

2.1.3 EMG Recording Technique

As with other skeletal muscles, EMG recordings of pelvic floor muscles involve the insertion of an electrode and the recording of abnormal spontaneous activity such as fibrillation potentials and high-frequency spontaneous discharges that provide evidence of active denervation. In addition, motor unit action potential morphology (amplitude, duration, number of phases) and the recruitment of motor units (interference pattern) during maximal voluntary or reflex contraction are analyzed (Lefaucheur 2006). A reduced interference pattern with an increase in the temporal recruitment of motor unit potentials indicates muscle denervation while an increase in the amplitude, duration, and phase of motor unit potentials indicates muscle reinnervation (Fig. 20.1). However, the procedure used for pelvic floor muscle EMG recordings is not the same as the procedure used for other skeletal muscles. First of all, the tonic firing pattern of anal sphincter and puborectalis motor units makes it impossible to produce the electrical silence in the muscles that is required to easily identify abnormal spontaneous activity. Second, the assessment of motor unit recruitment is difficult given the small volume of muscle tissue being sampled and given that it is difficult to reliably measure the force of contraction of the anal sphincter since it does not act on a bony lever through a tendinous insertion. In addition, normal individuals have varying abilities to voluntarily produce a strong contraction while a reduced pattern is most commonly caused by poor needle placement and/or a sub-maximal voluntary

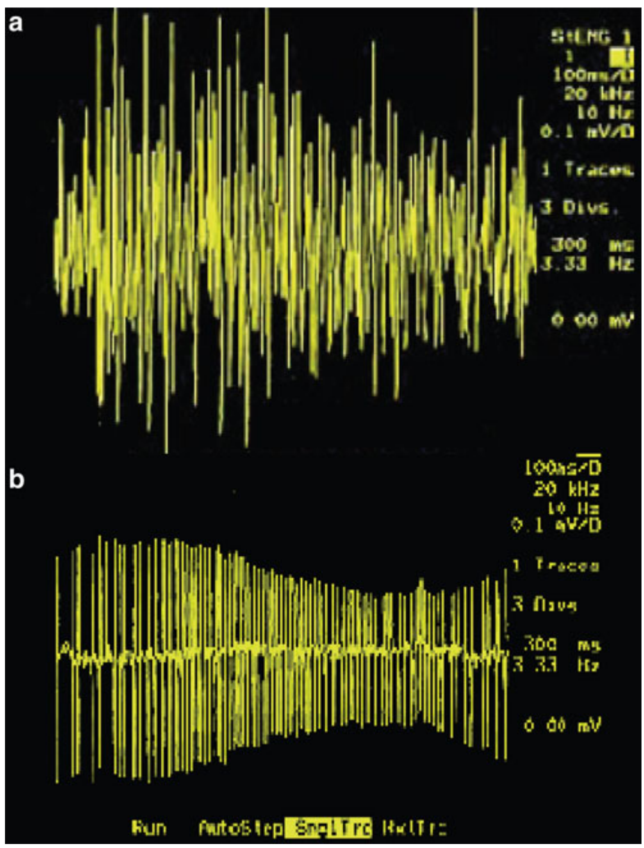

Fig. 20.1 Anal sphincter electromyography. Individual motor unit action potentials can no longer be seen at higher levels of muscle activation. (**a**) Normal (full) interference pattern; (**b**) reduced (neuropathic) interference pattern

contraction. To avoid these constraints and to improve the diagnostic sensitivity of anal sphincter EMG recordings, automated computer-based methods (multiple motor unit potentials and interference pattern analyses) are available with most commercially available EMG devices. After sampling at least 20 different motor unit potentials, a multimotor unit potential analysis is performed to quantify the amplitude, duration, area, and polyphasicity of the potentials (Podnar and Vodusek 1999; Podnar et al. 2000b; Podnar et al. 2002) (Fig. 20.2). The number of motor units, the firing frequencies of the motor units, and the recruitment of new motor units (since an increased contraction force is required) can be determined by analyzing the interference pattern (Gregory et al. 2008a). While these quantitative methods for analyzing anal sphincter EMG recordings are very useful because they are user-friendly and reduce operator-induced bias, they have to take variations in age (Podnar and Vodusek 2000), gender (Podnar and Vodusek 2000), vaginal delivery (Podnar 2000a), and chronic constipation (Podnar and Vodusek 2000) into consideration. The acquisition of a complete set of reference data is also an extremely demanding task due to the difficulty in recruiting asymptomatic subjects. As such, no electrodiagnostic laboratory possesses a complete set of valid normative values (Podnar and Gregory 2010).

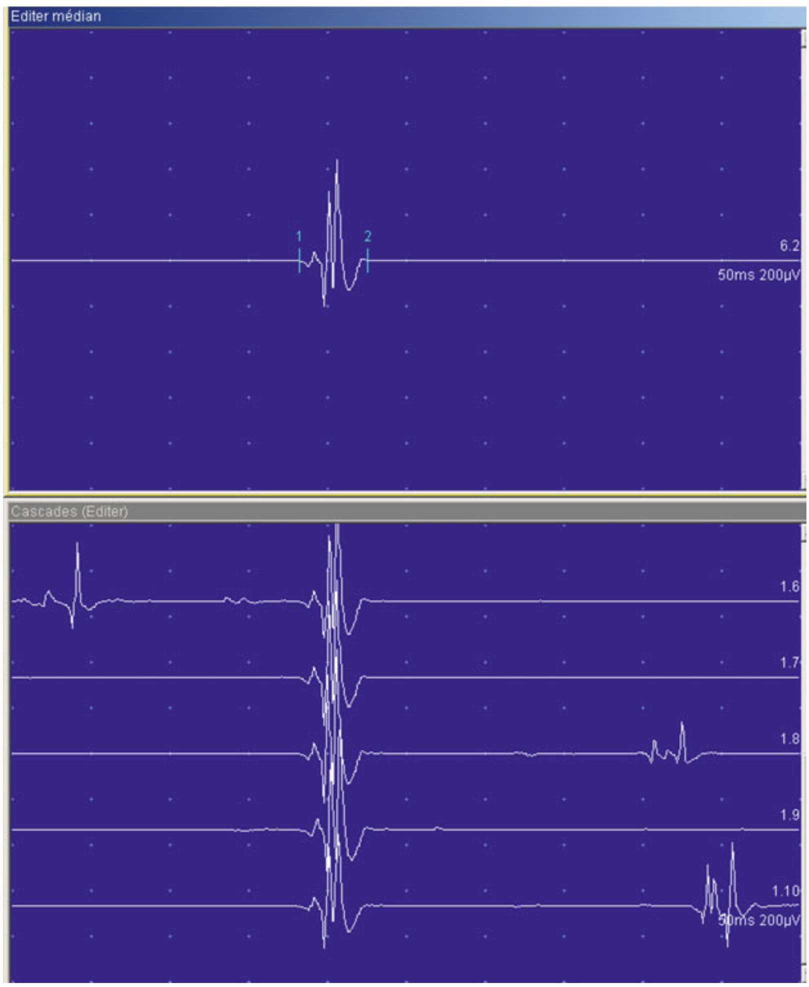

Fig. 20.2 Motor unit potentials analysis. After sampling at least 20 different motor unit action potentials, a multimotor unit potential analysis is performed to quantify the amplitude, duration, area, and polyphasicity of the motor unit action potentials

2.2 Indications

2.2.1 Fecal Incontinence

When anal sphincter weakness is detected clinically and/or at manometry in patients with fecal incontinence, anal sphincter EMG recordings are included in the pelvic floor assessment in order to identify anal sphincter neurogenic injuries resulting from damage to the sacral spinal cord, cauda equina, S2–4 spinal nerves, or pudendal nerve (pudendal neuropathy), and endoanal ultrasound imaging is used to detect anal sphincter anatomic lesions.

Several studies, including two controlled studies, using quantitative EMG analyses have shown that there is a significant prolongation of motor unit duration in the external anal sphincter and puborectalis muscles in fecally incontinent patients (Bartolo et al. 1983; Sorensen et al. 1991). The results of quantitative EMG recordings of the anal sphincter have been correlated with anal pressure during voluntary contractions (Sorensen et al. 1991; Cheong et al. 1995). Gregory et al. performed a quantitative analysis of anal sphincter EMG recordings in patients with fecal incontinence, with or without anal incontinence, after vaginal delivery (Gregory 2008b). The interference pattern analysis of the group of postpartum women with anal incontinence symptoms showed evidence of denervation and subsequent reinnervation (Gregory et al. 2008b). This study and others prompted the American Gastroenterological Association to recommend that concentric needle EMG recordings be used to look for denervation due to peripheral nerve lesions when assessing patients with fecal incontinence (Barnett et al. 1999).

2.2.2 Constipation

Dyssynergia is defined as a paradoxical increase in anal sphincter pressure (anal contraction) of less than 20 % relaxation of the resting anal sphincter pressure or inadequate abdomino-rectal propulsive forces leading to difficult defecation. In two-thirds of subjects, dyssynergia is a consequence of faulty toilet habit, painful defecation, obstetric or back injury, or brain-gut dysfunction (Rao et al. 2005). The diagnostic criteria for dyssynergic defecation are as follows (Rao 2008): patients must satisfy the symptomatic diagnostic criteria for chronic constipation (Rome III) and must show a dyssynergic pattern on manometry, imaging, or EMG of defecation during repeated attempts to defecate. Anorectal dyssynergia is suspected when there is no relaxation (EMG activity stable or higher) of the external anal sphincter and/or puborectalis muscles on EMG testing during attempts to defecate. Surface EMG recordings appear to be more useful than needle EMG recordings in providing evidence of non-relaxation of the anal sphincter or puborectalis muscle during defecation in constipated patients (Pfeifer et al. 1998). Only a few studies have investigated the predictive value of EMG recordings for excluding a diagnosis of pelvic floor dyssynergia in constipation. Bordeianou et al. studied constipated patients with and without pelvic floor dyssynergia and compared the results of anal sphincter EMG and the balloon expulsion test to the results of defecography, which is considered the reference method for diagnosing pelvic floor dyssynergia (Bordeianou et al. 2011). They reported that 84.1 % of the patients with abnormal EMG results did not expel the balloon. However, the presence of these abnormalities, in isolation or together, did not predict the presence of dyssynergia on defecography (Bordeianou et al. 2011). Consequently, it not clear which of the three tests (anorectal manometry, EMG, imaging) most accurately diagnose pelvic floor dyssynergia.

2.2.3 Degenerative Neurological Disease

Multiple system atrophy is a neurodegenerative disease presenting with a combination of Parkinsonian, cerebellar, and autonomic (including cardiovascular, urinary, and anorectal) dysfunction. While it is pathologically defined, there is no definitive clinical diagnostic test. The majority of patients with probable multiple system atrophy have an abnormal sphincter EMG because of the selective loss of anterior horn cells in Onuf's nucleus (Gilad et al. 2001; Palace et al. 1997).

However, patients with idiopathic Parkinson's disease do not show marked sphincter EMG abnormalities. As such, these abnormalities can be used to distinguish multiple system atrophy from idiopathic Parkinson's disease in the first 5 years after disease onset. In contrast, similar sphincter EMG abnormalities are found in some, though not many, patients with dementia with Lewy bodies, pure autonomic failure, progressive supranuclear palsy, or spinocerebellar ataxia type 3 (Winge et al. 2010). The limitations of sphincter EMG recordings should thus be kept in mind. Sphincter EMG recordings associated with sacral autonomic tests are also used as diagnostic tools for autonomic disorders (Winge et al. 2010).

3 Pudendal Nerve Motor Latency Measurement

3.1 Method

Kiff and Swash (1984) developed a stimulating electrode known as St. Mark's pudendal electrode (Fig. 20.3). A bipolar stimulating electrode is mounted on the tip of the gloved index finger, which is inserted into the rectum. Recording electrodes located 3 cm proximally at the base of the fingers pick up the contraction response of the anal sphincter. The ischial spine is located on transrectal examination, and electrical stimuli are applied at that site to stimulate the pudendal nerve where it leaves the pelvis through the greater

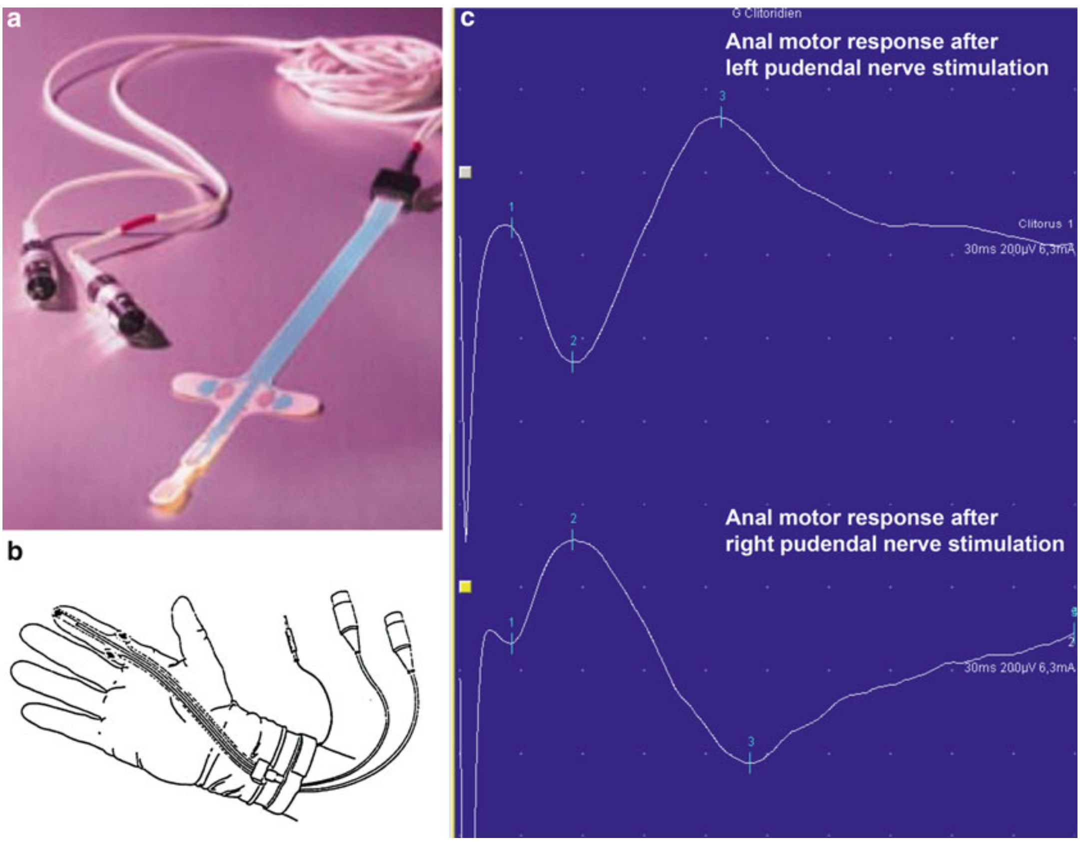

Fig. 20.3 St. Mark's pudendal electrode (**a**). A bipolar stimulating electrode is mounted on the tip of the gloved index finger, which is inserted into the rectum (**b**). Recording electrodes located 3 cm proximally at the base of the finger pick up the contraction response of the anal sphincter (**c**)

sciatic notch and before it branches into the inferior rectal nerve (to the anal sphincter) and the perineal nerve (to the periurethral striated muscle). Fowler (1995) applied a stimulation on both sides of the pelvis and reported that the mean latency of the response from the anal sphincter is 2.1 ± 0.2 ms (Fowler 1995).

The pudendal nerve latency technique is quite reliable (Tetzschner et al. 1997a). However, because of the very short latency value (less than 2.5 ms) and because of a frequent marked motor artifact, alternative stimulation and recording techniques have been developed. In women, the pudendal nerve can be stimulated through the vaginal wall using a St. Mark's electrode, with a recording surface or needle electrode on the anal sphincter (Tetzschner et al. 1997b). The stimulation can also be performed through the anus, but the response of the bulbo(clitorido)-cavernosus muscle must be recorded using a needle electrode in order to avoid the motor artefact and obtain easier-to-interpret responses. Lastly, magnetic shocks can be applied over the sacral roots at the sacral foramina (see chapter below).

3.2 Indications

Pudendal nerve terminal motor latency is the time lapse between the stimulation of the pudendal nerve and the response of the anal sphincter. Terminal motor latency is prolonged if the nerve between the site of stimulation and the muscle is demyelinated, as occurs with mechanical nerve injuries or diabetes (Fowler 1995).

Numerous studies have reported prolonged pudendal nerve terminal motor latencies in various anorectal disorders, including fecal incontinence (Cheong et al. 1995; Lefaucheur 2006), perineal descent (Lefaucheur 2006), and constipation (Vaccaro et al. 1994). Snooks et al. reported that terminal motor pudendal latency was prolonged for 2–3 days after vaginal delivery in 42 % of the women and that the abnormalities were more marked in multiparous women and in those who had a prolonged second stage of labor and forceps delivery. Two months later, the abnormality was resolved in 60 % of the women, but recovery was poorer in multiparous women (Snooks et al. 1984). A follow-up study 5 years later involving some of the multiparous women showed that the occult damage to the pudendal nerve persisted and became more marked with time (Snooks et al. 1990). An initial pudendal nerve injury at the time of the childbirth or during chronic straining at stools, such as with constipation, may explain abnormal pudendal nerve latencies associated with anorectal disorders that worsen with succeeding deliveries and/or repeated straining at stools, with traction on the pelvic floor leading to further stretch-induced injury to the pudendal nerve.

Pudendal nerve latency has been proposed as a predictive factor for the clinical outcome of biofeedback therapy (Leroi et al. 1999) and anal sphincter repair (Gilliland et al. 1998), but not for sacral nerve stimulation (Gallas et al. 2011). However, the conduction velocity of a nerve may have little bearing on its functional integrity. The prevalence of prolonged pudendal nerve terminal motor latency in patients presenting for anorectal neuropathies has been reported to be 20–28 % with unilateral neuropathy and 11–12 % with bilateral neuropathy (Ricciardi et al. 2006; Gurland and Hull 2008). The majority of incontinent patients with intact sphincters have a normal pudendal nerve terminal motor latency (Ricciardi et al. 2006). Bilateral neuropathy, but not unilateral pudendal neuropathy, is associated with diminished sphincter function and higher incontinence scores (Ricciardi et al. 2006). As such, there is no consensus on the significance of pudendal nerve terminal latencies, and the American Society of Gastroenterology does not recommend the use of this test for the routine assessment of patients with anorectal disorders (Barnett et al. 1999). However, the diagnosis of pudendal neuropathy should not be limited to an assessment of pudendal nerve terminal motor latency since pudendal nerve conduction velocity measurements are abnormal only when the largest and most heavily myelinated nerves are lost. This may explain the lack of sensitivity of pudendal nerve terminal motor latency measurements for detecting anal sphincter denervation.

4 Sacral Reflex Latency

4.1 Method

Sacral reflexes are reflex contractions of striated pelvic floor muscles that occur in response to stimulations of the perineum or genital region. One of the reflexes most commonly used in research is the bulbo(clitorido)-cavernosus reflex, which can be elicited by electrical stimulations of the dorsal nerve of the penis or clitoris. A needle electrode is used to record the responses of the bulbo(clitorido-)cavernosus muscle (Fig. 20.4). Following a stimulation of the dorsal nerve, afferent impulses are conveyed via the pudendal nerve to the sacral spinal cord through the posterior roots. After a variable synaptic delay, efferent impulses traveling in the pudendal nerve give rise to contractions of the bulbocavernosus muscle. There are two components to this reflex: the first response, which is used clinically, has a latency of the order of 35 ms while the second later response occurs after approximately 60–70 ms (Fowler 1995).

Sacral reflex latencies are influenced by age and gender (Pradal-Prat et al. 1998). Since it is often easier to elicit the reflex in males than in females, no significance should be given to its absence in women (Fowler 1995). A double-shock stimulation rather than a single shock can

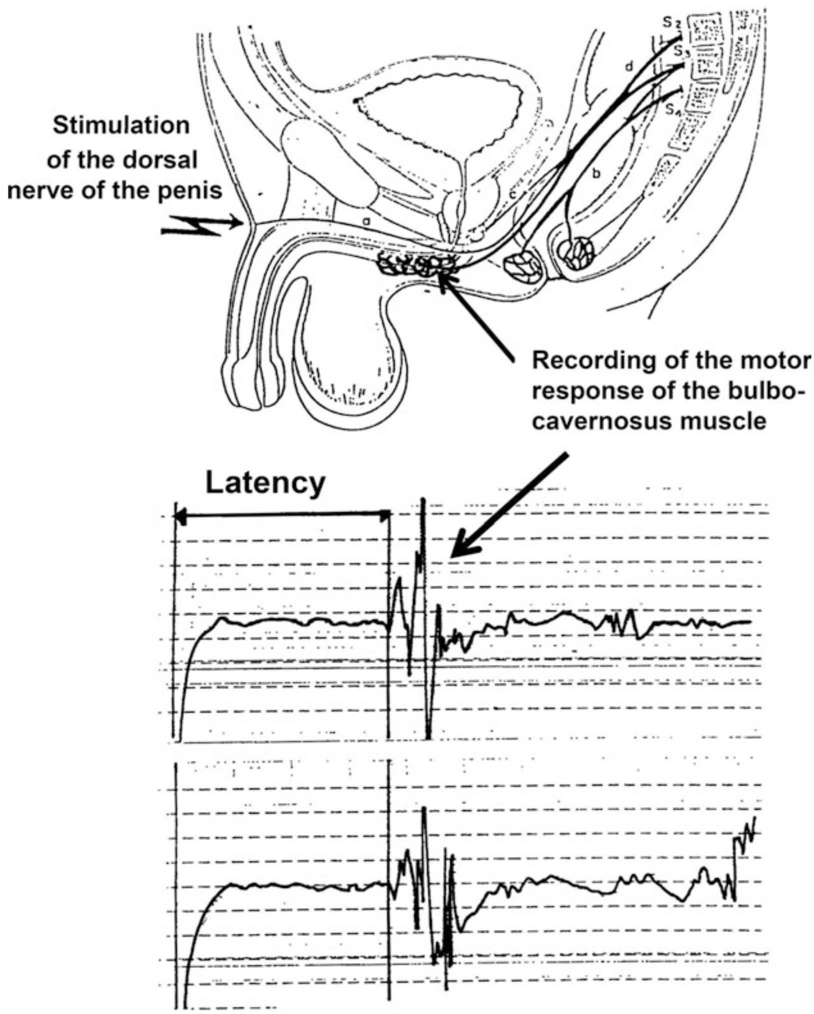

Fig. 20.4 Bulbo (clitorido)-cavernosus reflex. The reflex is elicited by electrical stimulation of the dorsal nerve of the penis or clitoris, and a needle electrode is used to record the response of the bulbo (clitorido-)cavernosus muscle

be used to decrease the rate of failure (Podnar 2014). The sacral reflex latency test should be part of the diagnostic armamentarium for investigating neurogenic pelvic floor disorders. The results, however, should be interpreted with caution since a normal latency does not exclude the possibility of a partial axonal lesion while, on the other hand, an abnormal latency may not be clinically relevant.

4.2 Indications

Sacral reflex latency investigations are indicated for suspected lesions of the conus medullaris or cauda equine, sacral radiculopathies, and sacral plexus. Only extreme pudendal nerve demyelination can cause a significant delay in peripheral conduction leading to an abnormal bulbocavernosus reflex latency (Podnar 2011). The few studies that have investigated the sacral reflex latency of patients with anorectal disorders seem to confirm the usefulness of this test for diagnosing sacral reflex arc lesions (sacral afferent fibers, sacral spinal cord, sacral efferent fibers) in the case of anorectal disorders. Ismael et al. described perineal electrophysiologic findings in 19 women with pelvic floor disorders (urinary and/or fecal incontinence, dysuria and/or dyskesia, sexual dysfunctions) after vaginal delivery (Ismael et al. 2000) and reported no associated lower limb sensory or motor deficits. However, perineal electrophysiologic examinations revealed signs of denervation with abnormal bulbo(clitorido)-cavernosus reflexes in all cases (Ismael et al. 2000). This study highlighted the value of bulbo(clitorido)-cavernosus reflex latency measurements in patients with suspected lumbo-sacral plexopathies but no lower limb deficits. In another study, Podnar assessed the sensitivity, specificity, positive predictive value, and negative predictive value of quantitative concentric needle EMG recordings of the external anal sphincter muscles and neurophysiologic measurements of the bulbo(clitorido)-cavernosus reflex, individually and in combination, for diagnosing sacral neuropathic lesions in 24 women with chronic cauda equine lesions with bladder, bowel, and/or sexual dysfunction (Podnar 2014). Podnar reported a high sensitivity and negative predictive value (98–100 %) and a reasonably high specific and positive predictive value (50–75 %) of the bulbocavernosus reflex associated with anal sphincter EMG recordings for confirming or excluding sacral neuropathic lesions (Podnar 2014).

5 Motor-Evoked Potentials (Central and Peripheral)

5.1 Method

It is possible to study central motor pathway conduction to the external anal sphincter by recording motor evoked potentials (MEPs) by the transcranial magnetic stimulation of the motor cortex (Lefaucheur 2006) (Fig. 20.5). Magnetic shocks can also be applied over the lumbar and sacral roots at the sacral foramina to evoke anal sphincter compound muscle potentials (Lefaucheur 2006). Central conduction along the pyramidal tract is calculated by subtracting the response latency to sacral root stimulation from the total conduction time to motor cortex stimulation (Fig. 20.5). Sacral magnetic stimulation has a number of advantages for measuring peripheral motor conduction time to the anal sphincter since it is less uncomfortable than intra-rectal stimulation and can be used to study the pudendal nerve along its entire length (Remes-Troche et al. 2007).

However, the anal MEP technique requires the use of a double-cone coil rather than a circular coil for the cortical stimulation to obtain interpretable results. The electromagnetic field induced by a double-cone coil is better adapted for stimulating the cortical representation of the anal sphincter, which is deep within the interhemispheric fissure (Lefaucheur 2006). Needle electrodes, surface electrodes, or anorectal pressures can be used to measure the anal sphincter response following cortical or sacral stimulation (Lefaucheur 2006). However, failure rates of 14–25 % have been observed for sacral magnetic stimulation because

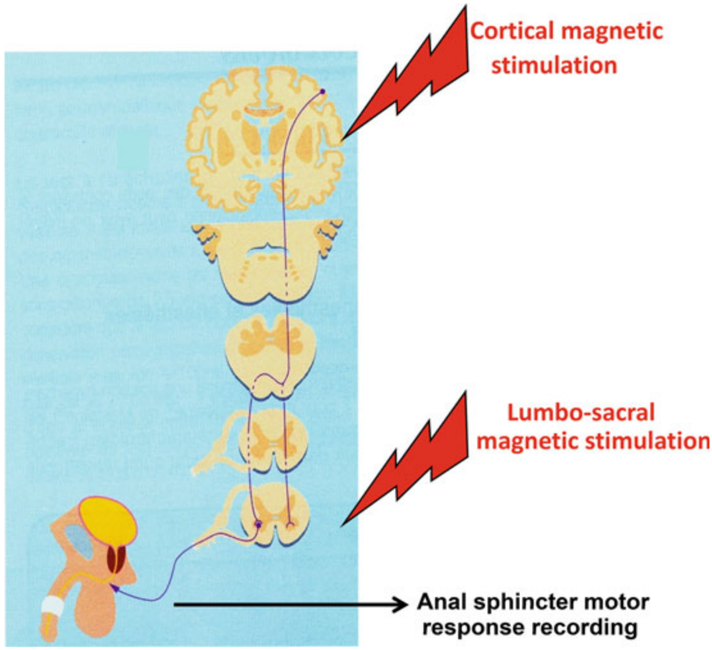

Fig. 20.5 Motor evoked action potentials of the external anal sphincter elicited by transcranial magnetic stimulation of the motor cortex. Magnetic shocks can also be applied over the lumbar and sacral roots at the sacral foramina to evoke anal sphincter compound muscle action potentials. Central conduction along the pyramidal tract is calculated by subtracting the response latency to sacral root stimulation from the total conduction time to motor cortex stimulation

the long recovery period of the stimulus artifact interferes with latency measurements, especially in the case of needle EMG recordings (Jost and Schimrigk 1994; Loening-Baucke et al. 1994; Sato et al. 2000). Intrarectal placement of the ground electrode substantially reduces the stimulus artifact and improves the reliability of the technique (Lefaucheur 2005). Normal MEP values depend on the stimulation (type of coil, stimulation parameters) and recording techniques used.

5.2 Indications

The relevance of investigating MEPs in patients with anorectal disorders remains to be confirmed. Such investigations may facilitate the diagnosis, understanding, and follow-up of anorectal diseases in which the brain-gut axis is involved (Lefaucheur 2006). For example, trans-lumbar and trans-sacral MEPs have been used to reveal significant lumbo-sacral neuropathies in 90 % of subjects with various levels of spinal cord injury (Tantiphlachiva et al. 2011). A study of 65 fecally incontinent patients with no known neurologic disease revealed abnormal anal cortical MEP latencies in 21.5 % of the patients (Paris et al. 2013). More recently, Rao et al. described abnormal lumbo-anal, lumbo-rectal, sacro-anal, and sacro-rectal MEPs in 44 (88 %) of 50 subjects with fecal incontinence (Rao et al. 2014). In these cases, abnormal MEP latencies might reveal undetected lesions of pelvic floor motor pathways that could help in the management of fecally incontinent patients. For example, patients with normal cortical MEP latencies might benefit more from treatments such as biofeedback than patients with abnormal motor responses who are unable to voluntarily contract their external anal sphincter muscle. However, this is pure conjecture and needs to be confirmed by other studies.

6 Somatosensory-Evoked Potentials

6.1 Method

With this test, the pudendal nerve is stimulated and the response is recorded using electrodes located on the scalp (Fig. 20.6). Unlike pelvic floor MEPs, this test does not require any special technical requirements other than the need to record

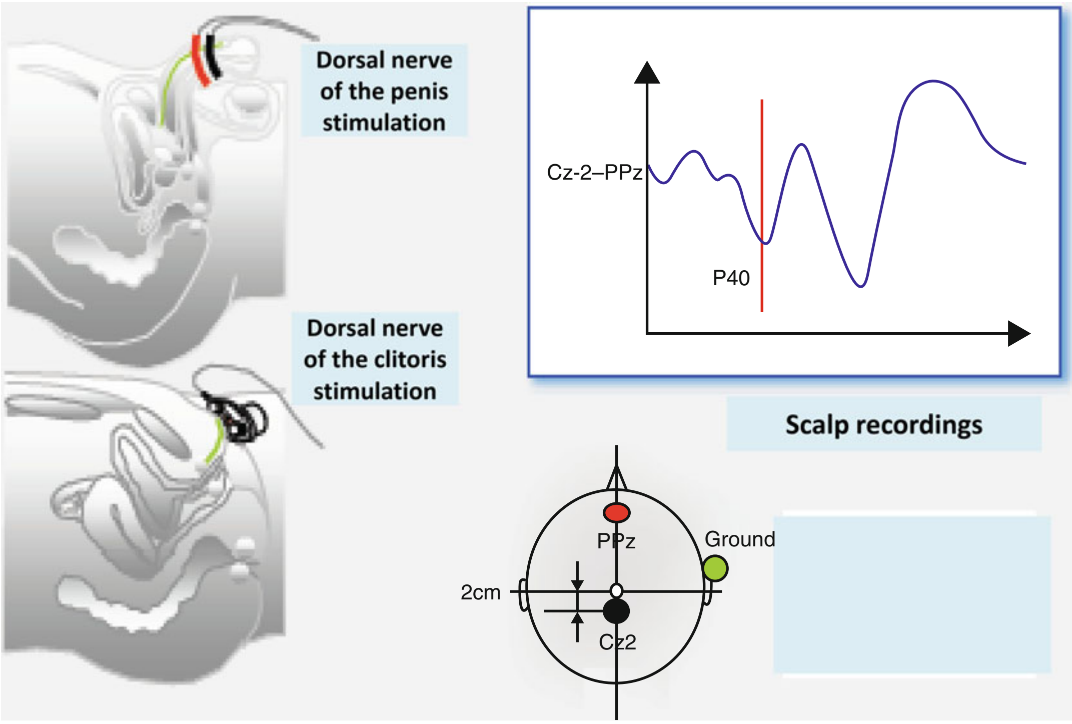

Fig. 20.6 Somatosensory-evoked action potentials recorded in response to the stimulation of the pudendal nerve in men and women

other somatosensory evoked potentials (SEPs). The electrical stimulation of the dorsal nerve of the penis or clitoris is a routine procedure in which a stimulation 2–4 times stronger than the sensory threshold is usually applied. After computer averaging of 100 responses in healthy subjects, the first positive peak is usually clearly defined and highly reproducible. The response is the highest amplitude (0.5–12 lV) at the central recording site (Cz':Fz of the international 10–20 system of electrode placement). SEPs recorded in response to the stimulation of the pudendal nerve in men and women have similar latencies (approximately 40 ms) and waveforms as those following tibial stimulation (Fowler 1995). The response latency is approximately 40–43 ms, but the result depends on the height of the subject (Tackmann et al. 1988).

Stimulations applied to the rectal wall (Speakman et al. 1993) or within the anal canal or at the anal verge (Delechenault et al. 1993) have also been studied, but are more difficult to perform and give less reliable results than stimulations applied to the dorsal nerve of the penis (clitoris).

6.2 Indications

Recordings of SEPs evoked by pudendal nerve stimulation are a very sensitive method for detecting large-diameter sensory nerve fiber lesions in the dorsal column and lemniscal sensory pathways as well as multiple sclerosis-related demyelination (Lefaucheur 2006). SEPs are abnormal in 72–100 % of multiple sclerosis patients (Sau et al. 1999). Recordings of SEPs evoked by posterior tibial nerve stimulation may improve the diagnostic sensitivity of pudendal SEPs since perineal SEP latencies are within the normal range of values but are longer than tibial SEP latencies and can thus be considered as pathologic. In contrast, simply recording SEPs from the tibial nerve may fail to reveal sacral dysfunctions (Lefaucheur 2006).

7 Sensory Thresholds

Various modalities of anal sensation testing have been investigated (Lefaucheur 2006). Several methods for quantifying anal sensations in response to electrical or thermal stimulations have been described (Roe et al. 1986; Miller et al. 1987; Beco et al. 2014). These methods have revealed marked alterations in anal sensation in patients with fecal incontinence. However, such semiobjective testing requires patient cooperation and cannot precisely pinpoint lesions between the sensory receptors and the somesthesic cortex.

8 Sympathetic Skin Potentials

Perineal sympathetic skin potentials (SSPs) can be used to assess the function of thoracolumbar sympathetic fibers innervating the perineal skin. An electrical pulse delivered to a limb (median nerve) or the sacral region can be used to elicit sacral SSPs. Responses, which have latencies of 1.5–2.3 s in control subjects, are recorded using surface electrodes positioned on the penis or labia. Sympathetic skin reflexes are sudomotor responses involving sensory afferents, unknown central pathways, and specific sympathetic efferents. Peripheral and central lesions of the sympathetic pathways can alter SSPs. Peripheral abnormalities can be differentiated from central lesions by determining the anatomic distribution of SSP abnormalities. Perineal SSP abnormalities are better interpreted by comparing them with palmar and plantar results (Lefaucheur 2006). SSPs can rapidly habituate and are critically dependent on a number of factors, including skin temperature. Since responses are variable in shape, only a lack of response can be considered abnormal. There are too few studies on the use of SSPs for assessing anorectal disorders to recommend the use of this approach in a clinical setting.

9 Pelvic Floor Electrophysiologic Studies and Anorectal Disorders

9.1 Which Tests for Which Neurologic Lesions?

Patient histories, clinical findings, and pelvic floor assessments (anorectal manometry, urodynamics) should provide a diagnosis of a neurogenic anorectal disorder and serve as a guide for further neurophysiologic investigations. The choice of pelvic floor explorations in turn should be guided by the type of pathology being investigated. If a central nervous system pathology is suspected (multiple sclerosis, spinal cord lesions, etc.), techniques for exploring the central nervous system such as SSPs and MEPs should be used. If a peripheral nervous system lesion is suspected (cauda equine syndrome, lumbosacral plexopathy, pudendal neuropathy, etc.), sacral reflex latency, lumbar and sacral MEPs, terminal motor latency, and pelvic floor muscle EMG recordings would be more appropriate. Both peripheral (EMG, sacral reflexes, and sacral MEPs) and central explorations (MEPs and SEPs) can be used to diagnose conus medullaris lesions, while SSPs can be used to evaluate pelvic floor autonomic functions in cases of suspected dysautonomia. In case of suprapontine disease, EMG performed during straining at stools may document anorectal dyssynergia in patients with Parkinson's disease and multiple system atrophy. Needle EMG recordings of the external anal sphincter, including quantitative motor unit potential analyses, are clearly indicated for patients with suspected multiple system atrophy, particularly in its early stages.

Whatever the type of neurologic pathology being investigated, electrodiagnostic assessments should be performed by experienced clinical neurophysiologists, and the results should be interpreted with caution, with the clinical context in mind. In addition, neurophysiologic investigations should not be restricted to single test, as is too often the case for the diagnosis of pudendal neuropathy, which is restricted to pudendal nerve latency measurements. For example, Podnar et al. have shown that neurophysiologic testing to diagnose cauda equina lesions is markedly more sensitive when a combination of EMG recordings and sacral reflex measurements are used (Podnar 2014). All neurophysiologic tests are complementary and not mutually exclusive (Lefaucheur 2006). For example, pudendal nerve latency or sacral MEP measurements combined with pelvic floor muscle EMG recordings can be used to diagnose pudendal neuropathy and to detect demyelinating neuropathy and axonal lesions.

9.2 Are Neurophysiologic Investigations Useful for Detecting Anorectal Disorders?

It is well known that urinary disorders may reveal neurologic pathology such as multiple sclerosis (Miller et al. 1965; Beck et al. 1994). A neurologic origin of an anorectal disorder (incontinence and/or constipation) of recent appearance and with no known underlying mechanisms should not be excluded, particularly if there are associated urinary and/or sexual disorders. Sun et al. suggested that neurologic injuries may be suspected in 10 % of patients referred for fecal incontinence and, more generally, for anorectal symptoms based on anorectal manometric abnormalities (Sun et al. 1990). In addition, Bardoux et al. reported four cases of patients who initially consulted for anorectal symptoms but who were eventually diagnosed with a neurologic disorder (multiple cerebral infarction, multiple system atrophy, amiodarone-related neuropathy, spinal neurinoma) (Bardoux et al. 1997). These four patients accounted for approximately 2 % of all patients consulting for anorectal disorders during the same period. When a neurologic disease is suspected, neurophysiologic assessments of the pelvic floor can be helpful for determining the pathophysiologic mechanism and the anatomic location of the lesion (i.e., peripheral neuropathy, sacral spine lesion, or suprasacral central lesion). Complementary information from neurologic examinations can be used to orientate further investigations (computerized tomography, magnetic resonance imaging, lumbar puncture, etc.) in order to confirm the neurologic diagnosis and determine the best treatment.

When the neurologic disease is known, neurophysiologic investigations could help to determine the role of the neurologic disease in the anorectal disorder since anorectal disorders in such patients are not necessarily due to a neurologic pathology but may be caused by other pathologies such as an anatomic trauma or a dynamic pelvic floor disorder. The exact mechanism of the anorectal disorder in these neurologic patients will help to determine the prognosis and therapeutic approach.

9.3 Particular Indications

9.3.1 Fecal Incontinence

Fecal incontinence is a common condition that can be caused by an external anal sphincter dysfunction secondary to compromised structural integrity and/or a neurologic disease. The importance of peripheral nerve lesions, such as pudendal neuropathy, in fecal incontinence has been highlighted by histometric and EMG studies of the external anal sphincter (Snooks et al. 1985).

Why attempt to diagnose pudendal neuropathy as the origin of sphincter incompetence, apart from identifying the cause of fecal incontinence? The diagnosis of a neuropathy may inform the therapeutic approach. For example, when medications for treating fecal incontinence fail to improve patient outcomes, biofeedback can be used as an alternative conservative therapy. However, pudendal neuropathy may result in a poor prognosis for biofeedback therapy (Leroi et al. 1999). In such cases, it could be useful to determine whether the patient is a good candidate for pelvic floor rehabilitative techniques. Anal sphincteroplasty should be considered for patients with fecal incontinence who do not respond to conservative therapies, including biofeedback, and who have a significant sphincter lesion.

While short-term improvements in fecal incontinence have been reported in up to 85 % of patients following anal sphincteroplasty, continence deteriorates thereafter, and there is a 50 % failure rate after 40–60 months (Cheung and Wald 2004). Anal sphincteroplasty is thus generally reserved for patients in whom fecal incontinence and an anal sphincter injury are diagnosed shortly after a perineal trauma (Wald et al. 2014). Pudendal neuropathy as evidenced by a prolonged pudendal nerve terminal motor latency is often cited as a factor for a poor prognosis of sphincter repair. However, this is still a matter of debate (Goetz and Lowry 2005). Some investigators have reported that prolonged pudendal nerve terminal motor latency is a strong predictor of poor postoperative function while others have reported that there is no statistically significant difference in postoperative continence in patients with or without a prolongation of pudendal nerve terminal motor latency (Goetz and Lowry 2005). As such, preoperative pudendal neuropathy diagnosed based on pudendal nerve terminal motor latency cannot be used to accurately predict postoperative function after sphincteroplasty and should not be used to exclude patients from surgery (Goetz and Lowry 2005). If anal sphincteroplasty is not indicated, sacral nerve stimulation (SNS) should be considered for patients with fecal incontinence. In recent years, SNS has become an increasingly important tool for managing patients with fecal incontinence. Gourcerol et al. were the first to suggest that patients with a prolonged sacral reflex and/or fecal incontinence of neurologic origin are more likely to have a favorable outcome with SNS, at least in the short-term (Gourcerol et al. 2007). However, this has not been confirmed by other studies that failed to identify any predictive factors, including pudendal neuropathy, for the mid- and long-term outcomes of SNS (Roy et al. 2014).

9.3.2 Anal Pain

Pudendal canal syndrome, which was first described by Amarenco in 1987 (Amarenco et al. 1987), is caused by the compression or stretching of the pudendal nerve in Alcock's canal. The complete syndrome presents with anal incontinence, pain, hypo or hyperesthesia, and urinary incontinence (and impotence in males) (Beco et al. 2004). The cause of pudendal canal syndrome is not always clear, but the medical histories of patients with this syndrome often include compression (biking, sitting for long periods, hematoma, etc.) or stretching (descending perineum, surgery, delivery, etc.) of the pudendal nerve in Alcock's canal (Amarenco et al. 1987). Diminished perineal sensation, a weak or absent anal reflex, reduced EMG activity of the external anal sphincter, and increased pudendal nerve terminal motor latency are used to confirm a diagnosis of pudendal canal syndrome before surgery (Beco et al. 2004). This syndrome is surgically treated by opening Alcock's canal (trans-perineal approach), with or without sectioning of the sacro-spinal or sacro-tuberous ligaments (trans-gluteal), to provide enough space to allow the pudendal nerve to relax and/or to relieve compression. EMG investigations of pudendal canal syndrome, which include pudendal nerve terminal motor latency measurements, anal sphincter muscle EMG recordings, and sacral reflex assessments, are limited by the fact that the techniques do not directly determine the pathophysiologic mechanisms of the pain but rather assess structural alterations to the pudendal nerve such as demyelination or axonal loss. In addition, only direct or reflex motor innervation is assessed, whereas sensory nerve conduction studies are more sensitive for detecting nerve compression. Lastly, electrophysiologic investigations cannot differentiate entrapment from other causes of pudendal nerve lesions (stretching induced by surgical procedures, obstetrical damage, chronic constipation, etc.). Perineal electrophysiologic investigations are thus of limited use for diagnosing pudendal nerve entrapment syndrome given their low sensitivity and specificity and, in addition, they do not provide direct information on pain mechanisms. Pudendal canal syndrome related to nerve entrapment is mainly suspected based on specific clinical features. Perineal electrophysiologic testing provides additional, if not definitive, clues for the diagnosis of pudendal canal syndrome and the localization of the site of compression. In fact, the main value of electrophysiologic testing is to objectively assess pudendal motor innervation when surgical decompression is considered. Perineal electrophysiologic testing may, however, predict the outcome of surgery (Lefaucheur et al. 2007).

10 Conclusion

Due to the limited usefulness of clinical examinations and anorectal manometric and imaging studies, clinical neurophysiologic methods continue to play an important role in determining whether anorectal disorders have a neurogenic etiology. A number of neurophysiologic methods have been used to assess patients with neurogenic anorectal disorders. Of these, EMG investigations of the external anal sphincter muscle are particularly useful in patients with focal lesions of the peripheral sacral nervous system or with atypical Parkinsonism. However, our ability to evaluate the sacral parasympathetic system and peripheral thin-diameter sensory afferent fibers remains limited. Since this system plays a pivotal role in anorectal function, further research aimed at developing clinically useful tests is needed.

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Integration of Diagnostics in Proctology: Assessment, Choice of Treatment, and Evaluation of Results

21

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Contents

Abstract

Proctologist has to face both structural and functional alterations of the complex unit of anorectum. In order to obtain a correct diagnosis and a careful patient's selection to treatment, the knowledge of the diagnostic tools is mandatory, because, to the date, several tests can contribute to an accurate diagnosis, but none of them is sufficient when used alone.

Four are the main symptoms which bring the patient to proctologist: bleeding, fecal incontinence, constipation, and pain. After a comprehensive history collection and a careful physical examination, those have the aim to place the clinical suspicion, there are many diagnostic tools in the quiver: endoscopic tests (e.g., anoscopy, colonoscopy), morphological tests (imaging studies both local (e.g., endoanal ultrasound), and traditional (e.g., CT scan, MRI)), functional tests (e.g., anorectal manometry, balloon expulsion test), morpho-functional tests (e.g., barium defecography), and neurophysiologic tests. However, not all these investigations are mandatory in every patient and for every symptom.

The integration of the diagnostic tools is crucial in order to assess the disease/dysfunction and its pathophysiology and address the patient to the best treatment, and, then, avoid wasting time and public money.

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1 Introduction

> Olim humani artus, cum ventrem otiosum cernerent, ab eo discordarunt, conspiraruntque ne manus ad os cibum ferrent, nec os acciperet datum, nec dentes conficerent. At dum ventrem domare volunt, ipsi quoque defecerunt, totumque corpus ad extremam tabem venit: inde apparuit ventris haud segne ministerium esse, eumque acceptos cibos per omnia membra disserere, et cum eo in gratiam redierunt. Sic senatus et populus quasi unum corpus discordia pereunt concordia valent. (cit. Tito Livio)

See Image 21.1.

In 494 B.C.E. the roman consul Menenio Agrippa, during the plebes revolt against the patricians, explained the roman social order with a metaphor about the human body, in which the survival depends on cooperation of every single part.

This metaphor is useful also to understand the importance of coordination of diagnostic tools in proctology, where if the identify of a symptom is the first step, the knowledge of the variety of diagnostic tools is mandatory to identify the pathology and its mechanisms to cure the patient, avoiding waste of time and public money. Whereas none of currently available test has sufficient positive predictive value when used alone, in term of positive and negative false results.

There are four main symptoms which bring patients to proctologist: bleeding, fecal incontinence, constipation, and pain. Clinical evaluation, along with the planning of a diagnostic strategy is mandatory to arrive to a correct diagnosis.

The first step is common to all of the previous symptoms: a detailed history collection. "What? How Much (severity)? How long (duration)? How often (timing)?" these questions are the pillars of the initial visit or contact with the patient. Coexisting problems, previous surgery and injury, a list of drugs, should be investigated too.

A detailed physical exam is essential for establishing an accurate diagnosis and for direct further investigation to confirm the diagnostic hypothesis. Digital rectal examination is an essential tool in proctologic patients. According to paper of Tantiphlachiva in the 2010, this examination is performed in three steps: (1) inspection

Image 21.1 Barloccini, 1849

of the anus and surrounding tissue, (2) testing of perineal sensation and the anocutaneous reflex, and (3) digital palpation and maneuvers to assess anorectal function (squeeze, pushing, and bearing-down maneuvers).

At this point, the clinical and physical findings detected guide and differentiate the next diagnostic steps.

2 Bleeding

Lower gastrointestinal bleeding is defined as hemorrhage originating distal to the ligament of Treitz (Davila et al. 2005). Rectal bleeding seems to be the sixth most common cause of presentation to emergency department, yet rarely requires blood transfusion or radiological/surgical intervention. In 80 % of patients it is self-limiting and resolves spontaneously (Lee et al. 2009). In case of acute bleeding the vital part is identifying risk in patients, to allow care to be appropriately directed. Naturally, hemodynamically unstable patients or in presence of fragile, medical comorbidities, or concurrent sepsis, the hospital admission is required. Three criteria are been identified to stratify the risk: (1) hemoglobin level, (2) systolic blood pressure, and (3) antiplatelet/anticoagulant therapy, to be significant factors in predicting the need for a blood transfusion (Patel et al. 2014). For the identification of the “high risk” patient Kollef et al. added elevated prothrombin time, erratic mental status, and unstable comorbid disease (1997). The patients identified as “low risk” could be booked for outpatient flexible endoscopy within 6 weeks, unless they have had a recent colonic investigation (Vanhegan et al. 2011; Patel et al. 2014).

Although rare, massive bleeding typically is thought to require more than 3–5 unit of blood transfused in 24 h. Mortality, ranging about 0.6 %, is lower than that from upper gastrointestinal bleeding, which is about 2 % (Kollef et al. 1997).

Massive lower gastrointestinal bleeding can occur at any age, even though there are specific diseases afflicting different age group, showing a familiarity which can help in planning diagnostic workup (Raphaeli and Menon 2012).

Table 21.1 Common causes of hematochezia (Modified from Raphaeli and Menon 2012)

Age group	Source of lower gastrointestinal bleeding
Adolescents and young adults	Meckel’s diverticulum IBD Polyps (juvenile polyps, hamartomas)
Adults to 60 years of age	Diverticula disease IBD CRC
Adults older than 60 years	Arteriovenus malformations Diverticula disease CRC

Meckel’s diverticulum, inflammatory bowel disease (IBD), and juvenile polyps are the most common causes of lower gastrointestinal hemorrhage in young people.

IBD can afflict adult people too, according to its bimodal distribution, with a second peak about age of 60. Bleeding is a typical initial symptom, while massive hemorrhage is present in about 1 % of cases (Robert et al. 1991).

Common cause of intestinal bleeding in adults is the diverticular disease, more frequently (about 50 %) arising from right colon. Colorectal cancer (CRC), the most frequent intestinal cancer and the second cause of mortality for cancer, can arise with bleeding, typically slow and insidious, till chronic anemia. Advanced age, personal or familiar history of polyps, personal history of IBD, or gynecological cancer increase the risk of presenting CRC (Table 21.1).

Less frequent causes of lower gastrointestinal hemorrhage, but more frequent causes of bleeding are the hemorrhoids, colitis (with ischemic, diversion, radiation, infectious nature), solitary rectal ulcer, stercoral ulcer, post-polypectomy bleeding, and small bowel tumors.

In presence of massive bleeding, after resuscitation maneuvers (i.e., crystalloid and blood product) and obtaining stabilization of the patient and information about his history, the goal is the identification of the source of bleeding. The literature reports that the first steps consist of sampling the gastro-duodenal contents, through carefully position of nasogastric tube and performing an esophago-gastro-duodenoscopy to investigate and exclude a proximal source (Jensen and Machicado 1988; Laine and Shan 2010). The second step consists of investigating the "distal potential source" of bleeding: anus, rectum, and till the entire colon. The digital rectal examination, followed by anoscopy, is a fast tool to check for palpable causes of bleeding. The digital rectal examination alone can reveal common cause of bleeding but rare cause of massive bleeding, such as hemorrhoids, fissure, external fistulous opening, perianal dermatitis, traumatic lesions, rectal prolapsed, condyloma, and mass. Anoscopy allows a direct vision of entire anal canal and the most distal part of rectum also, in order to obtain rapid and direct information about anorectal mucosa and its characteristic and rectal contents. The presence of blood and stools could obstruct and limit the inspection of the lumen, so the evaluation can be performed after an enema or a rectal washing. At this point the patient can be submitted to urgent rectosigmoidoscopy, or, if the bleeding has stopped and the patient can sustain a mechanical bowel preparation, a pancolonoscopy. If bleeding is not massive, endoscopy can be booked outpatient in patients who belong to risk group (i.e., older than 50 years, older than 40 years with familiar history of CRC or polyps, personal history of IBD), unless they have had a recent colonic investigation.

Colonoscopy is one of the mainstays of both diagnostic and therapeutic management in case of gastrointestinal bleeding, since the 1970s, though actuating this examination in presence of unprepared bowel or massive bleeding is difficult. Colonoscopy can survey a large spectrum of colorectal abnormalities which could be responsible of rectal bleeding, including benign or malignant diseases (Image 21.2).

If colonoscopy is not useful (e.g., unprepped bowel, negative examine, massive hemorrhage) other diagnostic tools to localize lower GI hemorrhage are:

- *Radionuclide scintigraphy*: a safe procedure with low morbidity and an accuracy ranging about 41–94 %. The slow washout of the tracer (Technetium-labeled red blood cell) is useful to better localize an intermittent bleeding source,

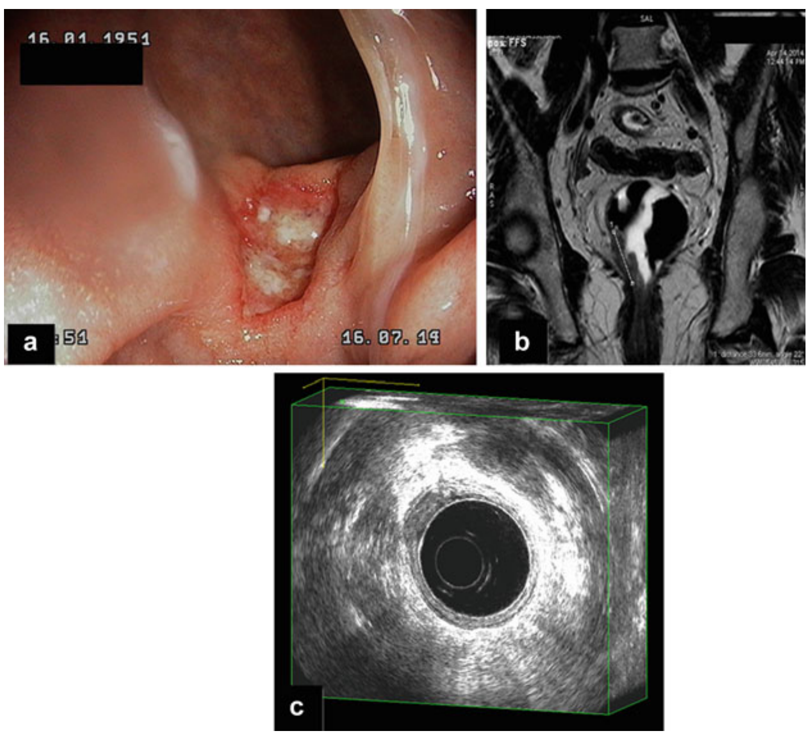

Image 21.2 Colonoscopy (**a**), RMN (**b**), and 3-D transrectal ultrasound (**c**) images of bleeding rectal cancer in 70 years old woman

due to the possibility to rescan the patient multiple time within 12–24 h (Howarth 2006).

- *Diagnostic angiography*: diagnostic mesenteric angiography is an invasive test, performable only in selected patients, which can also have therapeutic purposes (including selective embolization or vasopressin infusion) under provocative test to help the localization of intermittent bleeding.
- *Computed tomography angiography*: a quick and effective radiological examination with an intravenous contrast and a multidetector CT scanner, using dedicated angiographic protocols, allowing a sensitivity of 91–92 % in case of active bleeding (Table 21.2).

Table 21.2 Etiologies of anorectal bleeding (Modified from Daram et al. 2012)

Hemorrhoids
Anal fissure
Fistula
Post-polypectomy bleeding
Diverticula
Trauma
Fecal impaction
Ulceration Ischemia Infections (Cytomegalovirus) Stercoral ulcer Solitary rectal ulcer syndrome Diversion colitis
Infective colitis
Nonspecific proctitis
Inflammatory bowel disease (ulcerative colitis and Crohn's disease)
Uremic colitis
Polyps
Chronic radiation proctopathy
Rectal vascular lesion Rectal varices Angiectasia Angiodysplasia Hemangioma
Neoplasm Villous adenoma Rectal adenocarcinoma Anal squamous cell carcinoma Anal melanoma Local invasion from other pelvic neoplasm
Inflammatory polyps
Endometriosis

- *Evaluation of small bowel*
  - Wireless capsule endoscopy
  - Double-balloon endoscopy

In several diseases colonoscopy can represent the last diagnostic tool (e.g., direct disease visualization, indirect disease stigmata, obtaining a biopsy), and it can become an useful therapeutic tool, not only in case of polyps which can be removed by polypectomy. Endoscopic treatment options include thermal coagulation with argon plasma coagulation (APC), bipolar or heater probes, cryotherapy, or neodymium-doped yttrium aluminum garnet laser therapy, and endoscopic topical application of formalin (e.g., in case of chronic radiation proctopathy). Another important role played by colonoscopy is in the follow up period, to verify the pertinence of a therapy, due to the possibility of serial endoscopic monitoring (Image 21.3).

Either benign or malign proctologic disease can present a nontypical endoscopic scene as in case of a rectal ulcer.

A rectal ulcer, as reveled at colonoscopy, needs a biopsy to clarify its nature, and obtain a definitive diagnosis, because it presents a frequent misdiagnosis due to its nonpathognomic symptoms (including bleeding, mucus discharge, and anorectal pain) and varied endoscopic appearance. Solitary rectal ulcer syndrome is a common end-pathophysiologic process of focal rectal mucosa ischemia and ulcer formation. Many factors may play a role in its etiology: rectal intussusception, pelvic floor dyssynergia, and local rectal trauma. Contrary to the name, less than one third of the patients (more frequently women in the third-fourth decade) present a single lesion; many ulcers are observed in about 40 % of patients, hyperemic mucosa and polypoid mucosal change in less than one fourth of patients. The anterior rectal wall is the most frequent part involved. The absence of malignancy or suspect of IBD at biopsy, in young adults with no history of radiotherapy for pelvic tumor or suspect of ischemic colitis, is suggestive of solitary rectal ulcer. Either defecography or dynamic magnetic resonance is useful in finding suggestive anatomo-functional alteration frequently associated to this syndrome as the presence of rectal intussusceptions or pelvic

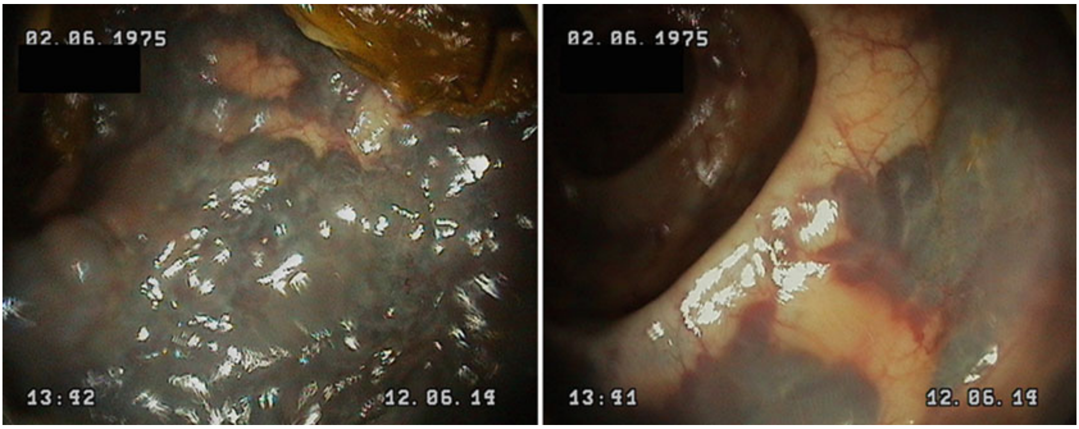

Image 21.3 Multiple cyanotic telangiectasia in a 39-year-old young man

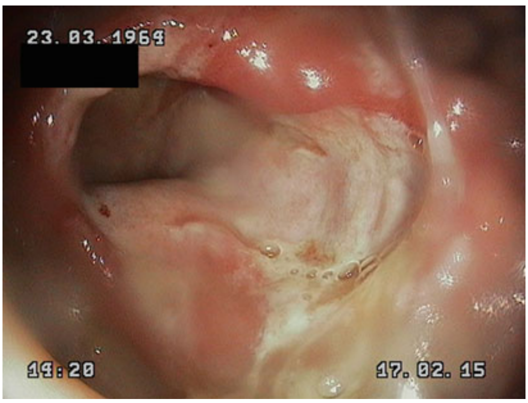

Image 21.4 Solitary rectal ulcer in a 50-year-old woman

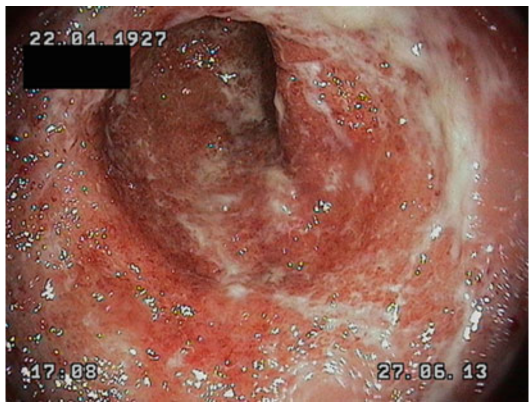

Image 21.5 Bleeding idiopathic proctitis in an 86-year-old woman

dyssynergia (Sharara et al. 2005). Moreover pelvic dyssynergia may be investigated with anorectal manometry (ARM) and balloon expulsion test (BET) (Image 21.4).

In case an ischemic proctitis is the responsible of rectal ulcers (e.g., when the patient is submitted to radiation therapy, aortic aneurysm repair, or aorto-iliac intervention with theoretical insult to blood supply to the rectum), the endoscopic findings are represented by mucosal ischemia with associated ulceration, and, sometimes, when the ischemia is severe, confluent ulcers, pseudomembranes, and poor bleeding when submitted to biopsies. Histopathologic confirmation is mandatory. The evidence of pneumatosis and extraluminal air-findings at CT scan suggest transmural necrosis and represents an indication to surgical treatment (Image 21.5).

The presence of a rectal mass, polyp, or ulcer with malignancy at biopsy starts the diagnostic falls for rectal cancer staging, according to NCCN guidelines (Version 2.2015). In case of noncomplete colonoscopy, it is important to "extend" the endoscopic examination to the whole colon. Blood tumor marker test should be performed to report the level of CEA (carcinoembryonic antigen) and Ca 19.9, to help monitoring the response to treatment or providing an early warning of recurrence.

Additional information regarding the extent of disease and distant metastases can be determined through: endorectal ultrasound and pelvic MRI to assess the depth of tumor penetration through the rectal wall and spread to local lymph nodes (N+); contrast CT scan of chest, abdomen, and pelvis for the preoperative staging of rectal cancer; PET/CT to evaluate an equivocal finding on a contrast CT scan.

Evidence supports an integrated therapeutic approach to rectal cancer. Local excision is appropriated for early stage without N+. Neoadjuvant chemoradiotherapy is indicated for patients with stage II-III rectal cancer and surgical strategy depending on the extent of disease. However, total mesorectal excision (TME) is recommended. Adjuvant chemotherapy is only suggest following neoadjuvant chemoradiotherapy and surgery in case of suboptimal treatment (e.g., positive circumferential resection margins, perineural invasion, and /or lymphovascular invasion) (Images 21.6, 21.7, and 21.8).

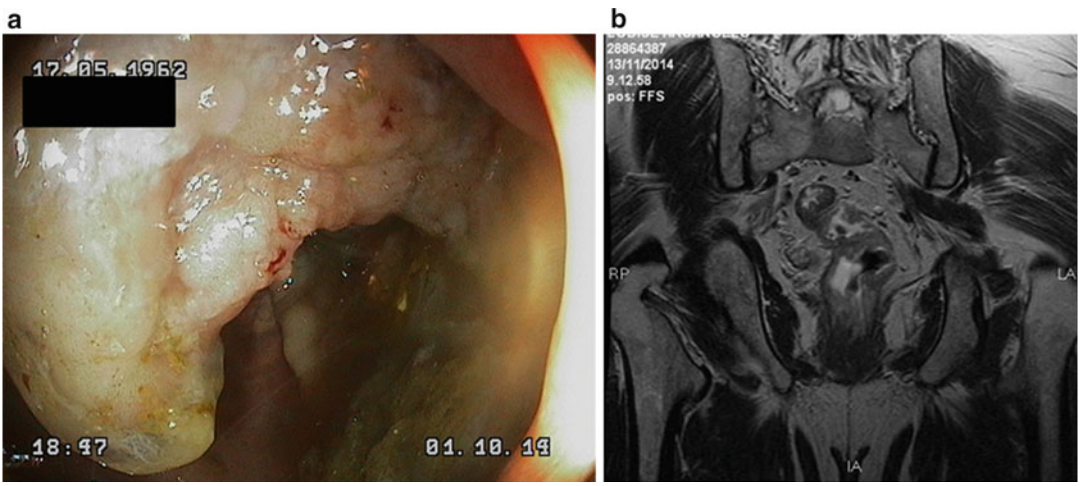

Image 21.6 Colonoscopy (**a**) and RMN (**b**) images of rectal cancer in a 52 years old man

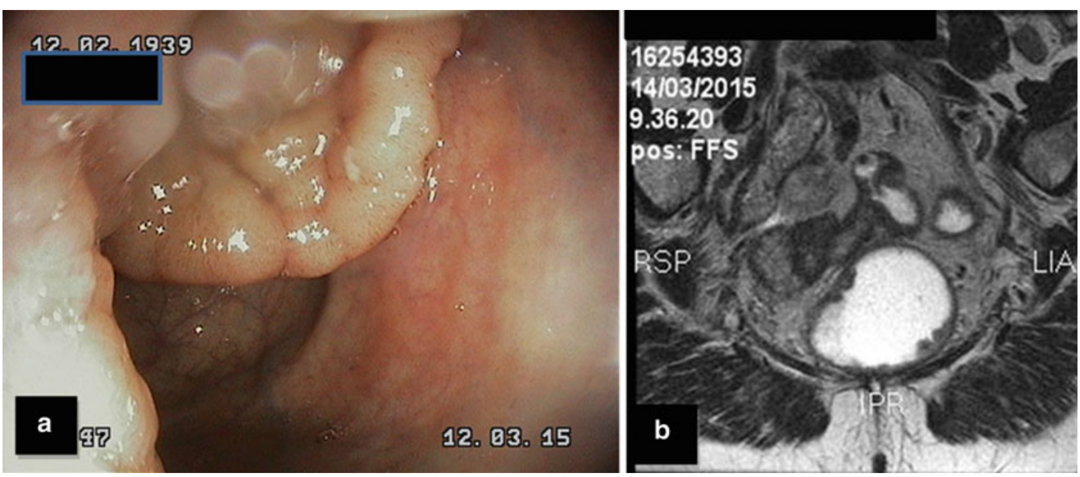

Image 21.7 Colonoscopy (**a**) and RMN (**b**) images of rectal adenoma in a 76-year-old woman

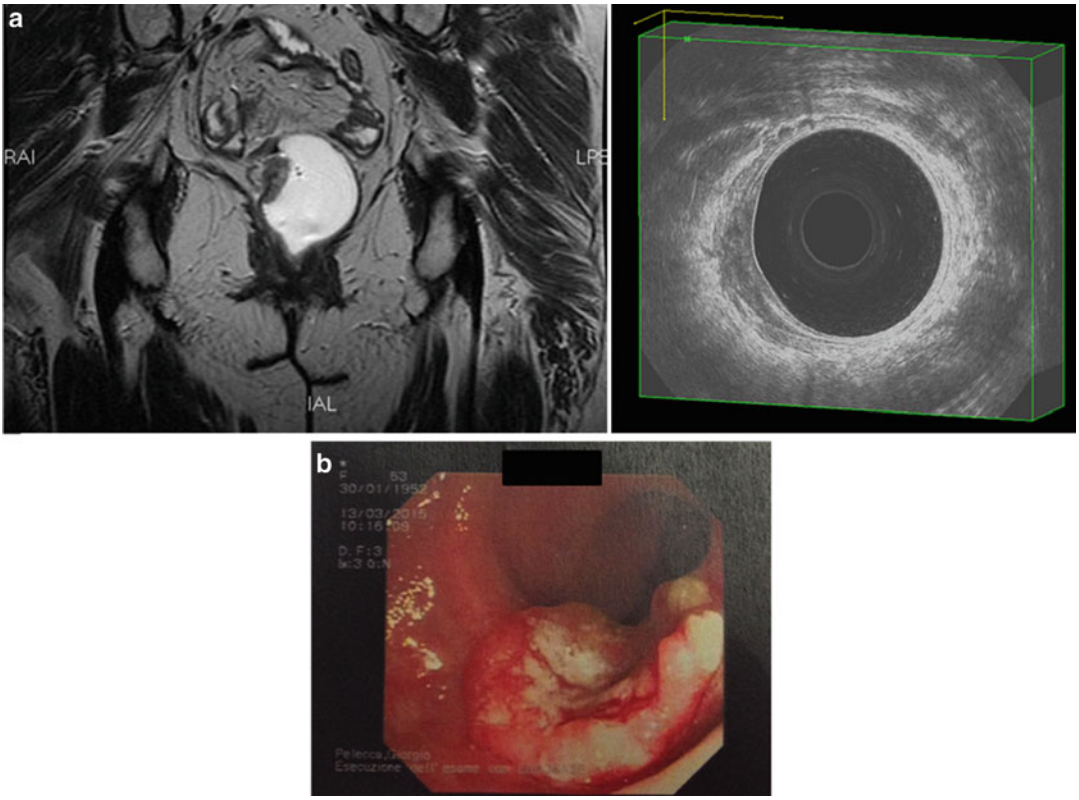

Image 21.8 RMN (**a**), TRUS (**b**) and colonoscopy images of rectal cancer in a 63-year-old woman

3 Fecal Incontinence

"Normal defecation is a process of integrated somato-visceral responses, which involve coordinated colo-recto-anal functions" (Chan et al. 2005).

Anal continence is a complex mechanism which can fail at many levels from central nervous system to the anal canal, due to deficit of nervous, muscular, supportive structures, or rectal ampulla, and it could carry to fecal incontinence when compensatory mechanism fail too.

Fecal incontinence (FI) is a distressing condition which affect the life of patients and their relatives, characterized by an involuntary loss of feces and the inability of delay the defecation. The prevalence of FI is similar between men and women at 7.7 % and 8.9 %, respectively, and increases with age, reaching 15.3 % in 70 years or older people (Whitehead et al. 2009), and up to 50 % of nursing home residents (Nelson et al. 1998). A recent population-based study found FI in 17 % of adults aged >65 years, where independent risk factors (controlling for age, comorbidity count, and body mass index) were: female gender, white race, depression, chronic diarrhea, and urinary incontinence (UI); while UI was the only risk factor in men (Marckland et al. 2010). Obstetrical trauma, with internal anal sphincter injury and reduced perineal descent, seems able to independently predict the development of FI in women (Bharucha et al. 2012). Other risk factors described have been: neurological disorders, cholecystectomy, current smokers, congenital anorectal malformations, trauma, iatrogenic anorectal injury, and multiple illnesses.

Causes of FI are: anal sphincter lesions or weakness, neurologic impairment related to damage of pudendal nerve, neuropathy, decreased rectal capacity, bowel disturbance, reduced mental awareness, and physical inability to reach toilet facilities (Lam et al. 2012).

At preliminary visit, FI should be recognized to be an affection of primary importance. Because only less than one third of patients with FI discuss their symptoms with a doctor, it is important to specifically investigate patients about the presence of FI. Thereafter, determining symptoms severity, investigating bowel habits through the use of Bristol Stool Form Scale and bowel diary, and the frequency, amount, type of leakage, and the presence of urgency, represents the second step. Patients who suffer for urge FI usually experience loss of stools despite their efforts to control them; they usually present reduced squeeze pressure and/or squeeze endurance, and/or reduced rectal capacity, and/or rectal hypersensitivity. On the other hand, patients who suffer for passive FI experience the loss of stools without sensation of their passage; they can show lower resting pressure.

During collection of clinical history, it is also important to assess if FI is secondary to diarrhea. In this case colonoscopy, stool test, and breath test may be useful.

Physical examination has three purposes: to identify those diseases causing FI as secondary effect; to identify rectal masses; and to characterize anorectal functions. During inspection the presence of fecal residuals, skin excoriations, dermatitis, scars, prolapsing hemorrhoids, patulous anus, loss of perineal body, and muscular deficit may be noted. Asking patients to simulate an attempt of defecation may reveal excessive perineal descent or rectal prolapse. Perineal pinprick sensation and anal wink reflex can assess the integrity of sacral lower motoneural reflex arc. An impaired or absent anocutaneous reflex suggests neuronal injury.

Palpation has to test anorectal function by evaluation of resting and squeezing tone, puborectalis motion, the presence of muscular deficit, and other anatomical alteration (including internal rectal prolapse, rectocele, and enterocele). In FI patients, frequent manometric findings are a reduced anal resting tone and/or weak squeeze response; they would suggest deficit of either the internal anal sphincter or external anal sphincter, respectively.

At this point, the use of diagnostic test has the aim of better characterizing anorectal function, by a correct assessment of pathophysiology in order to obtain a rationale choice of management. Specific and complementary tests which can define the underlying mechanisms include anorectal manometry (ARM), rectal sensation test, endoanal ultrasound (EAUS), magnetic resonance imaging (MRI), and neurophysiological tests. However, not all these investigations are mandatory in every patient.

ARM offers an objective measure of the anal canal pressure, quantifying the internal and external sphincter action, and determines rectal sensation, recto-anal reflex and rectal capacity. It can demonstrate a lower resting and squeezing pressure in FI patients with both sphincter defect and sphincter atrophy (Reddymasu et al. 2009). Either altered rectal sensation or compliance is other common feature in FI patients, who may exhibit rectal hyposensitivity or hypersensitivity. Rectal compliance is reduced in patients with colitis, low spinal cord injuries, and diabetes, but increased in patients with high spinal cord injuries. Cough reflex response (an external sphincter contraction in response to sudden increase of intra-abdominal pressure) is impaired in neurological patients (i.e., cauda equina lesions) (Rao 2004). Moreover, ARM, along with rectal balloon expulsion test (BET), can give additional information in suspicion of defecatory disorders which could contribute to FI. However, different studies showed a poor correlation between ARM findings and FI scores, advising against its routinely use (Lam et al. 2012); on the other hand, recent guidelines indicate, with strong recommendation, that the ARM evaluation, as first line diagnostic test, should be performed in FI patients who fail to respond to conservative measures (Wald et al. 2014; Image 21.9).

Anal imaging, provided by either EAUS or MRI, should be performed in FI patients with (or without) altered ARM features, where surgical treatment is a possible option. EAUS provides an assessment of sphincter structural integrity and echogenicity, evaluation of tissue (normal, scar,

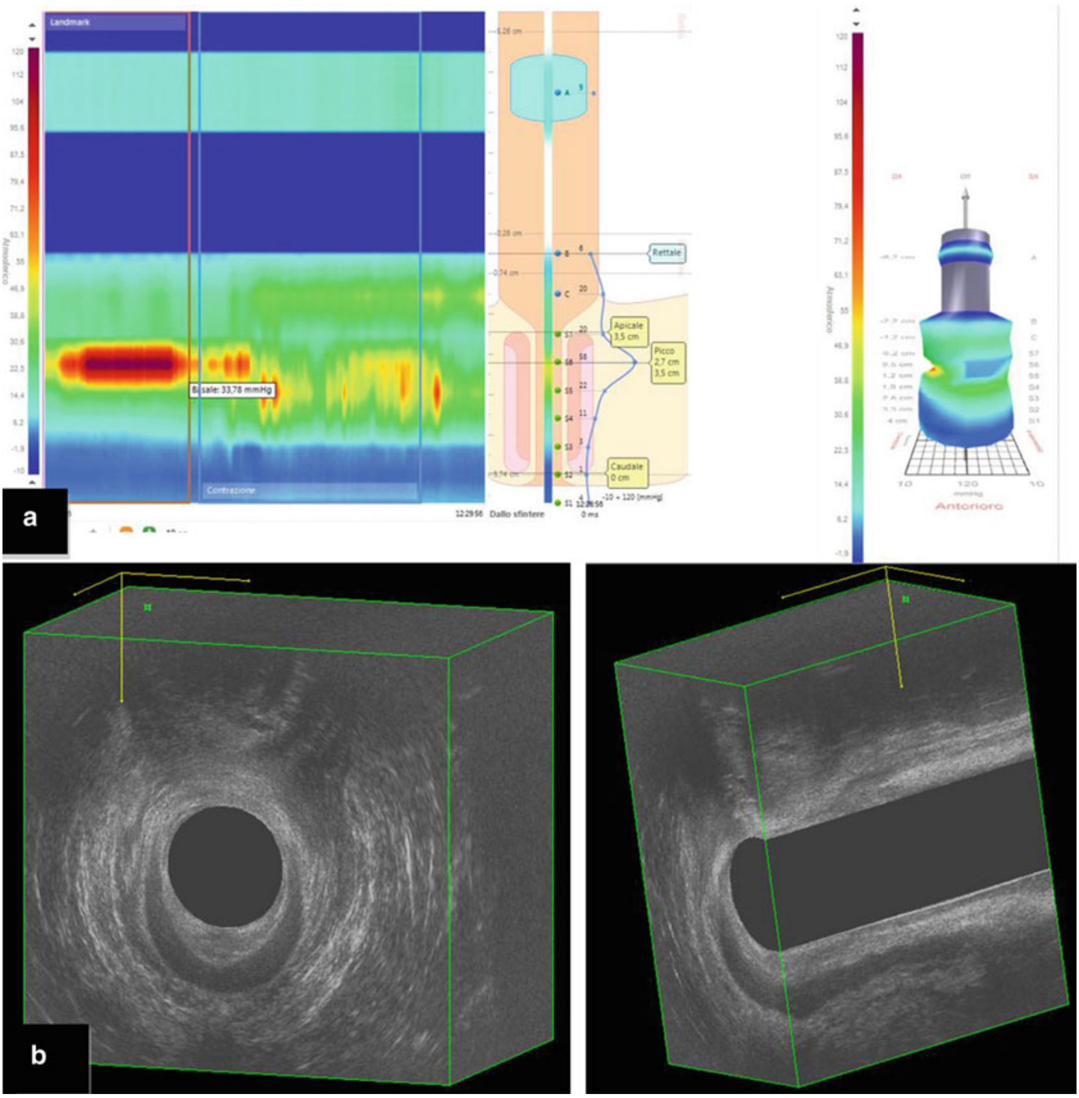

Image 21.9 ARM traces of low resting and squeezing pressure (**a**) and EAUS (**b**) images of sphincters lesion in a 35-year-old FI woman

atrophy), measurement of sphincter thickness or lesions extension (in terms of thickness, angle, length, area, and volume), presence of other local pathology and correlations with other diagnostic tools. It can play a role during diagnosis, treatment (echo-guided treatments), and follow up. It can distinguish internal from external anal sphincter injury but has a low specificity in demonstrating the etiology of FI. MRI provides superior imaging of external sphincter with better discriminating between tears and a scar and recognition of atrophy. Internal sphincter defect are probably associated to a worst anorectal lesion than external sphincter lesion alone. Moreover even asymptomatic women can have an occult postpartum sphincter lesion, with a prevalence of ~10 % at 3D-EAUS or MRI (Wald et al. 2014).

Further tests, not widely available (electromyography (EMG) of the anal sphincter, motor evoked potentials, dynamic MRI, or barium proctography), may be considered in presence of patients with refractory symptoms.

EMG of anal sphincter detects which indicate neuropathy (e.g., denervation-reinnervation potentials). In FI patients with clinically suspected neurogenic sphincter weakness, pudendal nerve injury (e.g., forceps assisted delivery) or cauda equine syndrome, fibrillation potentials, and high-frequency spontaneous discharge are suggestive of denervation.

History alone can detect an underlying cause only in a minority of FI patients (11 %), whereas specific tests reveal abnormal features in about 55 % of the patients (Wexner and Jorge 1994); but test alone has little or no predictive value in determining further FI and in the efficacy of treatment for FI.

In conclusion, specific morphologic and physiologic tests seem to be very useful both for providing a diagnosis and for assessing objective improvement in follow-up periods during and after therapeutic intervention.

4 Constipation

Constipation is a common, subjective, polysymptomatic, multifactorial disorder which affect up more than 27 % of the population, more prevalent in women and elderly people (Higgins and Johanson 2004). Frequent symptoms of severe constipation are incomplete, prolonged, difficult, rare and/or painful evacuation, abdominal pain, and bloating.

Constipation can be either primary or secondary. Primary constipation, defined according to Rome III criteria (Longstreth et al. 2006), is due to "intestinal" causes which altered colonic and anorectal function, whereas secondary constipation is related to "extraintestinal" causes, including endocrine, metabolic, neurologic, rheumatologic, psychological, medications taking, or dietary factors (e.g., poor water and fiber intake). Primary constipation recognizes three main overlapping conditions: *slow transit constipation* (STC), characterized by impaired propulsion of stools; *defecatory disorders* (DD), characterized by difficulty in evacuating stools; *constipation predominant irritable bowel syndrome* (IBS-C), characterized by association of constipation and abdominal discomfort or pain (Rao 2010).

Because constipation is a heterogeneous disorder, arising from the integration of multiple physiologic components, there is no test which is able to mimic the real-life stool progression and expulsion. Therefore, accurate assessment of patient's history and physical examination should be integrated with more than one test to define and characterize constipation.

A complete and detailed history should always been taken in patients affected by chronic constipation, with the aim of characterizing constipation (particular emphasis on stool habit and consistency) and identifying elements which may be related to a secondary constipation. Alarm features can be identified (such as new onset of constipation after age of 50, weight loss, anemia, bloody stools, family history of colorectal cancer, or inflammatory bowel disease) in order to seek and exclude a neoplasm.

There are no specific criteria based on neither the patient's history, nor pathognomonic symptoms, which can distinguish between normal subjects and those affected by subtypes of chronic constipation. However, the occurrence of two or more symptoms during at least 25 % of bowel movements distinguishes patients with chronic constipation from normal subjects.

Physical examination includes an accurate examination of the abdomen, perineum, and anorectum. The abdominal palpation may evidence the presence of stools, mainly in the left iliac fossa. The inspection of perineum can detect external signs of an anal disease, pelvic organ prolapse, or descending perineum syndrome. A digital rectal examination should detect signs of organic disease or obstructed defecation. The digital rectal examination may provide information about rectal content (stools presence and consistency), mass, or stenosis, and about the presence of some morphological abnormalities (rectocele, internal rectal intussusception, and enterocele). Moreover, digital rectal examination may give information of anorectal function, in terms of sphincter tone (internal anal sphincter, external anal sphincter, puborectalis muscle) at rest, and during squeezing and straining. Meticulous evaluation of anal sphincter tone and pelvic floor motion by digital rectal examination is reasonably accurate in the majority of patients in assessing anal resting tone, squeeze function, and in identifying dyssynergic defecation pattern (Tantiphlachiva et al. 2010). However, it is unable to give an accurate quantification of pressures. Some people, with normal pelvic floor function, are unable to simulate defecation attempts during digital rectal examination; therefore a normal digital rectal examination is more useful than a dyssynergic one in patients with chronic constipation (Rao 2010).

When indicated, endoscopy, rectosigmoidoscopy, and barium enema can integrate the physical examination, in order to identify or, on other hand, exclude anorectal diseases (neoplasm, stenosis, megarectum, inflammatory bowel disease, or solitary rectal ulcer). Melanosis coli is a frequent endoscopic finding in patients with history of chronic laxative abuse.

Once an organic alteration has been excluded, a neuromuscular disorder affecting the colon-rectum should be hypothesized.

Anorectal manometry (ARM), rectal balloon expulsion test (BET), followed by barium defecography or magnetic resonance proctogram or colonic transit time (CTT), if necessary, are recommended in constipated patients unresponsive to diet and lifestyle modification, to laxative therapy, or in presence of defecatory disorders, referred to difficulty in evacuating stool from rectum associated to chronic constipation.

ARM provides indirect information about anorectal function, and can identify defects involved in the pathophysiology of constipation. Common manometric features in constipated patients are: increased resting pressure, rectoanal inhibitory reflex defects, rectal hyposensitivity, and increased rectal compliance. In normal subjects, during attempt of defecation, ARM records an increase in rectal pressure (sign of adequate propulsive forces) and a synchronic decrease in anal pressure (sign of relaxation of puborectalis muscle and anal sphincters). This mechanism fails in patients with dyssynergic defecation, and ARM traces can show low rectal pressure (sign of weak rectal propulsive forces) and/or inadequate relaxation or paradoxical contraction of anal sphincters (sign of increased anal resistance) (Rao 2004). ARM is useful in identifying patients affected by dyssynergic defecation and impaired rectal sensation who could benefit with biofeedback therapy. ARM can be used to guide rehabilitation therapy, in order to develop a pelvic floor rehabilitation program for the patient, and to monitor the results (Image 21.10).

BET is a simple and cheap test, which can identify abnormal defecation. It consists of simulated evacuation in which a rubber balloon is inserted into the rectum. There is no standard technique: the filling volume of the balloon, the positions of the patient, and the expulsion time have differed in various studies. Nevertheless, BET has a high specificity for dyssyrgic defecation, and shows an high concordance (72–95 %) with ARM. Although failure to expel the balloon suggests dyssynergia, a normal test does not exclude the diagnosis. Hence BET, for the diagnosis of dyssynergic defecation, should be integrated with other physiological tests (Image 21.11).

Electrophysiological studies are useful in identifying a failure in relaxation of the puborectalis muscles or of the anal sphincters by recording their electrical activity, and seem to present a good agreement with ARM. Many studies on constipated patients have showed abnormal traces due to puborectalis muscle during straining (i.e., increased or unchanged activity). Electromyography is frequently used in biofeedback training for dyssynergic defecation.

Barium defecography is a dynamic morpho-functional test that, using fluoroscopy, provides information about anatomical and functional changes of the anorectum. Using barium to mimic stools, it shows the anorectum during pelvic floor contraction and before (at rest) during and after defecatory maneuvers. Anorectal angle provides an indirect measurement of puborectalis activity; in dyssynergic defecation this angle may not widen during defecation attempts, sign of failure of puborectalis muscle to relax in order to allow evacuation (Nielson et al. 1993).

In descending perineum syndrome, defecography reveals a distance between anorectal junction and pobococcygeal line >4 cm on straining (Somorowska et al. 1987).

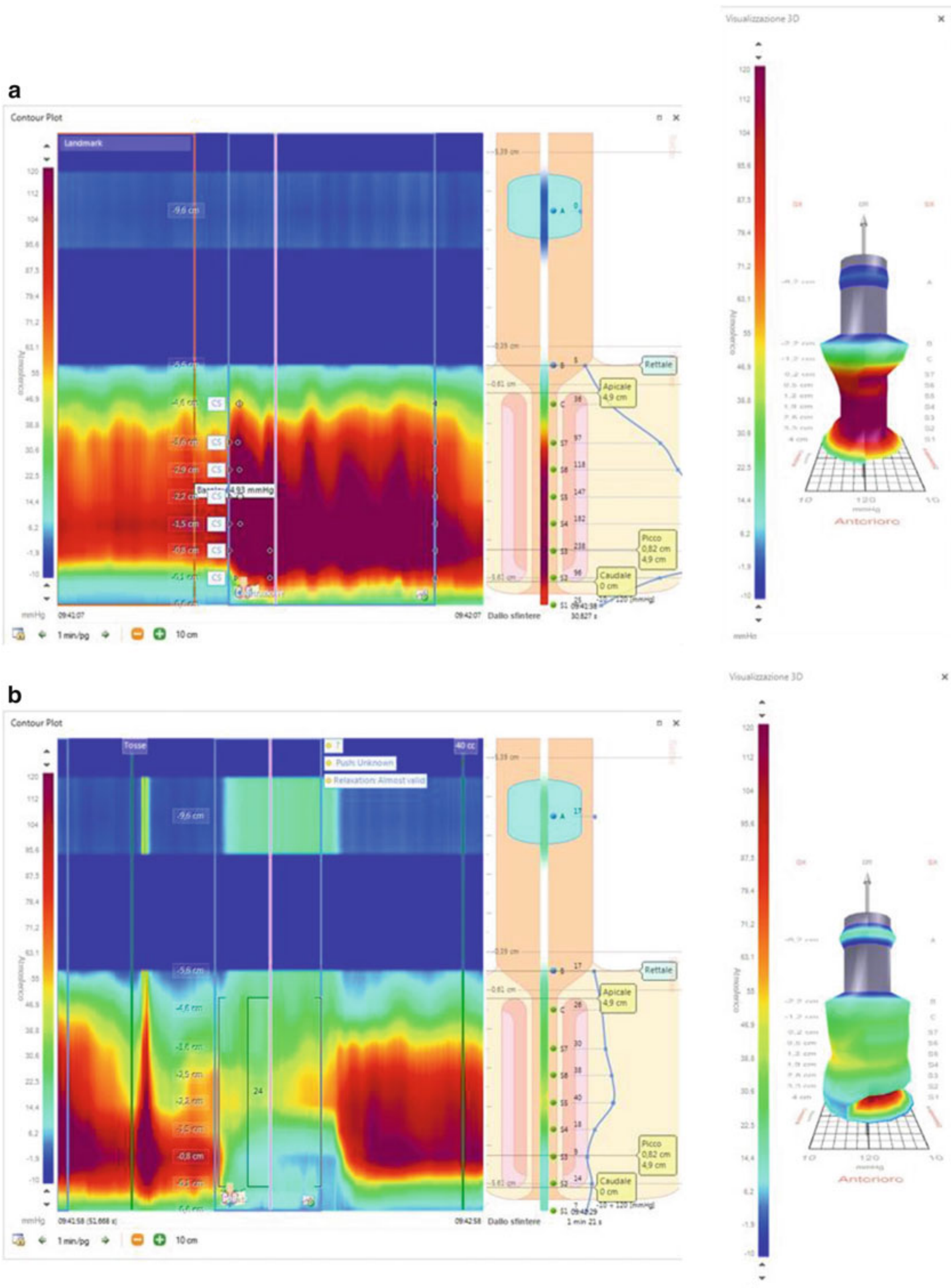

Image 21.10 ARM traces: high resting and squeezing pressure (**a**) and normal pushing pressure (**b**) in a constipated a 38-year-old woman

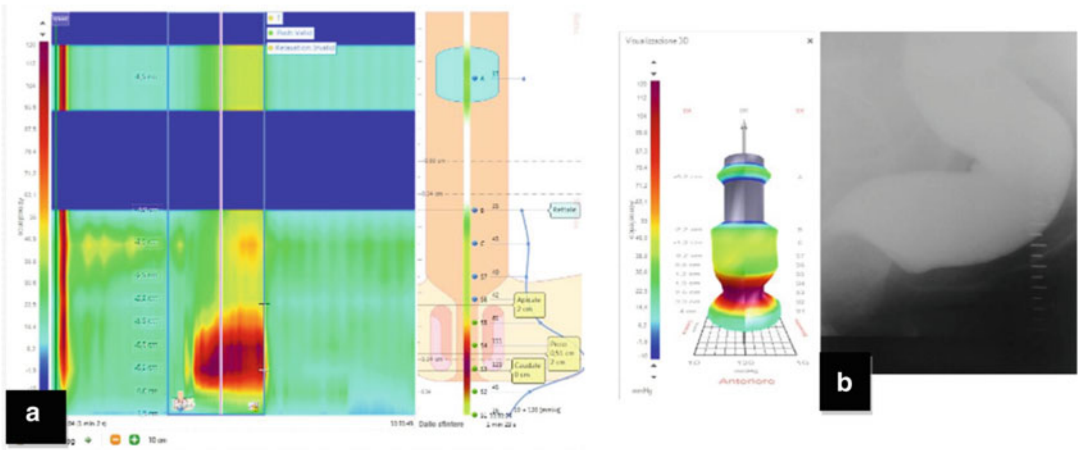

Image 21.11 Dyssynergic ARM (**a**) and defecography (**b**) pattern with a synchronous augmentation of endorectal and endoanal pressures, in a constipated 46-year-old man

Moreover, defecography may characterize structural causes of obstructed defecation including rectal prolapse (mucosal, internal, and/or external), rectocele, perineocele, and enterocele. Consequently, defecography is not a first line test in diagnostic work up of constipation; it is useful in case of equivocal ARM and BET, because it can identify impaired stools evacuation in patients with suggestive symptoms but normal BET and EMG, or in presence of suspected of structural causes of obstructed defecation.

An alternative to barium defecography is magnetic resonance (MR) proctogram. The test is a dynamic evaluation of pelvic floor image during defecation attempts, such as during defecography, with a global evaluation of pelvic floor anatomy and motion, free selection of imaging plane, lack of radiations, and a better resolution of soft tissue. MR can differentiate mucosal from full-thickness rectal prolapse (Bharucha et al. 2005). However, significant limitations are represented by high cost, the nonphysiological supine position, lack of availability, and lack of standardization.

Defecography and MR proctogram, providing the evidence of structural abnormalities, can guide the surgical treatment and be helpful in the follow up evaluation (Image 21.12).

In case of suspect slow transit constipation (STC), a whole-gut transit may be useful to demonstrate the speed of stool progression through the colon-rectum. It can be performed using radiopaque markers, scintigraphy, or wireless motility capsule. An initial transit time study can differentiate between patients with total or segmental colonic STC and patients with outlet obstruction, showing the site of accumulation of the radiopaque markers along the large bowel. Unfortunately, lack of standardization in the procedure makes it difficult to compare results among different centers (Image 21.13).

The wireless motility capsule provide a radiation-free, validated, standardized method of assessing both whole-gut and regional transit, with a good specificity and good agreement with radiopaque markers test (Rao et al. 2009).

Even if all the above mentioned diagnostic tests can contribute to an accurate diagnosis, to date none of them has sufficient positive predictive value when used alone.

5 Pain

"Chronic proctalgia" is one of the many definition of a syndrome characterized by recurring episodes of rectal pain, each lasting at least 20 min. This painful condition is defined as a nonmalignant and noninfective pain, constant or recurring over a period of at least 6 months, associated with negative behavioral and social consequences and with

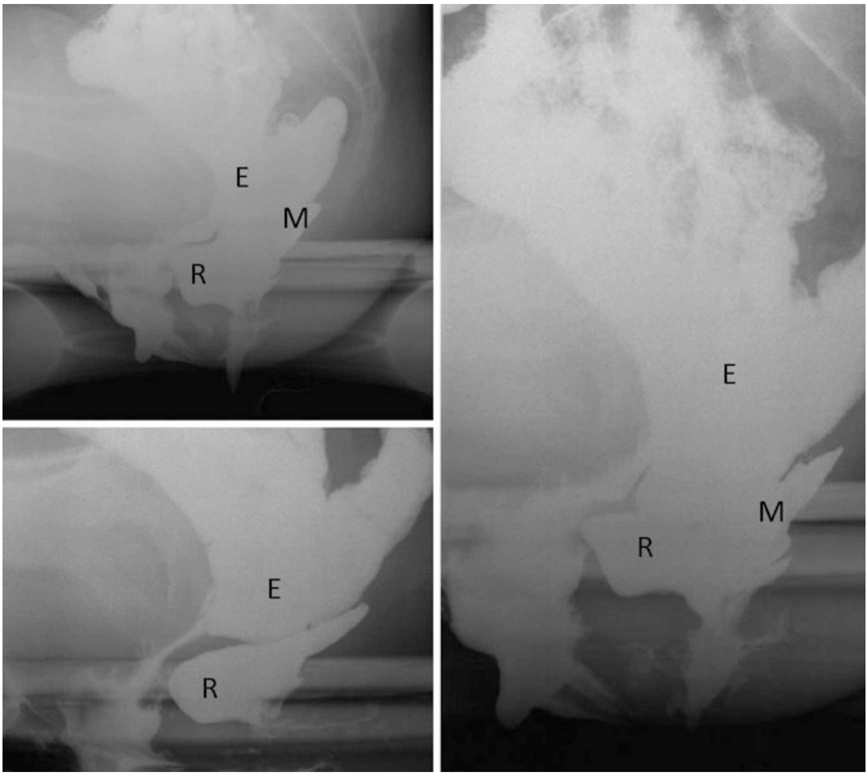

Image 21.12 Barium defecography showing rectocele (*R*), enterocele (*E*), and mucosal rectal prolapse (*M*)

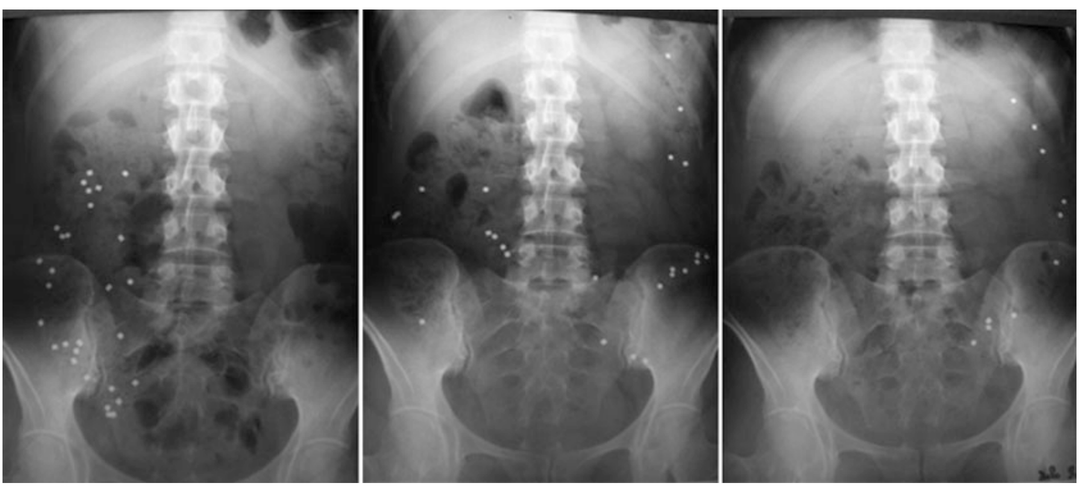

Image 21.13 Colonic transit time with radiopaque markers

bladder, sexual, or bowel dysfunction (Martellucci et al. 2012). Commonly, it is considered that chronic proctalgia is due to a sustained pelvic floor muscles spasm, and some authors suggest that the pathophysiology may overlap to that of dyssynergic defecation (Hart et al. 2012).

Diagnosis is based on a history of constant or recurring episodes of rectal pain, a digital rectal examination showing tenderness or pain to palpation of the puborectalis muscle and exclusion of other causes for rectal pain, by an adequate work-up based. Other pathological condition may mimic this syndrome, such as chronic pelvic pain, chronic prostatitis, and iatrogenic, or post-actinic pain. A suggestive history along with imaging study or endoscopy should be useful to exclude structural causes of rectal pain.

One large study showed that negative balloon expulsion test (BET) and manometric dyssynergic pattern (inability to relax pelvic floor muscles during simulated defecation) correlated with the presence of tenderness to palpation and were predictive of the success of biofeedback treatment in patients with history of chronic proctalgia (Chiarioni et al. 2010). Patients with clinical suspicion of chronic proctalgia may be submitted to BET and ARM in order to obtain a correct selection to treatment.

Other common causes of anorectal pain at physical examination could be the relief of hemorrhoidal thrombosis, anal fissure, or anorectal sepsis. The presence of purulent discharge and/or perianal swelling are suggestive of anorectal fistula or abscess. A comprehensive history and examination (including endoscopy) are fundamental, in order to determine the presence and configuration of the anal fistula (in relation to sphincter muscles) and the exclusion or association of other conditions (such as Crohn's disease). Careful examination of perianal skin and accurate digital rectal examination provide a large amount of information. Moreover, an evaluation under anesthesia could define the anatomy of the fistula more accurately. A simple fistula may be definitively treated at the time of evaluation but, in case of complex fistulas, it represents the first step of management; in fact, further investigations and procedures are usually necessary (Simpson et al. 2012). Endoanal ultrasound (EAUS) and magnetic resonance imaging (MRI) are the most useful imaging techniques to improve the characterization of fistula tract. EAUS is cheaper, provides anatomical detail of the tract and the sphincters, accuracy can be improved by injection of hydrogen peroxide into the fistula tract, and can be a useful intraoperative tool. Unfortunately, it can be considered operator-dependent, though the 3D-EAUS shows higher accuracy (~95 %) and a 98.8 % interobserver agreement. MRI provides excellent soft tissue resolution in multiple planes without recurring to ionizing radiation; moreover, it seems less operator-independent, and shows a superior field of view. Recently, a meta-analysis has confirmed that EAUS and MRI had similar sensitivity for detecting fistula (87 %), but MRI had higher specificity (69 %) (Siddiqui et al. 2012).

On the other hand, MRI is not applicable in patients with metallic implants or suffering from claustrophobia, whilst EAUS can be painful or impossible to perform in the presence of anal stenosis. In these cases, alternative methods to obtain information are represented by thin slice spiral computed tomography and transperineal ultrasound (Image 21.14).

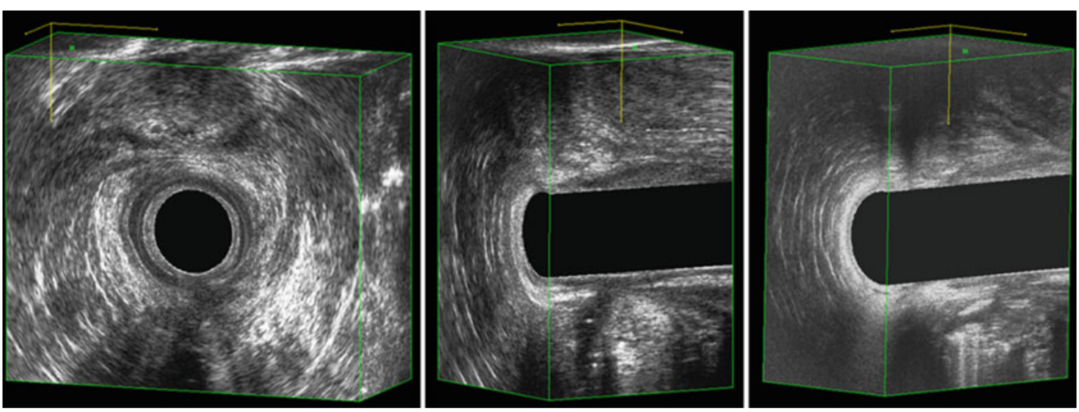

Image 21.14 Posterior transphincteric perianal abscess in 33-year-old women

The presence of an ulcer or a mass at the level of anal margin or anal canal raises the suspect of malignancy, especially in the higher risk population (e.g., human immunodeficiency virus (HIV), anal intercourse, high lifetime number of sexual partners, immune-suppression in transplant recipients, autoimmune disorders). The diagnosis of anal cancer is made on biopsy-proven histology. There are no pathognomonic symptoms; frequently the patients affected present a combination of mass, ulcer, bleeding, pain, itching, discharge. Medical conditions, current medications, and predisposing factors should be investigated. Digital rectal examination should examine anal lesion and perianal involvement, while, in women, a vaginal examination may determine the site and size of the primary tumor, recto-vaginal septum involvement, or the presence of a fistula. The vaginal involvement may require a prophylactic construction of a stoma, in order to avoid an anorecto-vaginal fistula. Proctoscopy or evaluation under anesthesia may facilitate the biopsy. Staging is mandatory. Imaging includes MRI of the pelvis and EAUS to provide information on tumor size, local extent and spread, invasion of adjacent organs, and,

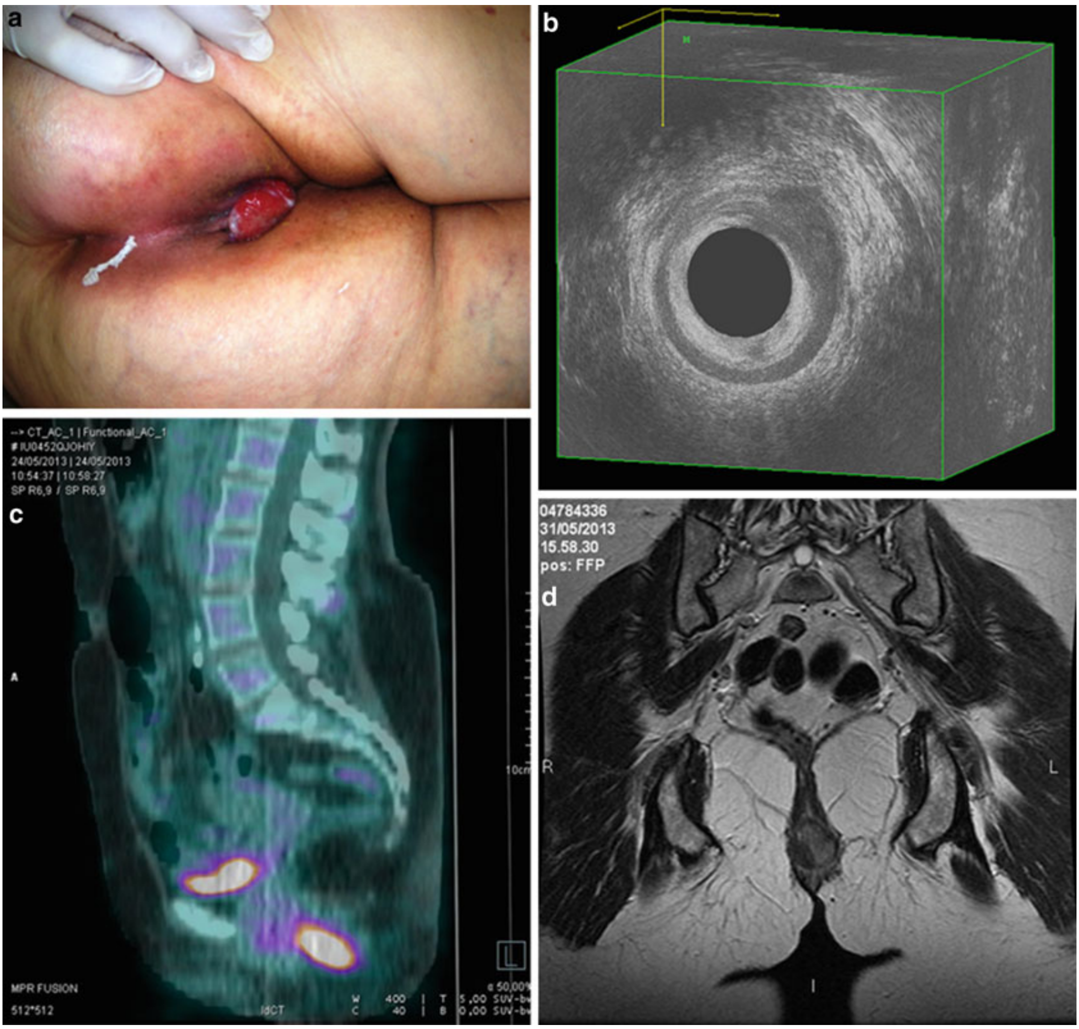

Image 21.15 Physical examination (**a**), EAUS (**b**), PET-TC (**c**), and RMN (**d**) images of anal cancer in a 54-year-old woman

especially with MRI, an accurate nodal involvement. Distant metastases can be assessed with a chest and abdomen CT scan. In the current USA National Comprehensive Cancer Network treatment recommendations, positron emission tomography (PET)/CT with fluorodeoxyglucose, with its high sensitivity in identifying nodal involvement and influencing radiation therapy planning by defining site of metabolically active tumor, has been recommended (Glynne-Jones et al. 2014; Image 21.15).

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Pathologic Evaluation of Colo-Recto-Anal Samples: Procedures and Clinical Significance

22

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Contents

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1 Introduction

The objective of every physician is to provide patients with high quality medical care. Traditionally, medical schools and postgraduate training programs have cultivated a somewhat unilateral approach to treat medical diseases in which each specialty operates in silos. There is mounting evidence that patients who are treated in the context of a multidisciplinary team, regardless of the disease process, fare better outcomes (Kesson et al. 2012; Burton et al. 2006). Diagnosis of surgical and biopsy specimens by pathologists ought to be rendered in a framework of sufficient clinical information, as many disease processes share gross and microscopic features (Robinson 1934). Similarly, it is critical that clinicians understand the processes pertinent to specimen preparation as well as some of the basic knowledge related to pathological diagnosis.

In this review, we will discuss important and practical information regarding the preparation and interpretation of pathological specimens of the colon, rectum, and anus.

2 Types of Tissue Received in the Pathology Laboratory

There are mainly two types of tissue specimens that are handed off to the pathologist for interpretation: (1) biopsies and (2) surgically resected specimens.

2.1 Biopsies

In the era of modern medicine, with the exception of emergencies, all radical surgeries are performed after the pathologist renders a definitive diagnosis on biopsy specimens, regardless of the nature of the disease. Frequently, diagnostic tissue is interpreted by pathologists at an institution different from the one at which the patient will undergo radical surgery. Therefore, it is advisable that the corresponding diagnostic slides are reviewed by the pathologists at the institution at which the definitive treatment will occur. This practice is not only relevant for cases in which malignancy is entertained, but also for conditions such as idiopathic inflammatory disease. The surgical treatment for ulcerative colitis (UC) and Crohn's disease (CD) (inflammatory bowel disease, IBD) frequently differs and therefore correct identification by the pathologist between these two entities is essential. Similarly, if the decision of radical surgery is based on a presumptive diagnosis of dysplasia in the background of IBD, it is imperative that a second opinion is sought since interobserver variability in the diagnosis of IBD is notoriously high (Odze et al. 2006; Allende et al. 2014).

Diagnostic biopsies for light microscopy are usually placed in a fixative medium (formalin) at the moment at which the tissue is retrieved; failure to comply with this step may lead to autolytic changes that hamper histological interpretation. Endoscopists are discouraged to send tissue on dry gauze or in saline as the artifactual distortion that results from poor fixation can render a biopsy unreadable. Adequate fixation is also of outmost importance for the performance of ancillary studies. The field of molecular biology has dramatically advanced in the last decade and the majority of the auxiliary tests that once required fresh tissue can now be performed on paraffin embedded material (Lee et al. 2012; Fairley et al. 2012). Unfortunately, it is not an infrequent event that critical studies with major treatment implications yield equivocal results due to improper specimen handling.

The majority of the common pathological processes found in colorectal and anal biopsies can be easily identified under the microscope using a routine hematoxilin and eosin stain. Special stains are occasionally used to support specific suspected diagnosis. For example, occasionally a trichrome stain is utilized to better visualize a thickened basement membrane in collagenous colitis (Jaskiewicz et al. 2006). Immunoperoxidase stains are a type of technique that relies on the recognition of a particular antigen present in the cell of interest – neoplastic, stromal, and inflammatory cells as well as microorganisms by synthetic antibodies both monoclonal and polyclonal (Mesa-Tejada et al. 1977). This type of diagnostic modality is especially valuable to identify specific infectious agents including CMV, herpes virus, and other organisms that affect the intestinal tract (Robey et al. 1988), as well as to differentiate primary colorectal and anal tumors versus those from metastasis from other primaries. Typically colorectal tumors show a distinctive immunophenotype characterized by positive labeling with cytokeratin 20 and CDX2. Although this staining profile is not entirely specific as other gastrointestinal tumors share this pattern, positivity with these markers coupled with absence of staining with other antibodies is usually specific enough to rule out metastasis from primaries arising in the gynecological or genitourinary tract (Chiang et al. 2012).

Pathology reports of diagnostic biopsies should not only be accurate but also concise and easy to understand. Use of terms such as "compatible," "suspicious," and "consistent" should be avoided as they create confusion amongst clinicians.

2.2 Intraoperative Consultation and Frozen Sections

The sole purpose of an intraoperative frozen section consultation should be to guide the surgeon in the choice of the appropriate surgical intervention. Therefore, the indications for this procedure are scarce and well-determined.

2.2.1 Confirmation of a Diagnosis of Malignancy

All efforts should be attempted to render a diagnosis of malignancy prior to the definitive surgical procedure. The diagnostic accuracy of frozen sections is suboptimal as the quality of frozen section slides is inferior to that of permanent slides; the architectural and cytological detail is often obscured by the artifact introduced during freezing of the tissue, which significantly increases the level of diagnostic difficulty. However, there are situations in which the confirmation of malignancy is not possible prior to radical surgery. For example, endoscopically malignant lesions that arise in large villous lesions in which preoperative biopsies only demonstrate the adenomatous component. Similarly, at surgery, peritoneal or liver lesions that were not discovered during the preoperative work-up may become evident. In these cases, knowledge of the nature of these lesions is essential to determine the correct procedure.

2.2.2 Evaluation of the Margins of Resection

At times, the surgeon submits separate fragments of tissue with the purpose of determining the adequacy of the margin of resection; this practice is more common for rectal cancer operations in which the distance of the tumor to the distal margin would dictate if a sphincter-sparing procedure is possible (Kwak et al. 2012). Alternatively, the pathologist may have to examine the entire specimen grossly, and apply India ink on the outer surfaces before submitting the margins for microscopic examination. Sections from the margin should be taken from the area closest to the site of the tumor, either en-face or perpendicularly. If the gross lesion is at less than 1 cm from the margin, perpendicular sections should be obtained.

3 Surgical Resected Specimens

3.1 Pathological Examination of Cancer Specimens

3.1.1 Gross Examination

The gross evaluation of surgically removed cancer specimens is often underestimated. Many pathology laboratories worldwide routinely delegate this task to pathologist assistants, typically residents and fellows with a disparate degree of training and experience in handling complex specimens. Not infrequently, this results in suboptimal sampling of the tumor and its relationship to adjacent structures, including margins.

As with biopsy material, proper fixation of surgical specimens is critical to guarantee adequate preservation of the tissue that is being submitted for histological examination. It is not uncommon that samples transferred from the operating room after hours and left unrefrigerated without fixative for an extended period of time impede satisfactory microscopic evaluation. Poorly fixed tumors may exhibit peculiar histological patterns that may lead to erroneous classification and can adversely impact treatment decisions.

It is important to consider that fixation in formalin results in tissue shrinkage of about 10 % after the specimen has been fixed for 24 h. Therefore, it is advisable, when possible, to procure relevant measurements including tumor size and distance to margins preceding fixation (Goldstein et al. 1999). Photographing the specimen and the tumor is encouraged as it may be useful when subsequently analyzing the case with clinicians or presenting the case in tumor board meetings.

It is highly recommended that colorectal cancer specimens are fixed for at least 24 h as complete sections of the tumor in the fresh state are notoriously difficult to obtain. This is due to the fatty nature of the pericolonic and perirectal soft tissue that causes disruption of the tissue during processing, making evaluation of critical histopathological findings such as depth of invasion, vascular, and perineural invasion, and adequate assessment of the circumferential resection margins in rectal cancer difficult.

The gross examination of colorectal cancer specimens includes measurement of the tumor, location relative to margins, as well as the quality of surgery.

4 Evaluation of the Quality of the Surgery

One of the duties of the pathologist is to assess the quality of the specimen and to provide feedback to the surgeon. It has been demonstrated that the integrity of the removed specimens is often associated with local recurrence. Rectal cancer is a typical example of how the quality of surgery impacts prognosis. As emphasized by Heald (MacFarlane et al. 1993), the rectal wall should be excised en block with the surrounding soft tissue without any disruption in the mesorectum. A number of studies have established that a disrupted mesorectum that exposes the muscularis propria increases the rate of local and overall recurrence (Bosch and Nagtegaal 2012; Garcia-Granero et al. 2009). The quality of the mesorectum is directly related to the plane at which the dissection between the rectum and mesorectum and the adjacent pelvic structures occurs. Accordingly, the quality of the mesorectum can be divided into three categories (Nagtegaal et al. 2002):

– **Complete** (plane of surgery at the mesorectal level): The mesorectum is intact with minimal irregularities and/or defects that are smaller than 5 mm in depth (Fig. 22.1).
– **Near complete** (plane of surgery at the intramesorectal level): Defects within the mesorectal fat larger than 5 mm but without exposure of the muscularis propria.
– **Incomplete** (plane of surgery at the muscularis propria): Deep defects in the mesorectal fat that exposes the muscularis propria (Fig. 22.2).

It is of outmost importance that evaluation of the mesorectal integrity is undertaken by someone other than the surgeon performing the operation to avoid any bias. Assessment of the mesorectal quality is a relatively simple task but one which involves some prior training. It is recommended that all individuals involved in grossing these

Fig. 22.1 Complete mesorectum in a TME specimen showing an intact mesorectal surface with no defects as well as a high tie

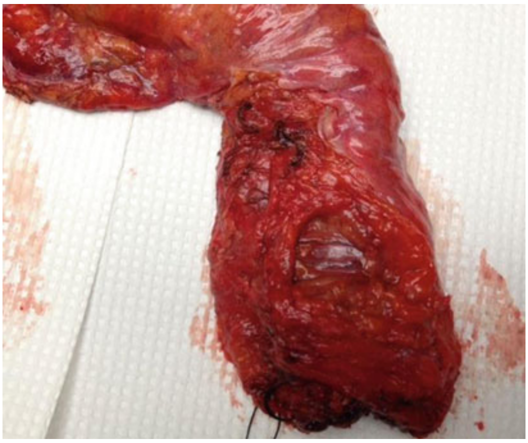

Fig. 22.2 Incomplete mesorectum in a TMS specimen showing a tear within the perirectal soft tissue exposing the muscularis mucosa

types of cases without supervision should take the proper training prior to handling these specimens. Under Quirke's leadership, the UK has created concise educational resources that are readily available to any pathologist worldwide (Quirke et al. 2012). In the USA, several multidisciplinary organizations are currently addressing this issue with the goal of providing access to educational materials that focus on training.

In recent years, a few studies have shown that, similar to total mesorectal excision (TME), colon cancer surgery performed along embryologically-defined planes to create an intact envelope of the mesocolonic fascia followed by high tie ligation is associated with better outcomes.

Typically, specimens obtained through CME are characterized by increased distance between the tumor and ligated vessels.

Analogous to rectal cancer surgery, the plane at which the surgeon performs the specimen dissection is classified as follows:

- **Mesocolic plane**: Intact mesocolon with a smooth peritoneal-lined surface
- **Intramesocolic plane**: Moderate bulk with irregularities that do not extend into the muscularis propria
- **Muscularis propria**: Little bulk to the mesocolon with disruptions that extend to the muscularis propria

Nagtegaal and Quirke (2008) designed a morphometric study demonstrating that CME is associated with a higher number of surgeries deemed to be at the mesocolic plane, longer segments of small and large intestine, greater distances between the tumor and the high vascular ligation, as well as a larger mesenteric area.

5 Sampling of Colorectal Cancer Specimens

Adequate sampling of colorectal cancer specimens should include at a minimum:

- Sections from the tumor
- Sections from the tumor and the interface with the surrounding nonneoplastic tissue
- Margins of resection
- Samples from any abnormal appearing area separate from the primary mass
- Random sections of nonneoplastic tissue in cases in which the malignant tumor arises in the background of a previously diagnosed condition specific to each organ such as IBD, polyposis syndromes, and diverticulosis, among others
- Lymph nodes

5.1 Tumor Sampling

The number of sections that should be submitted for histological examination varies according to the size of the tumor, as well as the degree of macroscopic tumor response in rectal cancer patients who have undergone preoperative chemoradiation. For large tumors, three or four sections usually suffice, provided that the areas of deepest tumor infiltration are sampled. At times, tumor regression postneoadjuvant rectal cancer therapy is so pronounced that the lesional area is reduced to a minute focus of thickening or ulceration. In these circumstances, it is essential that the entire area be submitted for examination to adequately assess tumor response.

5.2 Sampling of the Margins of Resection

The status of the resection margins is critical for all members of the multidisciplinary team involved in the care of cancer patients as a positive margin may imply additional therapeutic intervention. For the surgeon performing the procedure, it has added value since involvement of the resection margins may be reflective of the quality of the surgery (Nagtegaal and Quirke 2008).

In colon cancer, the proximal and distal margins are the most pertinent but are rarely involved. In rectal cancer, emphasis that once centered on the distal margin has now shifted to the radial or circumferential margins (CRM). Numerous studies have shown that the incidence of local recurrence in rectal cancer is more frequently related to inadequate circumferential rather than distal margins (Birbeck et al. 2002; Li Destri et al. 2014). The CRM should be measured microscopically and it is generally accepted that the presence of tumor at the CRM or at less than 1 mm represents a positive margin (Fig. 22.3). Due to the impact of the CRM status as a predictor of recurrence, it is essential for pathologists to consistently measure the distance of the tumor to the CRM. It is recommended that, following evaluation of the mesorectal integrity, the perirectal soft tissue surrounding the tumor is inked before the specimen is opened. Scissors should be used to open the specimen longitudinally to avoid cutting through the tumor as this may lead to artifactual retraction of the CRM. It is for this reason that surgeons are discouraged from opening colorectal cancer specimens in the operating room. This is to ensure that for rectal cancer cases, the distal resection margin is tumor-free. In these situations, it is advisable to request an intraoperative pathology consult so that assessment of the quality of the mesorectum and inking of the CRM are accomplished prior to opening the specimen. As previously stated, for cases in which the lesion is <1 cm from the distal margin, the sections should be taken in a perpendicular fashion to include the margin and the tumor in the same section. This allows accurate microscopic calculation of the distance between the margin and the tumor (Fig. 22.4).

Fig. 22.3 Transverse section through the specimen reveals the tumor reaching the circumferential margin of resection

5.3 Lymph Node Dissection

The presence of lymph node metastasis not only dictates prognosis but also impacts treatment decisions since the majority of patients with positive nodal disease are offered further treatment. It is important to acknowledge that the pathologist should have the same accountability as the surgeon in adequate lymph node retrieval. It is well known that the number of lymph nodes dissected from colon and rectal specimens has been used to judge the quality of the surgery and that a minimum of 12 nodes is required from each cancer specimen (Dillman et al. 2009; Stocchi et al. 2011). Although the number of retrieved nodes is, to a certain extent, related to the surgeon's skill, it has been shown that another important variable influencing the number of dissected lymph nodes is the effort and diligence invested by the pathologist (Sarli et al. 2005). Although the adverse impact of positive nodes has been clearly demonstrated in patients with colorectal cancer, more recently it has become evident that the total

Fig. 22.4 Sampling of the distal margin of resection in rectal cancer specimen in which the tumor is closely approximates the margin are taken in a perpendicular fashion

number of dissected lymph nodes, regardless of the status (positive or negative), influences outcome (Swanson et al. 2003; Le Voyer et al. 2003). Several studies have shown a positive correlation between the total number of lymph nodes dissected and survival (Chang et al. 2007). In a systematic review, Chang et al. showed that in 16/17 reports longer survival was noted with an increased number of lymph nodes evaluated. More recently, Budde et al. analyzed 147,076 colon cancer cases extracted from the SEER database and confirmed a small but significant association between the number of lymph nodes found and survival.

The reason for the influence of lymph node harvest on outcome remains unclear. It would be logical to assume that the higher the number of lymph nodes examined, the higher the possibility of identifying positive nodes (upstaging). However, other factors likely play a role. It is known that deep infiltrating (Swanson et al. 2003) and poorly differentiated lesions (Chen and Bilchik 2006) and patients' age (Soreide et al. 2006) are all associated with a higher number of lymph nodes retrieved. It is also possible that interactions between the tumor and the host occur at the molecular level, where malignant cells somehow stimulate the host's immune response. In this regard, it has been shown that microsatellite unstable tumors tend to be correlated with high lymph node yield (Soreide et al. 2006). In order to increase lymph node harvest, several enhancing techniques have been tested over the years and have been shown to boost the number of dissected lymph nodes. Universal acceptance of these enhancing methods among pathologists has been disparate due to higher cost and lengthier procedures. Furthermore, certain techniques contain toxic chemicals such as xylene and acetone (Sanchez et al. 1997). More recently, simpler methods that entail soaking the mesocolon in pure alcohol for 24 h have been applied with great success (Wang et al. 2009). It is now well established that patients with advanced rectal cancer benefit from preoperative chemoradiation (Bosset et al. 2006). It is also well known that lymph node assessment in these situations is challenging as radiation leads to shrinkage of the lymphoid tissue, rendering smaller lymph nodes more difficult to identify using the traditional palpation and visualization methods (Wang et al. 2009). The utilization of techniques such as xylene, acetone, and alcohol soaking to enhance lymph node retrieval has been encouraged in this situation (Wang et al. 2009; Chapman et al. 2013).

5.4 Evaluation of Tumor Specimens Following Preoperative Therapy

As previously mentioned, patients with advanced rectal tumors are often offered neoadjuvant chemoradiation (Bosset et al. 2006). The pathologist's role in these cases is to determine the amount of residual tumor burden or tumor regression. In the absence of obvious tumor after a known history of chemotherapy and/or radiation, areas of scarring should be searched and correlated with the patient's prior diagnostic procedure for the location of the original tumor. Once the area of interest is identified, it should be sampled in its entirety to evaluate for residual malignancy (MacGregor et al. 2012).

6 Microscopy

6.1 Permanent Sections

Since Dukes' initial observations regarding the prognostic impact of depth of tumor invasion into the colorectal wall, pathologists have consistently assessed and reported these variables. The tumor stage, both in Dukes and the TNM classification, is based on the degree of the anatomical involvement of the colorectal wall (Table 22.1). The influence on prognosis of the depth of tumor invasion into the colorectal wall is of great importance (Edge et al. 2010). This is best exemplified in the substratification of stage II colorectal cancer into Stages IIA, IIB, and IIC. In Stage IIA, the tumor infiltrates through the muscularis propria into the pericolorectal tissues (pT3). In stage IIB, tumor penetrates into the surface of the visceral peritoneum (pT4a), and in stage IIC, tumor directly invades or is adherent to other organs or structures (pT4b). Deeper tumor invasion through the wall is associated with a worse prognosis. According to the 7th edition of the AJCC staging manual, the 5-year observed survival rate for stage IIA is 67 %; for stage IIB, 59 %; and for stage IIC, 37 % (Edge et al. 2010).

Although the task of determining the depth of tumor invasion may appear simple and relatively straightforward, pathological staging may occasionally pose difficulties. The most common example is differentiation of extensive involvement of the pericolonic soft tissues (pT3) from the infiltration of the serosa by neoplastic cells (pT4a) (Fig. 22.5). Both situations are generally classified as stage II; however, the former falls into substage IIB while the latter would be classified as substage IIC, which has been related to poorer outcomes justifying the use of postoperative chemotherapy. Not uncommonly, histological sections are incomplete due to poor fixation or suboptimal technique and poor serosal visualization. This phenomenon may lead to substaging of

Table 22.1 AJCC colorectal cancer staging

Stage	T	NO	M	Dukes*
0	Tis	NO	MO	–
I	T1	NO	MO	A
	T2	NO	MO	A
IIA	T3	NO	MO	B
IIB	T4a	NO	MO	B
IIC	T4b	NO	MO	B
IIIA	T1-T2	N1/N1c	MO	C
	T1	N2a	MO	C
IIIB	T3-T4a	N1/N1c	MO	C
	T2-T3	N2a	MO	C
	T1-T2	N2b	MO	C
IIIC	T4a	N2a	MO	C
	T3-T4a	N2b	MO	C
	T4b	N1-N2	MO	C
IVA	Any T	Any N	M1a	–
IVB	Any T	Any N	M1b	–

*Dukes B is a composite of better (T3 NO MO) and worse (T4 NO MO) prognositic groups, as is Dukes C (Any T N1 MO and any T N2 MO)

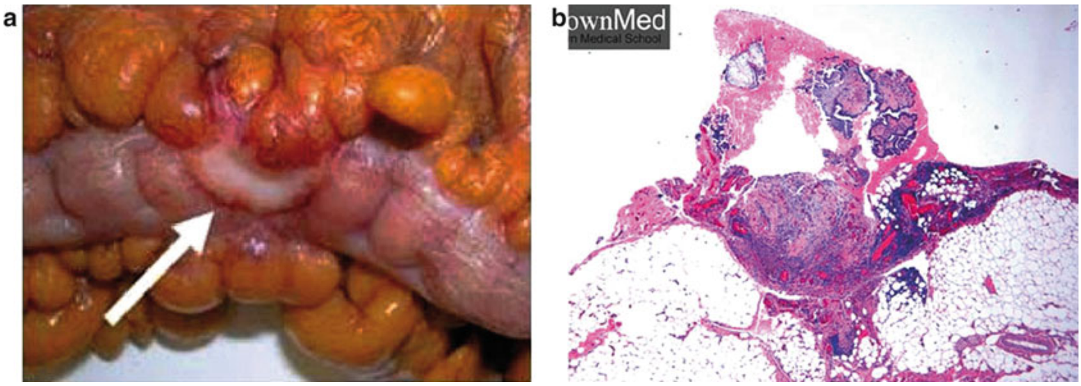

Fig. 22.5 (**a**) Colonic resection specimen viewed from the outer aspect shows and area of serosal involvement by the tumor; (**b**) Low power histological examination with H&E demonstrates malignant glands breaching the serosa and involving the peritoneal surface

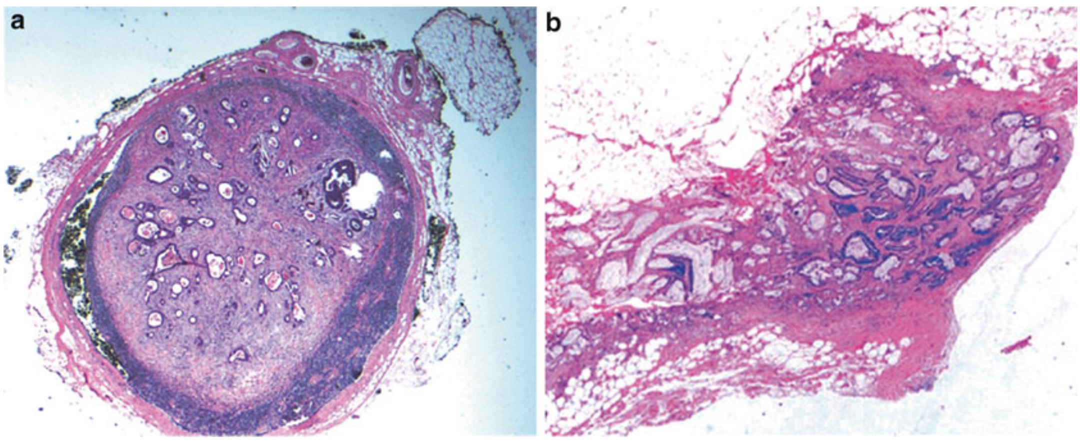

Fig. 22.6 (**a**) Low power H&E stains demonstrates a metastatic lymph node recognizable by the round shape and residual rim of lymphocytes (H&E ×1000); (**b**) Scanning magnification reveals a mesenteric tumor deposit, notice the irregular shape and the absence of identifiable residual lymph node structure (H&E ×1000)

a pT4 lesion to a pT3 lesion. It is therefore of outmost importance that pathologists carefully evaluate the quality of the histological slides being reviewed.

Generally, histological evaluation of the lymph nodes is a relatively simple task. However, small foci of metastatic cells or individual cells may be difficult to recognize, leading to understaging. Although the significance of isolated metastatic cells within the lymph nodes has been controversial over the years, more recently several authors have shown that micrometastasis in colorectal cancer is associated with poorer outcomes than actual pN0 (Nissan et al. 2012). It is for this reason that some pathologists have advocated the use of immunohistochemical stains such CK20 to highlight tumor cells (Nissan et al. 2012). Lymph nodes that have been entirely replaced by tumor and the so-called tumor deposits that result from vascular invasion in the soft tissue surrounding the colon and the rectum may be extremely difficult to locate. Moreover, this distinction is based on rather subjective and ever-changing criteria (Fig. 22.6) (Rock et al. 2014). In general, a stellate or irregular appearance of the tumoral focus would favor a metastatic tumor deposit due to venous invasion, whereas a round configuration would support a lymph node metastasis. This

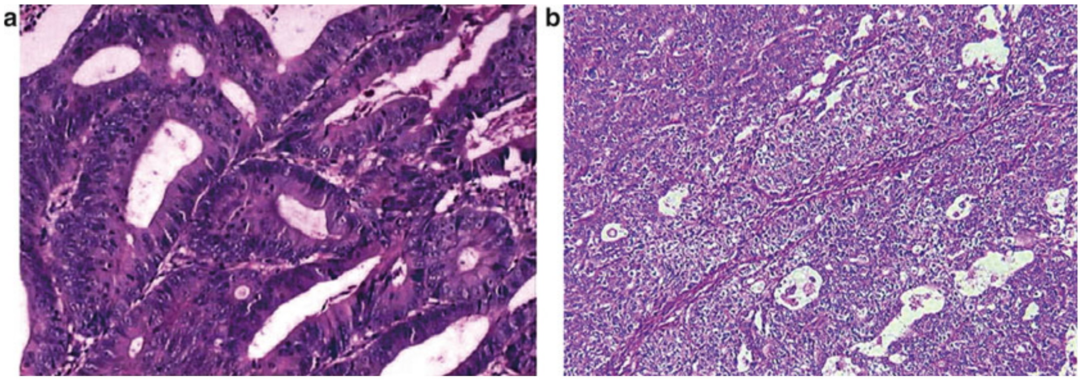

Fig. 22.7 (**a**) Medium power of a colonic adenocarcinoma exhibiting well differentiation. The malignant cells are arranged in clearly identifiable glands. (H&E ×2000); (**b**) Low power of this poorly differentiated malignant tumor demonstrates sheets of neoplastic cells with no recognizable glandular architecture, this type of malignancy may required of special stains to confirm its adenocarcinoma nature (H&E ×1000)

issue has generated much controversy as reflected by the evolving terminology applied in the various editions of the AJCC TNM classification in which the definition of tumor deposits and metastatic lymph nodes overlap according to each specific edition (Nagtegaal et al. 2011).

6.2 Evaluation of Other Prognostic Markers

In addition to the pTN status, there are several other histopathological factors that correlate with outcome and should always be present in the pathology report. These include tumor differentiation, the presence of lymphovascular and perineural invasion, and the pattern of growth at the edge of the tumor (Compton et al. 2000; Betge et al. 2012; Liebig et al. 2009; Nakamura et al. 2008). Identification of these elements is of particular importance in stage II cases (negative lymph nodes), as further treatment in the form of chemotherapy may be appropriate for these patients.

6.3 Tumor Differentiation

Several studies have shown the impact of tumor differentiation on prognosis (Compton et al. 2000; Compton 2006). Tumor differentiation ranges from well-differentiated in which there are well-formed glands, to poorly differentiated in which glands are no longer distinguishable and instead consist of solid sheets of markedly atypical malignant cells with numerous mitoses (Fig. 22.7). As a results of a notable degree of interobserver variability in histological grading, the CAP and AJCC UICC recommends a two-tiered grading system with <50 % gland formation designated as high-grade and >50 % as well-to-moderately differentiated disease (Compton et al. 2000; Compton 2006).

6.4 Lymphovascular and Perineural Invasion

Both lymphovascular (LVI) and perineural invasion (PI) have long been recognized as prognostic indicators in colorectal cancer (Betge et al. 2012) (Fig. 22.8). However, the power to predict outcome and influence treatment is somewhat overshadowed not only by the lack of consistency among pathologists in reporting these variables, but also by the inherent inter- and intraobserver variability in the assessment of these parameters (Compton 2006). Harris et al. (2008) studied this issue in a group of 7 expert gastrointestinal pathologists. These authors demonstrated that evaluation of H&E stained slides led to an overall fair level of agreement ($k = 0.24$). As the result of

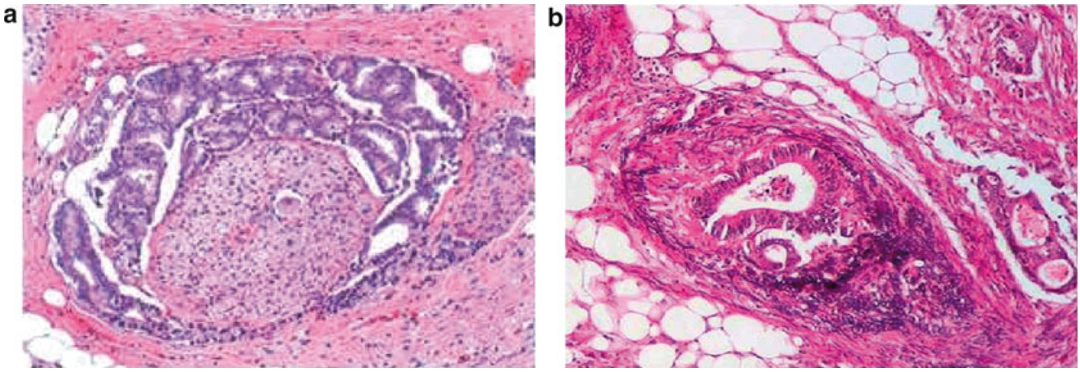

Fig. 22.8 (**a**) Perineural invasion: Malignant glands surround a large nerve trunk (H&E ×1000). (**b**) Vascular invasion: A vascular channel in the pericolonic soft tissue is completely occluded by a tumor emboli (H&E ×2000)

this phenomenon, some authors have encouraged the use of special stains to enhance lymphatic and blood vessel identification (Kingston et al. 2007). However, the application of these techniques remains controversial as they not only add time but also expense and are therefore not routinely performed.

Recognition of and reporting perineural invasion presents similar difficulties. This parameter is often underreported in published detection rates that range from 9 % to 42 % (Ceyhan et al. 2010). Similar to lymphovascular invasion, some authors have proposed the application of nerve-specific immunostains (S100) with the purpose of increasing the recognition of this marker (White et al. 2013). However, as previously stated, the added expense related to this technique renders this method unrealistic for use in routine practice.

6.5 Pattern of Growth at the Tumor Edge

Although the impact on prognosis of an infiltrative pattern of growth at the tumor border has been recognized for decades (Amato et al. 1994), in the past few years numerous publications have confirmed the finding that the phenomenon of "tumor budding" is a strong marker of prognosis (Harris et al. 2008). Tumor budding has been defined as the presence of individual cells or small clusters of cells at the invasive edge of the tumor. It has been shown that tumors that exhibit this feature are more frequently associated with lymph node and distant metastasis. It has been shown that, even in the presence of positive nodes (stage III), the presence of severe budding remains an independent poor prognostic factor (Choi et al. 2007).

6.6 Communication of Pathological Findings to the Clinicians

Many of the postoperative treatment decisions in cancer patients depend on the pathological findings present in the surgical resection specimen. The anatomical extent of the disease represented by pathological tumor (pT) and nodal stage (pN) most commonly conveyed through the TNM staging system is of outmost importance to plan additional therapy and to predict outcome. Although pTNM is the most powerful marker of prognosis in cancer patients, it does not provide any information concerning the adequacy of the resection (margin status), the quality of the surgery, and the presence or absence of other histopathological or molecular factors that influence outcome. It is therefore imperative that all the relevant data are communicated to the clinician through the pathology report. In this regard it has been shown that the use of structured synoptic reports significantly decreases the possibility of omitting critical information (Zarbo 1992).

Table 22.2 Gross and histological features that aid in the differential diagnosis between ulcerative colitis and Crohn's disease

Features	Ulcerative colitis	Crohn's disease
Pattern of involvement	Diffuse	Segmental
Rectal involvement	Present	Variable
Fissures	Rare	Common
Transmural involvement	Only in areas of deep ulceration	Common
Ileal involvement	Only in "backwash ileitis"	Common
Lymphoid aggregates	Only in areas of deep ulceration	Common
Granulomas	Only related to ruptured crypts	Epithelioid granulomas unrelated to ruptured crypts

7 Evaluation of Nonneoplastic Conditions Involving the Colon and Rectum

It is often the clinician's expectation that the pathologist will provide a specific diagnosis on a biopsy and/or surgical specimen for nonneoplastic conditions. Although some entities including microscopic colitis, ischemic colitis, radiation colitis, and IBD have relative specific features, it is important to realize that there are no pathognomonic characteristics for any of these conditions that would allow a categorical diagnosis. Instead, pathologists search for patterns of injury that would favor one disease over the other.

One of the most important concepts when evaluating colorectal specimens is the familiarity of the pathologist with the normal anatomy and histology of the colon and rectum. The intestinal tract could be viewed as part of the immune system, the presence of inflammatory elements such as plasma cells, lymphocytes, and eosinophils is expected and it is often the increase or decrease of these components that reflects a pathological process. Terms such as "mild chronic inflammation," "nonspecific inflammation," among others are devoid of meaning; furthermore they are confusing for the clinician and should be avoided.

8 Inflammatory Bowel Disease

Most pathologists are well aware of the importance of distinguishing MUC from CD, especially during surgery when creation of an ileal pouch is an important consideration.

Although in the majority of cases these two entities display distinctive features that allow easy differentiation (Table 22.2), occasionally cases will show overlapping characteristics rendering a definitive diagnosis difficult. This is particularly relevant for cases of CD in which no small bowel involvement is present and no granulomas are found.

A detailed description of the pathological findings in UC and CD is beyond the scope of this chapter; however, a few important concepts that relate to handling of the biopsies and surgical specimens need to be addressed.

8.1 Pattern of Involvement: Diffuse Versus Segmental

Although MUC typically tends to involve the colonic tract in a diffuse, continuous fashion, notorious exceptions occur, including:

– UC in children may exhibit a segmental pattern of inflammation (Glickman et al. 2004). It is therefore essential for the pathologist to know the patient's age when examining tissue biopsies.
– Medical treatment commonly induces an unequal healing of the mucosal surface leading to a "pseudoskip pattern" of involvement (Kim et al. 1999). Therefore, knowledge of previous therapeutic drugs is important before raising the possibility of CD in this situation.
– Involvement of the cecum (cecal patch) or appendix (Dendrinos et al. 2008) in an otherwise left-sided colitis is a well-described

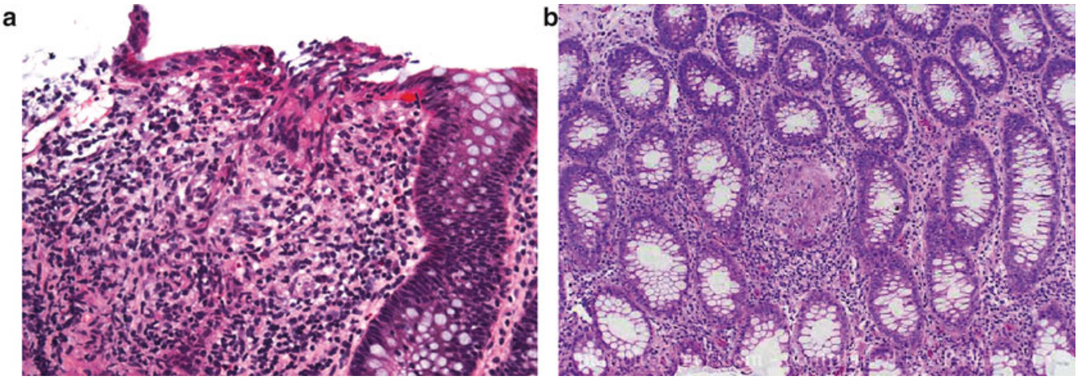

Fig. 22.9 (**a**) Granuloma surrounding a ruptured crypt in an ulcerative colitis case. Note the poor demarcation of the histiocytic collection and the pale cytoplasm related to the mucin content (H&E ×2000); (**b**) A case of Crohn's disease showing a well classic, well defined, granuloma within the lamina propria, not associated with inflamed crypts

phenomenon in UC. Pathologists need to be familiar with this occurrence to avoid misdiagnosis UC for CD.

8.2 Presence of Granulomas: Only in Crohn's Disease?

Although the presence of granulomas is extremely suggestive of CD, occasionally, mucosal granulomas surrounding injured crypts maybe seen in UC (Mahadeva et al. 2002). Once again, it is critical that pathologists are aware of this occurrence. Mucosal granulomas are usually comprised of mucin containing macrophages that result from the destruction of the glandular crypts. In most cases, experienced pathologists will be able to distinguish between these two histological findings (Fig. 22.9). The occurrence of granulomas in UC is restricted to the mucosa. Discovery of granulomas scattered throughout the rest of the colonic wall should raise a strong suspicion of CD.

8.3 Transmural Inflammation in Surgically Resected Specimens

The presence of transmural inflammation extending into the pericolonic soft tissue has traditionally been one of the differential features of CD. However, in severe forms of UC with deep ulceration, it is not at all uncommon to come across marked inflammation involving the entire thickness of the colonic wall. Therefore, pathologists need to be cautious and avoid relying on this sole finding to diagnose CD (Yantiss and Odze 2006; Abdelrazeq et al. 2005).

8.4 Involvement of the Terminal Ileum: Backwash Ileitis Versus Crohn's Disease

It is well known that patients with UC may develop inflammation of the distal 5 cm of the ileum due to retrograde flow of colonic content related to the incompetence of the ileocecal valve. This phenomenon has been referred to as "backwash ileitis" (Goldstein and Dulai 2006). Because the presence of ileal involvement is one of the characteristic features of CD, the finding of distal ileitis often raises diagnostic difficulties both in the biopsy and surgically-resected specimens.

Although clearly established criteria for the diagnosis of backwash ileitis are not defined, the most common findings that have been described include villous blunting, neutrophilic infiltrates within the lamina propria, crypt abscesses, and mild increases in the lmyphoplasmacytic cell population. Occasionally, surface ulcerations and pyloric metaplasia can be seen. The presence of extensive ileal compromise, transmural inflammation, "skip areas," and granuloma formation are not characteristic of backwash ileitis and should strongly raise the possibility of CD.

The prevalence and natural history of backwash ileitis remains controversial as long-term studies are lacking. In a study by Haskell et al. (2005) with 200 consecutive UC patients with a mean follow-up of 48.5 months, the authors reported an incidence of backwash ileitis of 17 %. The majority of patients in this cohort presented with pancolitis with severe activity. Interestingly, none of these patients demonstrated any increased pouch complications or development of dysplasia and cancer.

8.5 Examination of Biopsy Specimens

As can be deduced from the above discussion, recognition of the pattern of involvement (diffuse vs. segmental) can be of great help in differentiating UC from CD. It is therefore important that the endoscopist submits tissue obtained from the different anatomical sites in separate, properly labeled containers. In addition, it is highly recommended that no more than 4–5 fragments of tissue are placed in the same container as this makes processing difficult.

Although the search for granulomas in biopsies of IBD is customary and extremely helpful to diagnose CD, it is important to realize that the absence of granulomas does not exclude CD as this histological feature is only found in ≤40 % of CD cases (Rubio et al. 2007).

A few scoring systems to assess inflammatory activity in patients with CD have been described (Riddell et al. 1983). However, grading of the inflammatory process is not consistently recorded in the pathology report. This is an unfortunate fact as it has been shown that the histological severity of the inflammatory process predisposes to the development of dysplasia (see below).

As mentioned in the introduction, the vast majority of IBD biopsies can be adequately evaluated by routine H&E. One of the few exceptions is the use of immunostains for CMV. Pathologists should have a high index of suspicion when searching for viral inclusions, especially in the context of lingering severe symptoms refractory to therapy. Although sometimes CMV inclusions are readily seen on H&E, positive cells that were not recognizable on H&E are often discovered by immunostains (Robey et al. 1988).

8.6 Examination of Surgically Resected Specimens

Many of the basic principles displayed in the previous section also apply to the evaluation of surgical resected specimens in patients with IBD.

Gross examination of segmental, subtotal, or total colectomy for IBD should start with a thorough inspection of the serosal surface. A search for findings characteristic of CD including "fat wrapping," strictures, or fistulous tracts should always be performed. It is critical that resected specimens are promptly opened and soaked in formalin to allow preservation of the histological features. Unfortunately, not uncommonly, surgically removed samples for IBD are placed in formalin without previous opening, leading to extensive autolysis of the mucosa due to poor exposure to the fixative. Consequently, recognition of mucosal granulomas, "skip areas," and dysplasia becomes extremely difficult if not impossible.

Sections procured for histological examination should always include any suspicious areas that could harbor malignancy or dysplasia as well as random samples of the colonic mucosa every 5–10 cm.

8.7 Diagnosis of Dysplasia in IBD

The diagnosis of dysplasia in patients with IBD has traditionally relied on histological examination rendered by the pathologist. Based on this definition, dysplasia can be further subdivided according to the increasing degrees of cytological and architectural atypia into indefinite, low grade, and high grade dysplasia (Breynaert et al. 2008). As mentioned above, the development of colorectal cancer in patients with IBD is regarded as a continuum process spanning from normal, to inflamed mucosa, to dysplastic mucosa, and finally to frank invasive cancer (Navaneethan et al. 2013). The weak point of this concept is related to the significant inter- and intraobserver variability in the histological diagnosis of dysplasia in IBD. This is reflected in the wide variation in the reported incidence of dysplasia in the literature as well as the predictive value of dysplasia for the development of cancer (Montgomery 2005). It has been demonstrated that the accuracy in the diagnosis of dysplasia is higher among specialized gastrointestinal pathologists compared to general pathologists (Allen et al. 2010). Therefore, it is highly recommended that a diagnosis of dysplasia be confirmed by another pathologist. It is also important to point out that, as a consequence of the dramatic advances in endoscopic techniques achieved during the past two decades (Wanders et al. 2014), many dysplastic lesions can currently be identified endoscopically. As a result, the number of "random" biopsies obtained during surveillance for dysplasia has significantly decreased. Based on these concepts, today, more than ever, the diagnosis and treatment of dysplasia needs to be decided in the context of a multidisciplinary team. Communication between the endoscopist and the pathologist is critical. Furthermore, it is imperative that clinicians provide the pathologist with all clinical and endoscopic findings. A situation in which this concept is crystallized is the finding of the so-called adenoma-like dysplasia in a biopsy. In these cases, pathologists are often asked to differentiate between a sporadic adenoma and dysplasia associated with IBD. This task can be almost impossible in the absence of the proper endoscopic findings. Examination of the tissue surrounding an area of adenomatous changes is essential. Typically, in sporadic adenomas, the colonic mucosa in close proximity to the adenomatous changes is devoid of colitis-related inflammatory changes. Instead the presence of inflammatory activity in the tissue surrounding the adenomatous focus would be more in favor of dysplasia-associated colitis (Ingle et al. 2014). From the latter example, it can only be inferred that adequate sampling of the colonic mucosa with appropriate labeling is essential to arrive at the correct diagnosis.

9 Other Forms of Colitis

9.1 Microscopic Colitis

Most patients with microscopic colitis are middle aged or elderly women who suffer from several daily episodes of watery nonbloody chronic diarrhea that may be accompanied by abdominal pain, weight loss, and fatigue. Endoscopy and radiological images are not helpful in this entity. Instead, the inflammatory and pathological changes are only observed histologically, hence its name. The pathogenesis is unclear and while it could be multifactorial (Ingle et al. 2014), some emphasis is placed on autoimmunity (Bohr et al. 2014) The two better-known histomorphologic types of microscopic colitis are collagenous colitis (CC) and lymphocytic colitis (LC). Pathologists are making these diagnoses more frequently than in the past (Mahajan et al. 2012). While there are clinical and sometimes microscopic overlaps in the manifestations of these two entities (Münch et al. 2012; Bohr et al. 2014), the histology of each is very characteristic and they are therefore addressed separately for the purpose of this review. Multiple colonic biopsy tissues are required to establish the diagnosis and they should not be limited to the rectum (Offner et al. 1999).

9.1.1 Lymphocytic Colitis

The hallmark of this disease is the presence of epithelial lymphocytosis with >20 lymphocytes per 100 enterocytes of the surface epithelium (Lazenby et al. 1989). The epithelial cells show loss of mucin and goblet cells, and their height is decreased giving it a flattened withered appearance. In most cases the lamina propria shows moderate-to-marked increases of mononuclear cells, mostly lymphocytes but plasma cells may also be present as well as occasional neutrophils and eosinophils. A few cases with multinucleated giant cells have been described (Brown and Lambie 2008). The crypts are architecturally normal, but they may show lymphocytic cryptitis. Most cases do not require ancillary stains, but quantitation and confirmation for a histological diagnosis can be achieved by performing an immunohistochemical stain for the T-cell marker CD3. The changes in the right colon tend to be more severe than in other portions of the colon.

Lymphocytic aggregates are not infrequent in normal colonic mucosa and the lymphocytes may spill into the overlying epithelium. This creates a false picture resembling epithelial lymphocytosis but it should not be interpreted as such.

9.1.2 Collagenous Colitis

The diagnostic feature of collagenous colitis is the presence a thickened band of basement membrane of pink collagen underlying the surface epithelium. This layer is >10 μm in thickness and does not extend around the deeper crypts. It may contain capillaries, fibroblasts, and eosinophils. Most pathologists actually do not use an instrument (micrometer) to measure the collagen plate. Instead, the size of the nearby lymphocytes is used as reference, which normally measure 5 μm (Lazenby 2005). Mild increases in epithelial lymphocytes are normally seen, but occasionally these increases may be as intense as seen in LC. The lamina propria shows mild-to-moderate mononuclear infiltrates with a mixture of lymphocytes, plasma cells, mast cells, neutrophils, and eosinophils. As in LC, cases showing multinucleated giant cells have been described (Brown and Lambie 2008). The histotechnologist must attempt to properly orient the biopsy tissue to prevent tangential cuts that may mimic a thickened collagen plate. The overlying epithelium may show an absence of mucin and goblet cells and decreased cell height, appearing cuboidal instead of columnar. Detached strips of epithelium devoid of stroma that appear to have been peeled off the mucosa can be seen, leaving the mucosa denuded and exposing the thickened surface collagen (Fig. 22.10). The underlying crypt architecture is preserved. Extravasation of serum, which appears pink, may mimic collagen (Lazenby 2005). In addition, elevation of the epithelial nuclei may leave the pink cytoplasm of the basal cells, forming a band that may be erroneously interpreted as collagen. In these situations or cases of uncertainty, a stain for collagen such as trichrome will help to make the correct interpretation. A thickened collagen plate may also be seen in chronic ischemia, mucosal prolapse, diabetes, and hyperplastic mucosa (Lazenby 2005).

9.2 Ischemic Colitis

Hypoperfusion results in the pathological changes of colonic infarction and can be limited to the mucosa or may also involve the submucosa (mural); these can be acute or chronic (Herbert 2007). If they are observed in all layers of the intestinal wall, they are considered transmural.

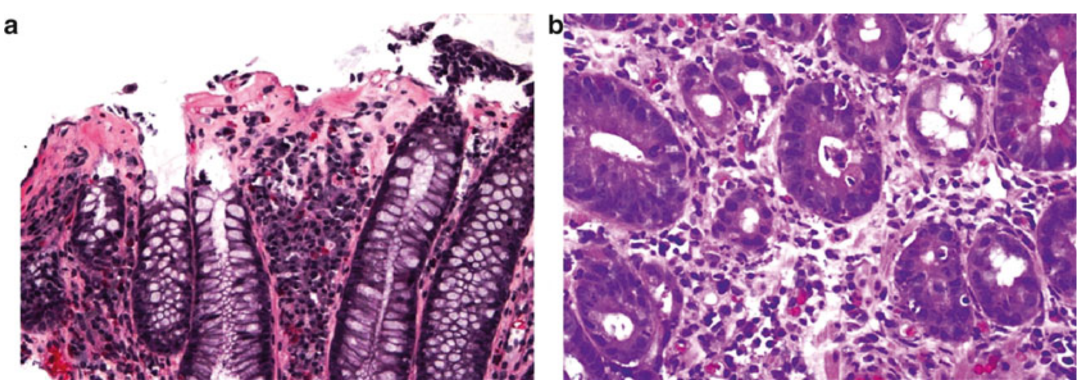

Fig. 22.10 (**a**) Typical appearance of collagenous colitis characterized by a thick and irregular basement membrane with surface epithelial injury (H&E; ×2000). (**b**) Classic case of lymphocytic colitis recognized by the prominent intraepithelial lymphocytosis (H&E; ×2000)

This usually occurs due to an occlusion of the major mesenteric arteries, causing acute injury to the intestines. Focal ischemic lesions occur when there is damage of the end arteries that penetrate the intestinal wall.

The initial hypoxic injury, when the blood supply is compromised, is followed by a secondary reperfusion injury when blood is resupplied to the hypoxic area. The main damage to the tissue is caused by reperfusion because of the presence of oxygen free radicals, neutrophils, and inflammatory mediators (Turner 2015).

The morphologic findings are dependent on the severity of the vascular compromise and the hypoxemic time. In more severe transmural infarction, there is a high risk of death because of the imminent possibility of perforation. Although any area of the colon may be affected, the most susceptible area is the splenic flexure because it is the watershed between the distribution of the superior and inferior mesenteric arteries (Washington and Carmichael 2012). The infarction appears hemorrhagic because of reperfusion to the damaged area. Initially, there is intense congestion and dusky to purple-red discoloration with ecchymosis is noted in the submucosa and serosa. In arterial occlusion, there is a sharp demarcation from uninvolved intestine, whereas in venous occlusion the injured tissue fades and gradually blends into the normal colonic wall. Microscopically, there is edema, hemorrhage, and necrosis of the mucosa with disappearance of the epithelial layer (Zou et al. 2009). The nuclei of the muscle layers fade and are difficult to observe. Perforation and gangrenous changes occur in 1–4 days with minimal inflammatory response. Because of the rapid evolution, the specimens that arrive to the pathology laboratory are either excisions or postmortem.

Mucosal and mural ischemia as well as chronic ischemia may be less catastrophic than transmural infarction with a more subacute and chronic evolution. Thus, they are more likely to be diagnosed histologically by means of biopsy preoperatively, or before the underlying condition is resolved (Zou et al. 2009). Because of the nonspecific clinical findings and less straightforward histological features, diagnosis can be challenging. Subtle changes in early ischemia may include congestion of lamina propria vessels, blood extravasation with patchy necrosis, and sloughing of the surface epithelium in the absence of inflammation. Fibrin thrombi can be observed. This finding is a hallmark of ischemia and very useful in subtle cases in which biopsy tissues are examined. However, fibrin thrombi can also be a secondary effect due to infection by enterohemorrhagic *E. coli* (Kelly et al. 1990). As the injury progresses, there is complete mucosal necrosis with no viable epithelium leaving only the presence of residual lamina propria. In chronic ischemia, these changes are accompanied by inflammation of the mucosa with ulceration, which can be confused with other types of colitis. Finally, the epithelial atypia may be so marked as to be confused with dysplasia. The observer must be aware of the clinical scenario prior to making a definitive diagnosis of dysplasia (Abraham et al. 2014).

9.3 Pseudomembranous Colitis

Pseudomembranous colitis is a form of acute colitis. It is usually caused by the enterotoxin TcdA and the cytotoxin TcdB, produced by *Clostridum difficile* that is a Gram-positive, anaerobic, and spore-forming bacterium (Rineh et al. 2014). The disease develops when the patient ingests the spores via personal contact or the environment. The term pseudomembrane refers to the histological observation of a layer of inflammatory cellular debris overlying a partially denuded mucosa in which dilated crypts filled with neutrophils open up to the surface in a volcano-like appearance, spilling the contents. The spilled contents become confluent with mucus and fibrin forming the pseudomembrane, which lacks epithelial elements and is, thus, not a true membrane. The main risk factor for the disease is prior use of antibiotics in the hospital setting. Diagnosis is confirmed by the detection of cytoxin in the stool (Kutty et al. 2010). This confirmation is necessary as the presence of pseudomembranes in biopsy tissues. Although characteristic, it is not pathognomonic of *C. difficile* and can be seen in ischemia and other types of infections.

Endoscopy may show plaques of yellow fibrin adherent to congested colonic mucosa.

9.4 Mucosal Prolapse Syndrome (Solitary Rectal Ulcer)

Mucosal prolapse syndrome involves a lesion generally located in the anterior wall of the rectum. The term mucosal prolapse is more accurate than solitary rectal ulcer in that the lesion is not always ulcerated or solitary. Symptoms include pain or discomfort with constipation, blood, and mucus from the rectum, and alternating episodes of diarrhea and constipation (Torres et al. 2007). The cause appears to be a malfunction of the puborectalis muscle leading to straining on defecation (Mackle and Parks 1986; Rutter and Riddell 1975). Straining causes mucosal prolapse of the rectum creating polypoid masses that can ulcerate because of local ischemic changes or the transit of fecal material eroding the surface epithelium. Histologically, there is a proliferation of fibroblasts and smooth muscle fibers in the lamina propria, some of which run perpendicular to the crypts. Ectatic capillaries are also present and inflammation of the lamina propria may be mild. In some cases, the fibrosis is more predominant than the muscle hyperplasia (Sharara et al. 2005). The epithelium may acquire a villiform appearance of the crypts and, thus, needs to be distinguished from a villous adenoma or a serrated polyp. The latter is of interest as it has been shown that up to 38 % of mucosal prolapse lesions have histological changes that mimic sessile serrated polyps; of the serrated polyps, 20 % have focal loss of MLH1 on immunohistochemistry, indicating a potential for preneoplastic change. This phenomenon may reflect an increased propensity for neoplastic progression in response to repeated trauma and repair process in certain cases of mucosal prolapse (Ball et al. 2005). Included in the differential diagnosis is a hamartomatous Peutz-Jeghers polyp; however, in mucosal prolapse there is absence of the branching arrangement of the glands. The presence of muscle fibers in the lamina propria argues against the possibility of IBD (Levine et al. 1988). As the lesion grows, dilated crypts may be seen in the deeper aspects of the lesion that mimic colitis cystica profunda.

9.5 Colitis Cystica Polyposa/Profunda

Colitis cystica polyposa/profundais a reactive process characterized by the presence of mucin-filled cysts in the submucosa, muscularis propria, or serosa. Although most cases described have been individual case reports, the lesions are associated with entities that have in common damage of the mucosal epithelium. This includes mucosal prolapse syndrome, IBD, diverticular disease, and radiation colitis in which the mucosal epithelium herniates into the deeper layers of the intestinal wall likely due to repetitive episodes of acute injury and repair (Qayed et al. 2011; Toll and Palazzo 2009). The cysts may be filled with mucin and are usually lined by colonic-type epithelium surrounded by a rim of lamina propria. Some cysts may have ruptured, further eliciting an inflammatory reaction and others may lack epithelium. The epithelium is benign in nature and shows no dysplastic changes unless a previous area of overlying dysplasia in the setting of IBD had herniated. The main differential diagnosis is mucinous adenocarcinoma in which there are irregular-shaped glands, papillary fronds, and cytological atypia.

9.6 Radiation Colitis

Patients at greatest risk of developing colitis after radiation are those in whom it was administered for treatment of cervical, prostate, or rectal carcinoma. In the acute form, damage occurs in the epithelium a few hours to days after exposure and then healing within 8 weeks. There is epithelial flattening, decreased mitotic activity, and loss of mucin of the enterocytes accompanying erosion and tissue eosinophilia. The overlying epithelium may show cytological atypia, which should not be interpreted as dysplasia. In the chronic form, manifestations may occur from 6 to 24 months after exposure and the targets of the injury are mesenchymal structures of the colonic wall, particularly submucosal and mesenteric blood vessels. The vessels undergo thickening with hyalinization of the wall, intimal fibroplasia, and foamy macrophages (Leupin et al. 2002). As a consequence, the luminal narrowing leads to ischemia and stricture. However, the capillaries in the lamina propria are characteristically dilated. The stroma appears hyalinized and fibroblasts are atypical. Differential diagnosis of acute radiation colitis includes eosinophilic gastroenteritis whereas for chronic radiation mimickers include collagenous colitis and mucosal prolapse.

9.7 Diverticular Disease-Associated (Segmental) Colitis

Diverticular disease-associated (segmental) colitis refers to the presence of inflammatory changes in the segment of mucosa that separates diverticular pouches in patients with diverticular disease, usually affecting the sigmoid of individuals older than 60 years of age. The inflammatory changes are independent of the presence or absence of diverticulitis. This entity has several names including crescentic colitis, crescentic mucosal fold, sigmoid colitis, sigmoiditis, and segmental colitis-associated diverticulosis (SCAD) (Haboubi and Alqudah 2012), which add to the confusion among pathologists and clinicians. Because of the histological and endoscopic features, it tends to be confused with IBD, in particular MUC, hence the importance of diagnosing it correctly. In contrast to MUC, colitis related to diverticular disease usually spares the rectum.

Histologically, the mucosa shows active colitis with the presence of cryptitis and crypt abscesses accompanied by features of chronicity such as glandular architectural distortion, plasmacytosis of the lamina propria, and Paneth cell metaplasia. In resection specimens, there may be findings that resemble CD with transmural inflammation and granulomas likely due to the associated diverticulitis. The distinction rests on the absence of CD in other areas of the gastrointestinal tract. In some cases, there is mucosal protrusion in a polypoid fashion and in others, a microscopic colitis appearance is noted (Makapugay and Dean 1996).

9.8 Drug-Induced Colitis

Various pharmaceutical agents have been implicated in the development of colonic inflammation. For instance, antibiotics may cause pseudomembranous colitis and chemotherapeutic drugs may induce ulceration throughout the colon. Other medications may cause lesions that are more difficult to distinguish form other colitides. One example is with the use of nonsteroidal anti-inflammatory drugs (NSAIDs) in which features of chronicity including architectural distortion and plasmacytosis of the lamina propria are observed in the biopsy tissues. In addition, active colitis is present with most cases showing cryptitis or erosion. As a result, the morphology resembles that of idiopathic IBD. Most cases of NSAID-induced colitis are located in the right colon (Deshpande et al. 2010), arguing against MUC. The absence of granulomas and ileal involvement would separate MUC from CD. As mentioned above, mycophenolate has been associated with the presence of apoptotic bodies in the crypt epithelium in a similar fashion to GVHD. Patients with hyperkalemia receiving kayexalate (sodium polystyrene), an ion-exchange resin administered with sorbitol in uremic patients to bind intraluminal potassium that is then excreted, may show crystalloid material associated with necrotic mucosa (Rashid and Hamilton 1997; Parfitt and Driman 2007).

9.9 Acute Infectious (Self-limited) Colitis

Acute infectious (self-limited) colitis describes a type of colitis caused by infection in which histological inflammatory changes along with clinical symptoms resolve within 2–4 weeks. The causative agents primarily include bacteria such as *Campylobacter*, *Salmonella*, and *Shigella*, viruses, and parasites. However, the pathophysiology of it is unknown (Surawicz 2008). Since the clinical presentation may be similar to acute MUC, a biopsy may be obtained to distinguish these two entities.

Unlike IBD, there is no overt crypt architectural distortion or heavy plasma cell infiltrates in self-limited colitis. However, there are neutrophilic infiltrates in the crypts whereas the lamina propria contains lymphocytes, histiocytes, and neutrophils. Ruptured-gland granulomas may be seen. *Yersinia enterocolitica* may present histologically with features very similar to those of IBD, particularly right-sided colonic CD with the presence of granulomas.

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Specifics and Pitfalls in Diagnosis of Inflammatory Bowel Diseases

23

Art Hiranyakas

Contents

Abstract
Inflammatory bowel disease is increasing in prevalence in Western society. Ulcerative colitis, Crohn's disease, and indeterminate colitis may manifest of the similar disorder causing difficulty in making the diagnosis, however on therapeutic grounds must be distinguished. Despite of the true diagnostic challenge, the two major groups of inflammatory bowel diseases (ulcerative colitis and Crohn's disease) often have different signs, symptoms, and histopathologic presentations that make it possible to be differentiated. Successful diagnosis is reliant on careful clinicopathological correlation and recognizing potential pitfalls. The accuracy in making the diagnosis will lead to the best available therapeutic plans and the reduction in physical and emotional morbidity and mortality.

Inflammatory bowel diseases are chronic inflammatory disorders of the gastrointestinal system affecting more than one million people in the United States, over 1.5 million people in Europe, and several millions worldwide. Studies have shown the trends that the incidence of the disease continues to increase in many areas of the world (Molodecky et al. 2012; Ng et al. 2013; Burisch et al. 2013; Cosnes et al. 2011).

The diagnosis of inflammatory bowel disease can be challenging. However, the more challenge is perhaps to subclassify between ulcerative colitis and Crohn's disease. This certainly requires

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efforts of a multidisciplinary team. Even though the pathogenesis of these disorders is unknown (Le Berre et al. 1995; Caroline and Evers 1987; Tanaka and Riddell 1990; Geboes 2001a), chronic inflammation has been reported as the major cause for both conditions. Despite of the similar etiology, the importance is to tailor clinical management, as each entity often involves specific therapeutic strategies and prognosis. For instance, there is a strong clinical need to classify inflammatory bowel disease patients definitively as ulcerative colitis or Crohn's disease (or other), since an **ileal pouch-anal anastomosis (IPAA)** "pouch" procedure is generally contradicted in Crohn's disease due to a high risk of morbidity related to **pouchitis**, fistulas, incontinence, or anastomosis leaks (Sagar et al. 1996; Grobler et al. 1993).

1 Diagnosis of Inflammatory Bowel Disease

Despite of other pathologic mimics of inflammatory bowel disease, such as NSAIDs colitis, diverticular disease-associated colitis, pseudomembranous colitis, radiation or ischemic colitis, or infectious colitis, approximately 5–15 % of inflammatory bowel disease cases, a definite diagnosis of ulcerative colitis or Crohn's disease cannot be established, in which case the term "**indeterminate**" **colitis** is used (Odze 2003; Meucci et al. 1999; Nuij et al. 2013; Burisch et al. 2014). Approximately 20 % of patients with indeterminate colitis develop severe pouch complications, which is intermediate in frequency between ulcerative colitis (8–10 %) and Crohn's disease (30–40 %) (Odze 2003). Up to 14 % of patients classified as ulcerative colitis and Crohn's disease, the diagnosis changes over time (Moum et al. 1997; Myren et al. 1988; Abraham et al. 2012; Marcello et al. 1997; Henriksen et al. 2006; Melmed et al. 2007). There are no specific clinical or laboratory features that may lead to an accurate diagnosis. The important clues in order to establish a reliable diagnosis of ulcerative colitis or Crohn's disease are derived from the evaluation of pathologic material in conjunction with clinical, laboratory, radiologic, and endoscopic features. A summary of the classic microscopic features of ulcerative colitis and Crohn's disease is noted in Table 23.1 (Itzkowitz 1986).

Table 23.1 Distinguished features of idiopathic inflammatory bowel disease

	Ulcerative colitis	Crohn's disease
Sign or symptom		
Rectal bleeding	Very common (90 %)	Uncommon (occult, 50 %)
Diarrhea	Early; frequent, small stools	Less distressing
Abdominal pain	Predefecation urgency	Colicky, postprandial
Fever	Uncommon	In 30–50 % if small bowel involved
		Frequent right-lower quadrant
Palpable mass	Rare	Common
		In <10 %
Fistula	Uncommon	
Toxic megacolon	In 3–20 %	
Endoscopy	Diffuse pinpoint ulcers, abnormal surrounding mucosa, continuous disease, pseudopolyps (in healing phase)	Discrete aphthous ulcers, normal surrounding mucosa, patchy disease, cobblestoning, fissures
Histology		
Inflammation	Mucosa	Transmural
Crypt abscesses	In >70 %	Uncommon
Granulomas	In 7 %	In 60–70 %; deep submucosa
		Slightly depleted
Mucus	Depleted	Seldom prominent
Vascularity	Prominent	
Radiography		
Rectal involvement	In 95 %	Often absent
Distribution	Continuous with rectum	Skips area
Terminal ileum	Usually normal	Stenotic, stricture
Fistula or stricture	Rare (exception: "backwash")	Frequent
Mucosa	Shallow ulcers, pseudopolyps	Longitudinal fissures, cobblestoning

Ulcerative colitis is a mucosal inflammation characterized by crypt abscesses, mucosal depletion, lack of **granuloma** (except those related to mucin or foreign bodies), and prominent vascularity (Odze 2003). The rectum is almost always involved, with continuous disease present to variable extents proximally. Rectal urgency, **tenesmus**, and, occasionally, severe constipation represent the classical complaints of rectal involvement. The left-sided or extensive disease typically has chronic diarrhea with nocturnal defecation and crampy abdominal pain (Dignass et al. 2012a). Terminal ileum involvement is rare with the exception of a minor degree of inflammation associated with backwash ileitis (Odze 2003). Endoscopically, diffused pinpoint ulcers and friability are seen, and the mucosa surrounding the ulcers is usually abnormal. Frequently, bloody diarrhea and predefecation **urgency** are characteristic complaints (Itzkowitz 1986).

The diagnosis of Crohn's diseases may be relatively easy when the different gastrointestinal tract areas or extraluminal complications such as strictures, abscesses, or fistula are involved. However, approximately a third of Crohn's disease patients have a pure colonic location (Louis et al. 2001; Peyrin-Biroulet et al. 2010; Veloso et al. 1996; Freeman 2003; Nikolaus and Schreiber 2007). Crohn's disease is typically a patchy, segmental inflammatory process that has less severe disease in the distal colon compared to the proximal colon (Odze 2003). Crohn's disease often spares the rectum and results in a deeper, transmural disease. Submucosal granuloma formation is the hallmarks of the disease but is by no means pathognomonic. Endoscopically, **aphthous ulcers** with relatively normal surrounding mucosa may be seen early in the course of the disease. More severe inflammation may cause cobblestone appearance of the mucosa with longitudinal **fissures**, fistula formation, and strictures. Perianal disease can occur in up to 10 % of new-onset Crohn's disease (Burisch et al. 2014; Peyrin-Biroulet et al. 2012; Bouguen et al. 2010). Bloody diarrhea is somewhat less common than in ulcerative colitis, and postprandial colicky pain is often described. If terminal ileum is involved, fever and palpable right-lower-quadrant mass may be detected (Itzkowitz 1986). Upper gastrointestinal tract involvement as well as a less pronounced degree of mucosal architectural changes, and mucin depletion, compared to ulcerative colitis can also be found (Odze 2003).

A definitive diagnosis of inflammatory bowel disease is not always straightforward. Historically, most cases of indeterminate colitis are related to **fulminant colitis** (i.e., severe colitis with systemic toxicity and often associated with colonic dilatation), a condition in which the classic features of ulcerative colitis or Crohn's disease may be obscured by severe ulceration with early superficial fissuring ulceration, transmural lymphoid aggregates, and relative rectal sparing (Odze 2003). Recently, the pathologists use the term "indeterminate" colitis when a definite diagnosis cannot be established at the time of surgical sign out (Price 1978). This may be due to either insufficient data or prominent overlapping features between these two disorders (Marcello et al. 1997; Yu et al. 2000; Nicholls and Wells 1992; Farmer et al. 2000). Indeterminate colitis has been associated with worse prognosis (than ulcerative colitis) because of the higher frequency of relapses (Stewenius et al. 1996), the increased risk of **colon cancer** (Stewenius et al. 1995), and less favorable outcomes after ileal pouch-anal anastomosis (Tyler et al. 2013). Approximately 80 % of cases, the true nature of the patient's underlying inflammatory bowel disease usually becomes apparent within a few years (Meucci et al. 1999).

The international guidelines have recommended that for a definitive diagnosis of inflammatory bowel disease, the pathologist requires the following: (a) a minimum set of patient's information about clinical history and endoscopic pattern and (b) biopsy sampling and handling procedures of adequate quality in both the endoscopy room and histology laboratory (Dignass et al. 2012a, b; Magro et al. 2013; Van Assche et al. 2010; Bernstein et al. 2010; Kornbluth and Sachar 2010; Mowat et al. 2011). Recently, Canavese et al. demonstrate that the recommended guidelines for diagnosing IBD are frequently disregarded in clinical practice (Canavese et al. 2015). Three hundred forty-five cases from 13 centers were retrospectively analyzed. The diagnosis was conclusive only in 47 % of the cases. The date of onset and treatment were available for 13 % and 16 % of the cases, respectively. Endoscopy information was accessible for 77 % of the cases. Endoscopic mapping was completed in 13 % of the cases. In no cases were the biopsies oriented on acetate strips. The authors concluded that multidisciplinary education should be emphasized for making an adequate diagnosis of inflammatory bowel disease and managing the condition.

Biomarkers may be helpful in classifying ulcerative colitis and Crohn's disease. **Anti-neutrophil cytoplasmic antibodies (ANCA)** and **anti-saccharomyces cerevisiae (ASCA)** have been widely used; for instance, ANCA are detected in the serum of 60–70 % of ulcerative colitis patients, but in only 10–40 % of Crohn's disease patients. ASCA are present in 50–60 % of Crohn's disease patients and has a sensitivity of 67 % and a specificity of 92 % as a serum marker for Crohn's disease. Interestingly, of the Crohn's disease patients who are ANCA positive, most have left-sided colitis with clinical, endoscopic, and/or histologic features of ulcerative colitis. Furthermore, biomarkers is also very helpful in the prediction of further development of ulcerative colitis and Crohn's disease in unclassified patients, determination of disease activity, risk stratification, and prediction of response to therapy (Bouguen et al. 2015; Lewis 2011; Iskandar and Ciorba 2012).

2 Diagnostic Pitfalls

Apart from the group of indeterminate colitis (confined only to the operative specimen) and a spectrum of architectural damage and inflammatory features that are often nonspecific and may overlap with the features of the comprehensive group of noninflammatory bowel disease colitis (Yantiss and Odze 2007; Dejaco et al. 2003; Geboes 2001b; Tanaka et al. 1999), diagnostic confusion may occur with a number of other exceptions to the classic principles of inflammatory bowel disease pathology. Inflammatory bowel disease restricted to the colon that cannot be allocated to the ulcerative colitis or Crohn's disease category is best termed "inflammatory bowel disease unclassified" (Van Assche et al. 2010; Silverberg et al. 2005). In 2003, Odze et al. summarized unusual morphologic patterns of disease in ulcerative colitis as follow (Odze 2003).

1. Effect of oral and topical therapy

   Odze et al., in 1993, reported the first report demonstrating that "fixed" chronic features in ulcerative colitis may revert to normal in the natural course of the patient's illness (Odze et al. 1993). One hundred twenty-three rectal mucosal biopsies all from the same anatomic location from 14 patients with pathologically confirmed ulcerative colitis treated with either **5-aminosalicylic acid (5-ASA)** or placebo enemas. Overall, 29 % of biopsies from 64 % of patients were histologically normal. Patients treated with 5-ASA enemas showed a significantly higher percentage of normal biopsies (36 % ASA group *vs.* 12 % placebo group; $p = 0.005$). Kim et al. in 1999 reported in a retrospective review of 32 patients (median follow-up of 15 years) that in 47 (27 %) follow-up endoscopies demonstrated either patchy disease, rectal sparing, or both in 59 % of the patients. No significant difference in treatment, including steroid use and rectal therapy, between those with patchiness and/or rectal sparing and those without (Kim et al. 1999).
2. Ascending colon, cecum, and appendiceal involvement as "**skip**" **lesions** in ulcerative colitis

   Approximately 65 % of ulcerative colitis patients present initially with limited left-sided involvement, which may spread to involve more proximal portions of the colon in 29–58 % of cases (D'Haens et al. 1997; Ekbom et al. 1991). D'Haens et al. reported that in "**left-sided**" **ulcerative colitis,** distal involvement may be accompanied by more proximal areas of inflammation, particularly in the periappendiceal area of the cecum. Ileocolonoscopy with biopsy was performed prospectively in 20 patients with left-sided ulcerative colitis. Segmental inflammation, separated from the distal inflamed segment by apparently uninvolved mucosa, was found in 15 patients (75 %) and always included the area around the appendiceal orifice.

3. Initial presentation of pediatric patients with ulcerative colitis

   The absence of features of chronicity, mild active disease, and microscopic skip areas, at initial presentation in pediatric patients, does not exclude the possibility of ulcerative colitis. Glickman et al. presented at the 2002 USCAP meeting comparing the rectal mucosal biopsy appearance of 70 pediatric ulcerative colitis patients to 44 adult patients, all at initial presentation. The pediatric group showed significantly fewer chronic active diseases, and more microscopic skip areas and relative **rectal sparing** in comparison to adults. Furthermore, two of pediatric patients showed completely normal rectal biopsies (Glickman and Bousvaros 2002).

4. **Backwash ileitis**

   A mild degree of active inflammation in the distal few centimeters of terminal ileum is commonly recognized among patients with severe pancolitis. This is presumably related to reflux of colonic contents (Mc et al. 1949; Saltzstein and Rosenberg 1963). Although backwash ileitis has not been shown to be a significant risk factor for the development of pouchitis, rarely, premalignant dysplastic changes, and even adenocarcinoma, have been shown to develop in this setting (Schlippert et al. 1979; Gustavsson et al. 1987).

5. Upper gastrointestinal involvement in ulcerative colitis

   In 2000, Valdez et al. reported four cases of diffuse duodenitis associated with well-documented ulcerative colitis that multiple endoscopic biopsies show histologic features of ulcerative colitis and colectomy specimens confirming severe ulcerative **pancolitis**. Endorectal pull-through (ERPT) procedures were performed in three of four patients, and an end-to-end ileorectal anastomosis was done in one patient. None of the patients developed postsurgical Crohn's-like complications during a follow-up period of 12–54 months (Valdez et al. 2000). More precise characterization of cases with long-term follow-up is needed to help establish specific criteria for upper gastrointestinal tract involvement in patients with ulcerative colitis.

6. Unusual patterns of disease in Crohn's disease

   Crohn's disease may involve the colon similar to ulcerative colitis consisting of continuous mucosal inflammation of the colorectum, minimal or no submucosal inflammation, and an absence of upper gastrointestinal tract involvement (Guy et al. 2001; Kleer and Appelman 2001). In these cases, the diagnosis relies heavily on the finding of granulomas or transmural lymphoid aggregates in the resection specimen. These cases may account for some of the cases of presumed ulcerative colitis which have Crohn's disease-like complications (Goldstein et al. 1997).

The initial diagnostic work-up of a patient with symptoms suspicious for inflammatory bowel disease requires the optimal integration of clinical, radiology, laboratory, endoscopic, and histological data to avoid misdiagnoses and therapeutic **pitfalls** (Magro et al. 2013; Dignass et al. 2010, 2012b; Annese et al. 2013; Lichtenstein et al. 2009).

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Diagnostic Algorithm for Constipation and Obstructed Defecation 24

Guido Manfredi, Claudio Londoni, Massimo Bellini, and Elisabetta Buscarini

Contents

Abstract

Chronic constipation is a common and disabling problem in many patients all over the world, in particular in elderly. There are two main pathophysiologies, but with possible overlapping situations: disorders of transit and evacuation disorders.

Functional constipation has many causes, including the kind of diet and lifestyle, and it can also be secondary to medications, other many medical conditions, and/or disease. Alarm symptoms sometimes coexist, and it is mandatory to underline these conditions in order to manage the therapeutical approach properly.

Treatment options for chronic constipation include changes in lifestyle, drugs, and rehabilitation of the perineum as well as biofeedback therapies; commonly first-level therapeutical approach is undertaken before the diagnosis of chronic constipation will be cleared, but understanding its etiology is necessary to determine the most appropriate and tailored therapeutic option; history and physical examination of the patients can orientate in an intricate instrumental diagnostic approach which consists of imaging and functional tests.

Our aim is try to clarify on these complicated diagnostic choices in order to optimize therapeutical interventions.

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1 Chronic Constipation and Obstructed Defecation: Definition, Diagnosis and Clinical Approach

Chronic constipation is a worldwide problem increasing with age. It can be either primary or secondary. It is often, erroneously, considered as a single disease but it is a complex and multifaceted syndrome. There are many different causes able to induce secondary constipation (Tables 24.1 and 24.2).

The term "primary constipation" itself hides different conditions, such as irritable bowel syndrome with constipation (IBS-C), functional constipation, functional defecation disorders, and rectal hyposensitivity (Bellini et al. 2015; Bharucha et al. 2006; Longstreth et al. 2006) (Tables 24.3 and 24.4).

Particularly IBS-C is characterized by abdominal pain or discomfort improved by defecation, whereas functional constipation is a functional bowel disorder that presents as persistently difficult, infrequent, or incomplete defecation. Functional defecation disorders are characterized by paradoxical contraction or inadequate relaxation of the pelvic floor muscles during attempted defecation (dyssynergic defecation) or inadequate propulsive forces during attempted defecation (inadequate defecatory propulsion) (Bharucha et al. 2006).

Rectal hyposensitivity is a relatively new disorder defined by Gladman (Gladman et al. 2003) as an elevation beyond the normal range in the perception of at least one of the sensory threshold volumes during anorectal manometry. There are as yet no specific criteria that can differentiate the subtypes of chronic constipation based on history (Bharucha et al. 2006). Also performing a full assessment of defecation using specific tests (e.g., anorectal manometry, colonic transit time, and defecography) may not distinguish these different conditions (Wong et al. 2010; Rey et al. 2014; Jones et al. 2007; Gambaccini et al. 2013). However, a careful attempt to understand the pathophysiological mechanisms underlying the constipation of each patient is mandatory in order to suggest an effective therapy. This should be strictly tailored to each individual patient and therefore different from one patient to another (Bellini et al. 2015).

Even if there are no specific criteria that can definitely distinguish among the different subtypes of chronic constipation, a careful medical history should always be collected. It is the first approach to the patient and is aimed to detect symptoms and events possibly linked to the onset of symptoms themselves (Bove et al. 2012).

Table 24.1 Medications more commonly related to constipation

Antidiarrheal agents	Antiparkinson drugs
Antiepileptics	Antispasmodics
Antihistamines	Calcium channel blockers
Antidepressants	Diuretics
Antipsychotics	Sympathomimetics
Antiacids with aluminum or calcium	NSAIDs

Table 24.2 Condition associated to constipation (Diamant et al. 1999)

Mechanical obstruction	Colorectal tumor, diverticulosis, strictures, ab-extrinseco compression, postsurgical abnormalities, volvulus
Pelvic – anal diseases	Levator ani syndrome, rectal prolapse, rectocele, anal abscess, anal fissure, hemorrhoids
Myopathies, neuropathies	Autonomic neuropathy, cerebrovascular disease, spinal cord disease, multiple sclerosis, Parkinson disease, systemic sclerosis
Metabolic conditions	Chronic renal failure, diabetes mellitus, electrolyte disorders, dysthyroidism, Porphyria
Psychiatric conditions	Cognitive impairment, anorexia, depression
Other conditions	Fluid depletion, low-fiber intake, immobilization, dehydration, cardiac disease

Table 24.3 Rome III criteria for differential diagnosis between functional constipation and irritable bowel syndrome (Bharucha et al. 2006)

Functional constipation *Diagnostic criteria* *
1. Must include two or more of the following:
Straining during at least 25 % of defecations a. Lumpy or hard stools in at least 25 % of defecations b. Sensation of incomplete evacuation for at least 25 % of defecations c. Sensation of anorectal obstruction/blockage for at least 25 % of defecations d. Manual maneuvers to facilitate at least 25 % of defecations (e.g., digital evacuation, support of the pelvic floor) e. Fewer than three defecations per week
2. Loose stools are rarely present without the use of laxatives
3. Insufficient criteria for irritable bowel syndrome
* Criteria fulfilled for the last 3 months with symptom onset at least 6 months prior to diagnosis
Irritable bowel syndrome with constipation
Diagnostic criterion *
Recurrent abdominal pain or discomfort ** at least 3 days/month in the last 3 months associated with two or more of the following:
1. Improvement with defecation 2. Onset associated with a change in frequency of stool 3. Onset associated with a change in form (appearance) of stool (hard or lumpy stools ≥*25 % and* loose or watery stools <*25 % of bowel movements*)
Criteria fulfilled for the last 3 months with symptom onset at least 6 months prior to diagnosis ** "Discomfort" means an uncomfortable sensation not described as pain

Table 24.4 Roma III diagnostic criteria for functional defecation disorders

Criteria fulfilled for the last 3 months with symptom onset at least 6 months prior to diagnosis	
The patient must satisfy diagnostic criteria for functional constipation During repeated attempts to defecate must have at least two of the following:	
	Evidence of impaired evacuation, based on balloon expulsion test or imaging
	Inappropriate contraction of the pelvic floor muscles or less than 20 % Relaxation of basal resting sphincter pressure by manometry, imaging, or EMG
	Inadequate propulsive forces assessed by manometry or imaging

History can also identify alarm symptoms (Table 24.5), such as weight loss, bloody stools, anemia, or a family history of colon cancer and conditions and/or diseases potentially associated with constipation, such as dietary mistakes (Altringer et al. 1995); low physical activity (Diamant et al. 1999); the use of constipating drugs; metabolic, psychiatric, or neurological diseases; and previous perineal-pelvic-abdominal or obstetric-gynecological surgery (Tables 24.1 and 24.2). In case of alarm symptoms/signs, colonoscopy is recommended (Table 24.10).

Also assessing the stool form using the Bristol stool form score (Lewis and Heaton 1997) is of paramount importance to obtain an objective evaluation; moreover stool consistency is considered an indicator of colonic transit; hence, it can address the diagnosis.

A physical examination is essential in the initial workup of a patient with chronic constipation (Lindberg et al. 2011). The examination can detect a possible gastrointestinal mass and should include inspection of the anorectal region and exploration of the rectum. This process can provide evidence of external signs of anal disease, pelvic organ prolapse, or descending perineum syndrome. A digital rectal examination should detect any signs of organic disease or obstructed defecation (rectal masses, fecal impaction, stricture, rectal intussusception, or rectocele). The examination is particularly important if functional alterations in defecation are suspected in order to evaluate puborectal and anal sphincter activity.

Blood tests do not provide useful input in functional constipation but can be performed to exclude conditions of secondary chronic constipation (Bove et al. 2012) (Table 24.2). They also can be mandatory when alarm symptoms are present.

Once excluded on a clinical basis organic lesions and secondary constipation, many patients will benefit from abolishing or reducing medications that cause constipation and recommending changes in lifestyle and diet with correct fluid (at least 1.5 l/day) and fiber (25 mg/day) intake (Table 24.6).

Table 24.5 Clinical approach to constipation

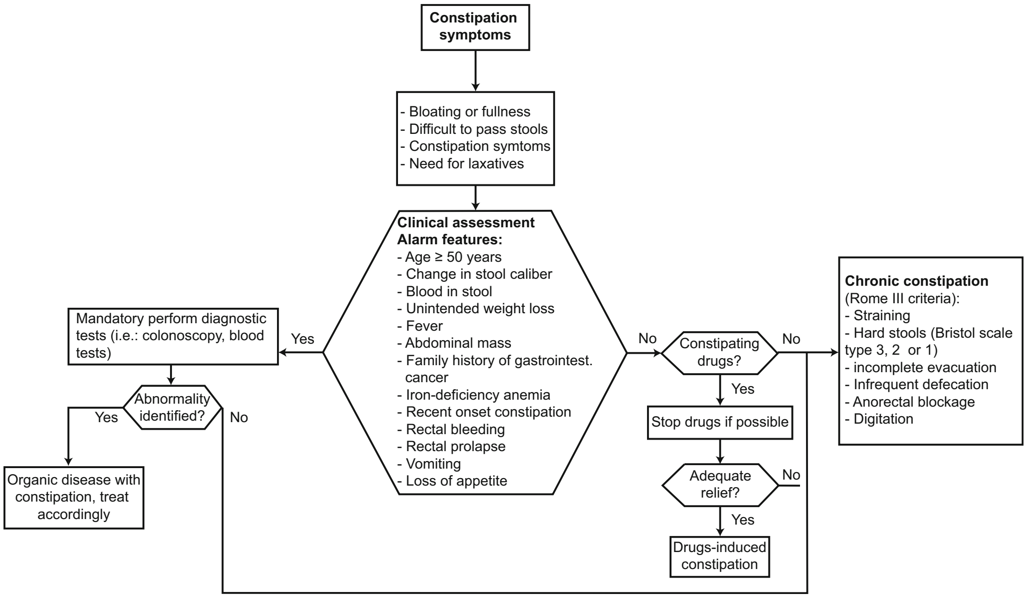


Table 24.6 Stepwise approach to chronic constipation

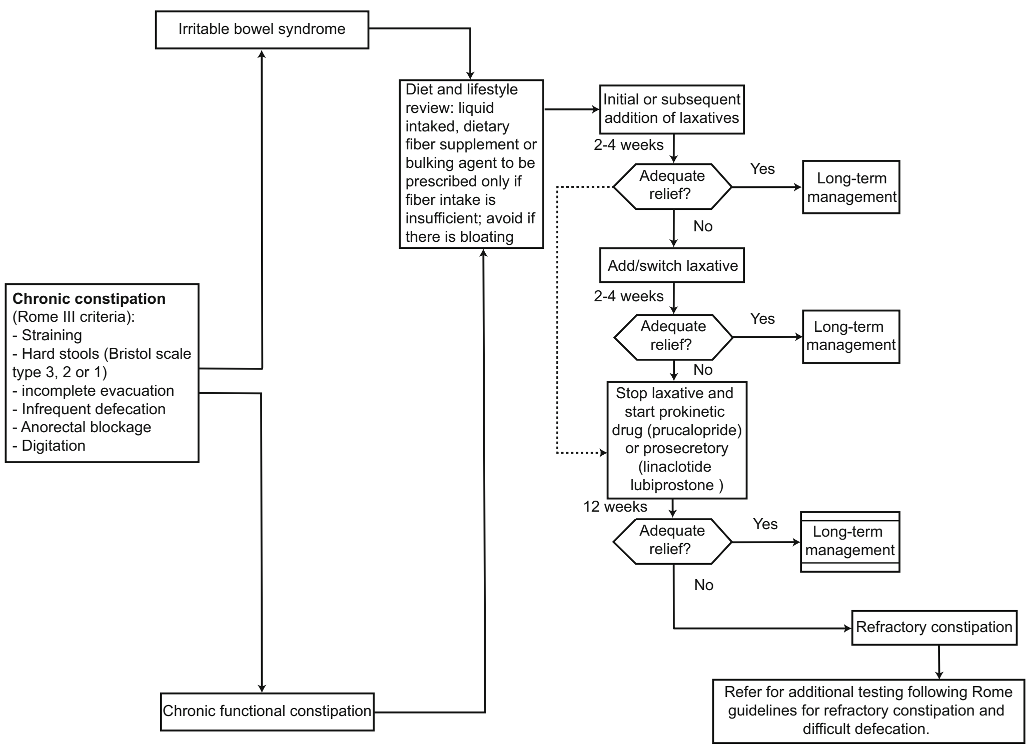


Table 24.7 Stepwise approach to constipation) refractory to high-fiber diet, laxatives, and prokinetic/prosecretory drugs

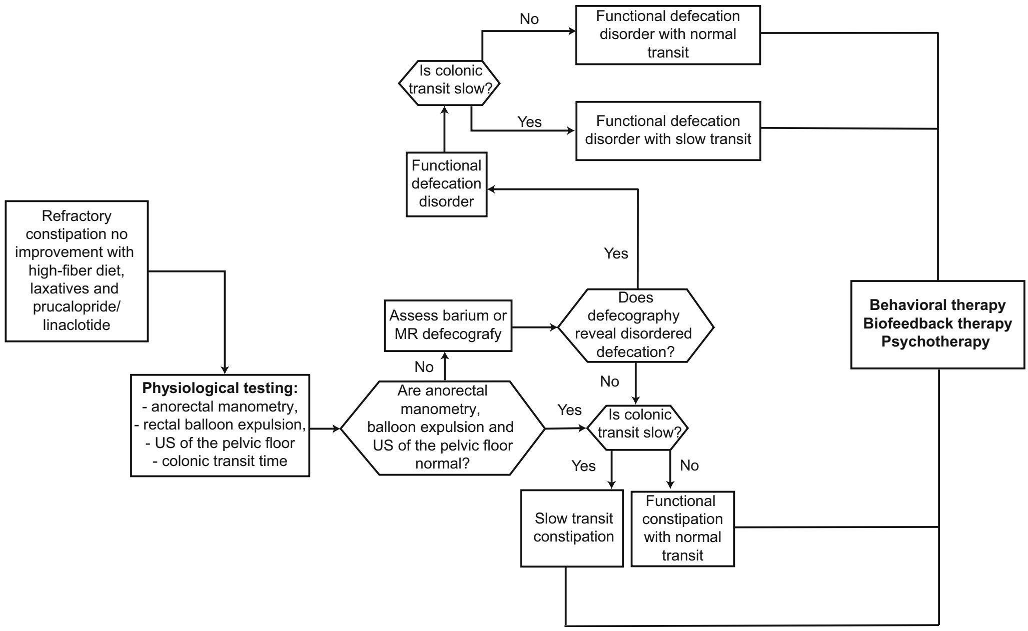


Table 24.8 Diagnostic tools and relevant information provided (Pehl et al. 2002)

Diagnostic tool	Provided information
Rx colonic transit time, entero-defecography, RM defecography, DTP-US	Morphological and dynamic
Anorectal manometry	Anorectal sensitivity and motility
Balloon expulsion test	Dynamic abdominoperineal synergy

If this management is not sufficient, it is mandatory to move to a second step encompassing the use of fiber supplementations and osmotic laxatives.

If also these therapies are ineffective, it is possible to use old (stimulant, softening, or saline) laxatives or new prokinetics or prosecretory agents even if in this subset of patients, further tests such as anorectal manometry and/or entero-defecography and/or colonic transit time are advisable, in order to better characterize the type of constipation (Tables 24.7, 24.8, and 24.9) and to evaluate other therapeutic options (e.g., pelvic floor rehabilitation, sacral nerve stimulation, anorectal surgery) (Ratto et al. 2015); colpo-cysto-entero-defecography (Altringer et al. 1995; Maglinte et al. 1997), magnetic resonance (MR) defecography (Lienemann et al. 1997), and dynamic transperineal ultrasonography (DTP-US) (Beer-Gabel et al. 2002, 2004; Dietz and Steensma 2005; Brusciano et al. 2007) are also available and increasingly utilized. Colonic and/or gastrojejunal manometry should be performed in patients with serious slow-transit constipation because they can be helpful in the diagnosis and in decisions regarding therapy (whether conservative or surgical) (Bove et al. 2012).

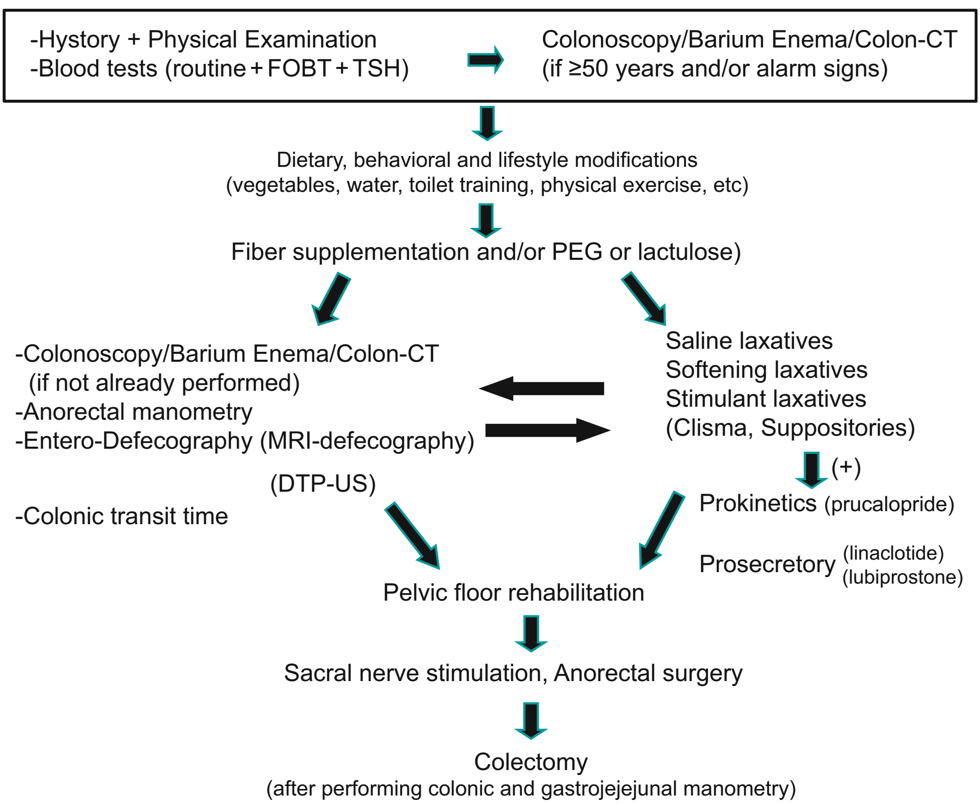
The global approach to chronic constipation integrating available tests and treatments is summarized in Table 24.10.

Table 24.9 Interpretation of the manometric data (Bove et al. 2012)

Test	Parameter evaluated	Interpretation
Resting pressure	IAS (70 % of resting pressure) and EAS (30 % of resting pressure)	P increased: Hypertonic sphincters (IAS and/or EAS) Oral nitroglycerin can identify the sphincter involved because it relaxes IAS, but not EAS
Squeeze pressure	EAS	The fatigue rate index can be calculated based on the pressure and duration of the contraction. However, the usefulness of the test in both constipated and incontinent patients is disputed
Rectoanal inhibitory reflex	IAS relaxation during rectal inflation	Absent: Possible Hirschsprung If present with elevated volume inflation: Megarectum
Rectal sensitivity	Rectal sensory function at different volumes	Elevated sensory thresholds may be linked to changes in rectal biomechanics (megarectum) or to afferent pathway dysfunction
Rectal compliance	Mechanical rectal function	Increased compliance: Megarectum
Attempted defecation	Synchronization between the increase in rectal pressure and the decrease in anal pressure during attempts to defecate	Three types of dysfunction may be detected: Type 1: Adequate rectal P increase but associated with anal P increase Type 2: Inadequate rectal P increase associated with anal P increase or inadequate anal P decrease Type 3: Adequate rectal P increase but inadequate anal P decrease

IAS: Internal anal sphincter; EAS: External anal sphincter; P: Pressure

Table 24.10 Global stepwise approach to chronic constipation



Legend: FOBT: Fecal occult blood test

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Index

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